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ELLEN P. TAPPERO
MARY ELLEN HONEYFIELD

Physical Assessment of the Newborn

Sixth Edition

A COMPREHENSIVE APPROACH TO THE ART OF PHYSICAL EXAMINATION

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Foreword

Systematic, accurate, ongoing physical assessment is a critical component of managing neonates across all settings. The purposes of neonatal assessment include identifying influences of the prenatal environment, evaluating transition to extrauterine life, recognizing early the subtle indicators or changes that may be harbingers of serious problems, and evaluating a plethora of clinical findings to distinguish between normal variations and problems. Assessment is an integral part of care planning and parent teaching.

There is a tremendous range of variation even among normal newborns. These variations include normal differences characteristic of infants in general and traits that are specific and unique within an individual family. Family traits, due to genetic variation, may or may not have pathologic significance. Variations seen in newborns may also be abnormal. Minor or subtle anomalies may be missed unless the person assessing the infant has heightened awareness and skill in assessment and is specifically looking for anomalies during the newborn examination. These subtle alterations are important in that they can provide clues to the presence of internal malformations or syndromes that have significant consequences for the infant and family. In addition, many problems in the neonate, such as infection, metabolic alterations, necrotizing enterocolitis, or changes in skin integrity, can be detected early, long before electronic monitors or other equipment record them, by an astute nurse, nurse practitioner, or physician with good assessment skills. Identification of infant state and behavioral cues and signs of pain is critical in recognizing and reducing stress and in providing individualized care.

This landmark 25th anniversary edition of this book continues to be a valuable and essential resource for all those involved in neonatal care. Editors Ellen P. Tappero, DNP, RN, NNP-BC, and Mary Ellen Honeyfield, DNP, RN, NNP-BC, have developed a comprehensive text with a wealth of detailed information on the assessment of the newborn. This book is an excellent resource for beginning and experienced practitioners on gestational assessment, neurologic assessment, neonatal history, and assessment of the dysmorphic infant, as well as the systematic evaluation of individual body systems. Chapters also discuss behavioral and pain assessment, including the use of specific tools with various groups of infants ranging from term to extremely preterm infants. Implications of antepartum and intrapartum testing for the newborn are also addressed. All of the content has been updated and, in some areas, expanded to continue to provide state-of-the-art knowledge. New to this sixth edition of the book is a chapter focusing on assessment of the drug-exposed neonate. Because substance exposure is a growing problem in perinatal and neonatal care, this is a timely addition and valuable resource.

The numerous tables, figures, and illustrations, including many color illustrations and photographs, are a major strength that enhances this book's usefulness as a clinical resource. A glossary provides a quick resource for looking up definitions of findings and techniques. The chapter authors clearly identify areas for

assessment, provide the scientific basis for and rationale underlying its various assessment techniques, review standard terminology, and define and exemplify normal and abnormal findings and common variations. The book not only illustrates the principles and skills needed to gather assessment data systematically and accurately, but also provides a knowledge base for interpretation of these data.

The text is both an excellent teaching tool and a resource for anyone who performs newborn examinations, including nurses, neonatal and pediatric nurse practitioners, nurse-midwives, physicians, and therapists. It should be a core text for any program preparing individuals for advanced practice roles in perinatal and neonatal care, and it should be a resource in every setting providing care to neonates. Individual practitioners have varying degrees of familiarity and comfort with the many areas of newborn assessment. This text can serve as an in-depth, systematic introduction to the major components of and techniques for evaluating all the major systems. For more experienced practitioners, it can reinforce, update, and improve knowledge and techniques. At all levels of practice, this book serves as a convenient reference to normal parameters, common variations, and less commonly seen abnormalities.

Understanding the unique physical, physiologic, neurologic, and behavioral findings in the neonate helps practitioners to recognize alterations and prevent or minimize their effects. Skillful newborn assessment reduces the risks associated with the transition to extrauterine life and pathophysiologic problems of the neonatal period. Yet because assessment skills are such an integral aspect of practice, many individuals take them for granted. To maintain excellence, practitioners must continue to expand, update, and validate their skills. This book provides a resource to do so and has been a significant contribution to and essential resource for neonatal care for 25 years. This edition continues that legacy, for which the editors and authors are to be congratulated.

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Preface

Physical Assessment of the Newborn has been a leading resource for newborn healthcare providers since it was first published 25 years ago. Support from academicians and clinicians places a great deal of responsibility on editors and chapter authors to earn continued acceptance and future endorsement of each new edition.

To accomplish this goal, we continue to preserve aspects of the book that have been regarded by readers as essential, including its easy-to-read style, logical and user-friendly organization with numerous tables, graphs, and photographs. The overall organization is identical to that of previous editions. In addition to extensive updates and revisions of previous existing chapters, cultural changes have compelled us to add a new chapter to address the assessment of the antenatally substance-exposed newborn.

This is a unique book, relevant to a wide audience. Our original goal remains the same: to make the material understandable to a diverse group of individuals ranging from practicing staff nurses, nurse practitioners, and nurse-midwives to physician assistants, medical students, and pediatric residents. Our objective is based on the premise that newborn healthcare providers must be confident in their knowledge and demonstrate clinical competence with "well" newborns, whether preterm, late preterm, early term, or term gestations before they move on to the study of ill newborns. Diagnosis by physical assessment is considered by some to be "old" medicine and lacks the glamour of high-tech diagnostic modalities. Although it may seem "old fashioned" because it relies on clinical experience and the ability to discriminate among normal, normal variations, and abnormal findings, physical assessment remains a vital component of our practice. There should be a balance between the new and old with technology supplementing, rather than replacing, the skill of physical assessment.

The information contained in the text is only an introduction and is not meant to be an exhaustive review. Along with other references, this book is intended to be useful as a resource for physical assessment of the newborn to form the foundation that supports the clinical decision-making process. We hope that this edition continues to arouse the interest of beginning students in newborn physical assessment and allows experienced practitioners to further refine their assessment skills.

Each author, many of whom were chapter authors in the first edition, has written material distinctly and no attempt has been made to achieve a completely uniform format. We are grateful to our authors, who revised or wrote their chapters willingly and enthusiastically, in spite of the myriad of other responsibilities in their lives.

This book is possible because of the efforts of many individuals. Thanks go to our teachers, who introduced us, as novices, in a positive way to the challenge of newborn physical assessment. We offer thanks to our many colleagues who have shared their photographs and slides as well as parents who allowed photographs to be taken of their infants.

Finally, no book is ever a reality without the perseverance of the editorial staff. We are grateful to the Springer Publishing editorial staff who assisted in the preparation, editing, and design of this 25th Anniversary edition.

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Ellen Tappero
Mary Ellen Honeyfield

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Acknowledgments

It is both exciting and challenging to have the opportunity to prepare yet another edition of *Physical Assessment of the Newborn.* I would like to express my gratitude to the many people whose contributions have made this work a valuable and sustainable text for students and practicing clinicians.

My thanks to all of the authors and colleagues who provided support, shared knowledge, read, wrote, and offered comments and suggestions for each new edition. Thanks to my coeditor, Mary Ellen Honeyfield, for the stimulation to begin the project initially and mutual respect generated by our continued collaboration over the past 25 years. For valuable help in the preparation of the manuscript, I would like to acknowledge the Springer Publishing editorial staff, who showed confidence in our work and assisted in editing, proofreading, and designing this 25th-anniversary edition.

I am especially indebted to my husband, who has been supportive of my career goals and worked to provide me with the protected time to pursue those goals. Without his encouragement, patience, and understanding, this text would still be an item on my professional "bucket list."

Ellen P. Tappero

As someone who enjoys celebrating milestones, this 25th anniversary edition of *Physical Assessment of the Newborn* brings sentimental emotions. From the first musing, "There needs to be a book on newborn physical assessment," to this 6th edition has been a personal and professional journey of growth and gratitude. I have come to recognize the many "families" we gain throughout our careers, and this edition is dedicated to those families.

Families become our community. My first family was The Children's Hospital in Denver where I acquired the skill of observation and lifelong learning from those who would now be considered some of the founders of neonatology. Mentors and peers became family, as did the many families who followed: P/SL, NCC, NANN, NNP Services of Colorado, NCMC; and also my Kansas City families at MMC, CMH, and PMC. There are no words big enough to thank everyone who contributed in their own ways to the knowledge I gained, ultimately making this text successful. And how can one thank the families of all the babies I was privileged to observe and care for at the most intimate times of their lives.

Every edition of this book had its own family, beginning with my extraordinary co-editor, expert authors, script editors, publishers, and international readers who have continued to support the importance of the contextual content, learning how to assess newborn babies.

Finally, professional endeavors are always aided by one's own personal family, whose support, and probably tolerance, allows success. Thanks to all for your continued backing of that very first thought about what text was needed.

Mary Ellen Honeyfield

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Physical Assessment of the Newborn: A Comprehensive Approach to the Art of Physical Examination, Sixth Edition

Principles of Physical Assessment

Mary Ellen Honeyfield, DNP, RN, NNP-BC

1

The importance of approaching newborn physical examination with a sense of anticipation cannot be overemphasized. This first examination offers a unique opportunity for early recognition of any problems the infant may have. Seeing each infant as a mystery to be unraveled requires mindful curiosity from the examining clinician. The newborn's inability to provide verbal information tests the acuity of the examiner's skills. When the examiner views this responsibility as a diagnostic challenge, newborn physical assessment provides both personal and professional satisfaction, even though most infants examined are normal.

Initially, the inexperienced examiner finds physical assessment time-consuming. Lack of familiarity with essential tools and limited practice with basic techniques both slow assessment. In fact, the student often views the infant as a series of systems to be examined. Repetition and experience help the examiner learn to see the newborn as a whole and process multiple observations while examining individual systems. For example, if a previously quiet infant begins to cry during palpation of the abdomen, the experienced examiner continues the abdominal examination but also notes the quality of the cry, the infant's color while crying, respiratory effort, facial movements, the tongue, intactness of the palate, and movement of the extremities.

Clinical expertise develops throughout a practitioner's career. With experience, each clinician develops her or his own unique approach to newborn physical assessment. The sequence of how the examination is performed is not as important as developing a consistent and organized approach. Consistency—performing the newborn physical assessment in the same organized manner every time—maximizes information gained from each examination performed, thus adding to the clinician's knowledge base and ensuring that portions of any individual examination will not be inadvertently forgotten.

TECHNIQUES OF PHYSICAL ASSESSMENT

Observation

Observation, or inspection, is the most important physical assessment technique for practitioners to master. Observation also is the most difficult skill for the fledgling examiner to incorporate into his or her clinical approach. Often the novice clinicians' first instinct is to touch, talk, and unwrap the infant, thus possibly startling the infant and missing an opportunity to observe the infant's presenting status.

In 1860, Florence Nightingale wrote these thoughts about the art and skill of observation:

> In the case of infants, everything must depend upon the accurate observation of the nurse or mother…. For it may safely be said, not that the habit of ready and correct observation will by itself make us useful…, but that without it we shall be useless

> with all our devotion…. If you cannot get the habit of observation one way or [the] other, you better give up…, for it is not your calling, however kind and anxious you may be.[1]

Using the visual and auditory senses, the practitioner observes the infant, assesses, and makes decisions about what has been seen or heard. A specific observation may alert the examiner to assess a particular system more thoroughly. For example, the observation of an active precordium (visual cardiac pulsations) directs the examiner to careful cardiovascular assessment. Auscultation may reveal a heart murmur. This finding may lead the examiner to palpate the precordium and peripheral pulses and obtain four extremity blood pressures, actions that are not normally part of the examination of an otherwise healthy-appearing newborn.

The practitioner can collect most of the information needed for a complete physical assessment solely through observation (Table 1.1).

Auscultation

Auscultation is the technique of listening to sounds produced by the body (e.g., the lungs, heart, and gastrointestinal tract). Direct auscultation involves application of the examiner's ear to the body surface being assessed. Some sounds, such as stridor, wheezing, and expiratory grunting, may be heard simply by being near the infant. Indirect (mediate) auscultation utilizes a stethoscope to listen to these same sounds.

Accurate indirect auscultation of the newborn requires a stethoscope fitted with a pediatric-sized, double-headed chest piece with an open bell and a closed diaphragm. The stethoscope should be placed firmly on bare skin rather than over the infant's clothes. A quiet infant and environment as well as a warm room and warm stethoscope facilitate auscultation.

Palpation

Palpation is a technique in which the examiner uses the sense of touch to assess both superficial and deeper body characteristics. During palpation, the tips, palmar, and lateral surfaces of the fingers of both hands are used to assess external structures (e.g., skin, hair texture), vibrations (e.g., peripheral pulses, precordial activity, point of maximal impulse of the heart against the chest wall), and internal structures (e.g., liver, spleen, kidneys). One hand can be used, or a bimanual technique may enhance the palpation of deeper organs, such as the kidneys (Figure 1.1).

For accurate palpation, the infant ideally should be quiet and relaxed at the beginning of the abdominal examination. Warm hands, progressing from superficial to deeper palpation, and elevating the infant's hips off the bed keep the abdominal muscles relaxed. The examiner should take care not to palpate too deeply for superficial abdominal organs, such as the liver and spleen, as the fingertips may instead palpate stool-filled intestine or abdominal wall muscle mass rather than the organ itself. In palpation of the extremities and genitalia, a grasping action of the fingers is used to evaluate such accessible features as skin texture, skin lesions, descent of testes into the scrotum, muscle mass, and muscle strength.

Percussion

Percussion refers to tapping or striking a part of the body to put underlying tissue into motion. This movement produces audible sounds and palpable vibrations, which are then assessed for quality and duration of their tone and notes.

Indirect (mediate) percussion is performed (for a right-handed person) by hyperextending the middle finger of the left hand and placing only the distal interphalangeal joint firmly on the part of the body to be percussed. Contact by any other part of the hand will alter the sounds produced. The right wrist is then hyperextended and the middle

TABLE 1.1: OBSERVATIONS FOR PHYSICAL ASSESSMENT

TO ASSESS	OBSERVE
Distress	Facial expression, respiratory effort, activity, tone
Color	Tongue, mucous membranes (centrally pink vs. cyanotic), nail beds, hands, feet (peripherally pink vs. cyanotic), skin (jaundice, pallor, ruddiness, mottling), perfusion, meconium staining
Nutrition status	Subcutaneous fat, breast nodule
Hydration status	Skin turgor, anterior fontanel
Gestational age	Skin (smooth vs. peeling), ear cartilage, areola and nipple formation, breast nodule, sole creases, descent of testes, rugae, labia
Neurologic status	Posture, tone, activity, response to stimuli, cry, state, state transition, reflexes
Respiratory/chest status	Respiratory rate and effort, retractions, nasal flaring, grunting, audible stridor or wheezing, chest shape, nipples (number and position), skin color
Cardiovascular status	Precordial activity, visible point of maximal intensity, skin perfusion and color
Abdomen	Size (full, distended, taut, shiny), shape (round, concave), distention (generalized or localized), visible peristaltic waves, visible bowel loops, muscular development/tone, umbilical cord, umbilical vessels, drainage from cord, periumbilical erythema (redness)
Head	Size, shape, fontanels, suture lines, swelling, hair distribution, condition of hair
Eyes	Shape, size, position, pupils, blink, extraocular movements, color of sclera, discharge, ability to fix and follow
Ears	Shape, position, external auditory canal, response to sound
Nose	Shape, nares, flaring, nasal bridge
Mouth	Shape, symmetry, movement, philtrum, tongue, palate, natal teeth, gums, jaw size
Neck	Shape, range of motion, webbing, masses
Genitalia (male)	Scrotum, descent of testes, rugae, inguinal canals, foreskin, penile size, urine stream, meatus, perineum, anus
Genitalia (female)	Labia majora, labia minora, clitoris, vagina, perineum, inguinal canals, anus
Skin	Color, texture, firmness, vernix caseosa, masses, lanugo, lesions (pigmentary, vascular, trauma-related, infectious)
Extremities	Posture, range of motion (involuntary movement), digits, palmar creases, soles of feet, nails

right finger partially flexed and "cocked" upward so as to strike the middle left finger. With a quick downward motion of the wrist, the tip of the right middle finger strikes the left middle finger's distal joint (Figure 1.2). Vibrations are transmitted through the bones of this joint to the underlying tissue being percussed. Direct percussion is performed

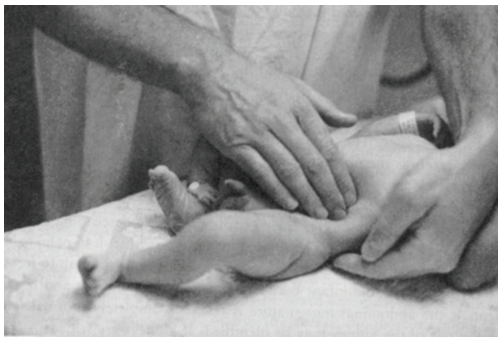

Figure 1.1 Bimanual inspection of the kidneys

Source: From Coen RW, and Koffler H. 1987. *Primary Care of the Newborn.* Boston, MA: Little, Brown, 30. Reprinted by permission.[2]

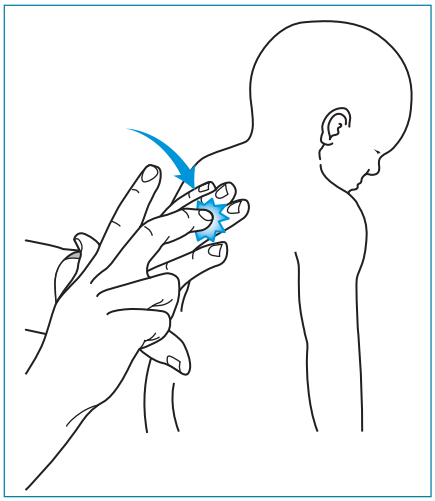

Figure 1.2 Indirect (mediate) percussion technique to assess underlying structures

Source: Adapted from Jarvis C. 2012. *Physial Examination & Health Assessment*, 6th ed. Philadelphia: Saunders, 117. Reprinted by permission.[3]

by directly striking the body surface to be assessed with the tip of the middle right finger.[4]

Translation of sounds heard during percussion into descriptive words is difficult at best. Practice and multiple examinations of normal infants and those with pathology is required to develop skill in describing percussed sounds.

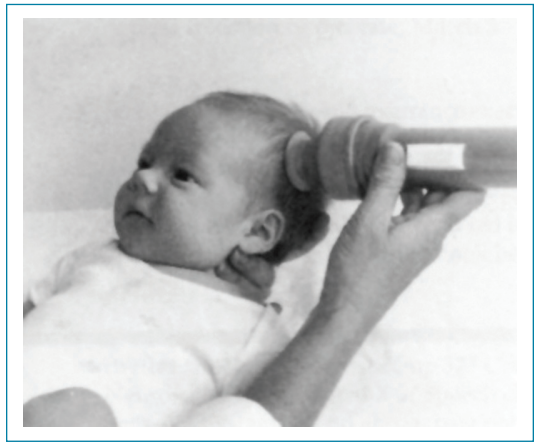

Figure 1.3 Transillumination

Source: From Jarvis C. 1992. *Physical Examination and Health Assessment.* Philadelphia, PA: Saunders, 297. Reprinted by permission.[5]

Although the technique of percussion is commonly taught to students, in practice it is not a technique universally used on the newborn. When the examiner, using mediate percussion, suspects a pathology, such as pleural effusion or air leak, the common approach in the newborn is to confirm the suspected diagnosis by x-ray. A description of comparative sounds produced during percussion has therefore been omitted here.

Transillumination

Transillumination is the technique of applying a high-intensity light directly to a body part, such as the head, chest, or scrotum, and assessing the amount of pink light that can be seen as a corona (halo) around a cuffed flashlight or fiberoptic device (Figure 1.3). Transillumination of the enlarged scrotum of an infant with a hydrocele reveals a fluid-filled mass that transmits light (Figure 1.4) rather than a solid mass that does not transmit light, as in the case of an inguinal hernia. As with the other physical assessment techniques described, practice enables the examiner to recognize the difference between normal and abnormal halos of light.[4]

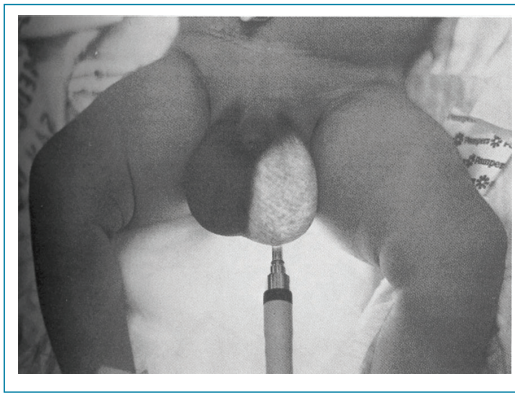

Figure 1.4 Transillumination of hydrocele

Source: From Coen RW, and Koffler H. 1987. *Primary Care of the Newborn.* Boston, MA: Little, Brown, 33. Reprinted by permission.[2]

TIMING OF THE EXAMINATION

Timing of newborn physical assessment often depends on circumstance and hospital guidelines. A quick overall exam can be done in the delivery room unless the infant is clinically stable and has been placed with the mother. While disconcerting when parents are first to discover a problem, the clinician is guided by hospital practice. A complete physical examination is done during the first few hours after birth when the infant becomes accessible.[6] Daily and discharge examinations are completed thereafter.

BASICS OF PHYSICAL ASSESSMENT

Certain themes recur throughout this text in discussions of assessments of each system. Repetition is intentional and reflects the importance of these activities. These basic principles of physical assessment include the following:

- **Review the perinatal history for clues to potential pathology.** The newborn's history begins with conception and includes events that occurred throughout gestation, labor, and delivery. The newborn is also affected by genetic histories of both parents and of their families. For example, a maternal history that includes diabetes mellitus directs the experienced practitioner to carefully assess the cardiovascular and neurologic systems and the extremities, because infants of diabetic mothers show an increased incidence of abnormalities in these systems. Labor and delivery history may reveal the mother received medication for pain relief just before delivery; this could account for depression of the newborn's respirations and/or muscular tone. With this knowledge, the examiner need not pursue a more serious etiology for the depressed respirations, as long as the respiratory pattern or muscle tone improves over time.
- **Assess the infant's color for clues to potential pathology.** The infant's color provides important information about several body systems. For example, very red, or ruddy, infants may have polycythemia and be more prone to complications such as respiratory distress or hypoglycemia associated with this phenomenon. The infant whose tongue and mucous membranes are pale or blue (central cyanosis) may be anemic or may have a heart lesion or respiratory disease. Proper lighting is essential for accurate assessment of color. If the examiner is concerned about central color, pulse oximetry can aid assessment.
- **Auscultate only in a quiet environment.** Assessing sounds produced by the body is difficult if there are noises, such as people talking or a radio playing, in the room. External interferences impede accurate evaluation of heart and breath sounds.
- **Keep the infant warm during examination.** After the infant is undressed, preventing heat loss is crucial to the infant's comfort and maintaining a normal temperature and glucose homeostasis. The undressed infant is examined in a warm environment with an external heat source, such as an overhead radiant warmer. To keep from startling the newborn and maintaining a stable metabolic status, warming the stethoscope as well as the examiner's hands aids accurate assessment.

- **Have the necessary tools at hand.** A stethoscope, an ophthalmoscope, and a tape measure are used in all newborn examinations. Having these tools ready saves time.
- **Calm the infant before beginning the examination.** A quiet infant provides the best opportunity for data gathering. If a crying infant must be examined, patience—and possibly the aid of a second person to help calm the infant—is required.
- **Handle the infant gently.** The newly born infant is amazingly cooperative when the examiner is gentle. A soothing voice and a soft touch often allow the examiner to complete the entire physical assessment without disturbing the infant greatly or at all. Parents enjoy watching their infant interact with the examiner and appreciate a gentleness of touch. Certain portions of the examination cause the infant more distress than others. Examination of the hips, usually the most disturbing part of the examination, is therefore performed last.
- **Complete the examination.** Redress the infant to maintain a normal temperature. Notify the primary caregiver the examination is over and of any abnormalities that were found or further observations that need to be made.

A SAMPLE APPROACH

One method of organizing examination of the newborn is described here as a guide for the inexperienced examiner. With practice, each examiner develops a personal style. It is assumed that the infant is unclothed and supine under a radiant warmer.

Observation

It can be very difficult for a practitioner to just stand at a crib and observe the infant. The immediate inclination is to touch and talk to the infant. This natural response must be repressed until later in the examination, however, because observation alone produces important information about every organ system. These initial observations allow the examiner to develop a visual differential diagnosis before employing other assessment techniques (auscultation, palpation). Table 1.1 catalogs the many observations the skilled examiner must process in this initial assessment of the infant's status.

If these multiple observations prove normal, the examiner is less likely to find a significant abnormality upon auscultation and palpation. Each observation of normality serves to reassure the examiner—just as an observation of abnormality should heighten the examiner's suspicion that further inspection is necessary.

Observation is not an isolated technique for use only at the outset of the examination. Although it is important to spend a moment or two at the bedside before touching the infant, observation of the infant's responses takes place throughout the examination. Depending on the infant's state and responses, all the observations listed in Table 1.1 can be made during the hands-on examination. The examiner must learn to take advantage of every opportunity the infant's behavior offers for observation. If, for example, the infant awakens spontaneously during the examination, the examiner should take advantage of that opportunity to examine the eyes.

Auscultation

After observing the infant closely, many examiners next auscultate the lungs, heart, and abdomen. Concentration allows the examiner to separate the sounds of the heart from those of the lungs. Listen first to one type of sound, then to the other. For example, listen first to the heart—its rate, rhythm, regularity, and any added sounds. Then listen to breath sounds, ignoring cardiac sounds.

Palpation

The examination continues with palpation. Palpating certain parts of the body disturbs the infant more than palpating others. An

ordered approach keeps the infant calm through much of the process. Because femoral pulses are difficult to assess in a crying infant, palpate them first. Then palpate the brachial pulses. Next palpate the abdomen, beginning with the more superficial liver and spleen. (Learning to palpate the liver and spleen with the tips of the fingers as well as with the lateral edges of the index fingers facilitates examination from either side of the bassinet.) Palpate for abdominal masses; then use deeper palpation for the kidneys. At this point, the infant may be disturbed and crying, but this will not impede the remainder of the examination.

Head-to-Toe Examination

After palpation, the examiner may then assess the infant from head to toe, examining the head and neck, upper extremities, genitalia, lower extremities, and back, in sequence. This hands-on inspection also includes measurements and tactile examination of the skin. Maneuvers to assess symmetry and reflexes are also done.

The Integrated Examination

The skilled examiner integrates examination tasks. For example, after palpating the head, neck, clavicles, arms, and hands, the examiner can perform the pull-to-sit maneuver to assess palmar grasp, arm strength, and tone. At that point, the infant is being held in an appropriate position to elicit the Moro reflex. Genitalia can be examined next, before progressing to the lower extremities. While the infant is positioned prone on the practitioner's hand to assess truncal tone and the truncal incurvation reflex, the back can be examined. These shortcuts facilitate multiple inspections and save time. Hips should be examined last because this procedure causes the most stress to the infant.

Usually it is not necessary to assess reflexes as a separate step. The examiner will likely have observed root and suck by this point. Moro and palmar grasp can be incorporated into the upper extremity examination, as just explained.

Although an extremely cooperative infant may sleep through the entire examination, assessment is not complete until the infant has been observed through various behavioral states. Facial asymmetry, for example, cannot be seen until the infant cries.

Ideally, the parents should observe the first complete examination. Parents appreciate demonstration of their infant's normality and uniqueness as well as early identification of unusual or abnormal findings.

EQUIPMENT

Ophthalmoscope

In addition to developing hands-on confidence with the newborn and performing the examination in the parents' presence, the student must become proficient with what may be unfamiliar equipment. Facility with the tools reduces stress to both infant and examiner. Again, practice is key.

If the ophthalmoscope is not already assembled, attach the head of the device to the handle by pushing the head toward the handle and turning the head clockwise. To turn the ophthalmoscope on, depress the on/off button, usually on the neck of the handle, at the same time turning the button clockwise. To gain familiarity with the various apertures, rotate the aperture selection dial and project the light onto the palm of the hand or a piece of paper. The lens selector compensates for the visual acuity of the individual examiner. Adjust the lens selector dial to bring the structure being examined into focus.[4,7] The small and the large round beams are the two apertures used to assess the gemini choroidal light reflex (referred to as the *red reflex*) and pupillary response in the undilated eyes of the newborn. The most opportune time to examine a newborn's eyes is when the infant is in a quiet alert state. An infant who is in light sleep will often open the eyes if the room is darkened. To examine the infant's eyes, hold the ophthalmoscope

in the right hand, with the viewing aperture as close as possible to the right eye, and use the right index finger to turn the lens selector dial to the appropriate lens for proper focus. While positioning the infant's head with the free hand, align the illuminating light along the infant's visual plane. If both the infant's eyes are open, the Bruckner test, a simultaneous bilateral red reflex test, may be performed. When using the large, full spot and holding the ophthalmoscope 18 to 20 in. (45–50 cm) from the infant's face, the light circle from the spot will cover both eyes. The examiner assesses symmetry and brightness of the red reflex as well as pupillary constriction.[7] The red reflex appears as a homogenous bright red-orange color. Any opacity along the central optical pathway will block all or part of the red reflex. Note that in infants with dark pigmentation, the reflex appears pale or cloudy rather than red.[7] Use the ophthalmoscope to observe pupillary constriction after assessing the red reflex.

Stethoscope

The acoustical stethoscope excludes environmental sounds, making it easier for the examiner to hear sounds coming from the infant; it does not magnify these sounds. The earpieces and connecting binaurals should be angled toward the wearer's nose to project sound onto the examiner's tympanic membrane. To minimize sound distortion, the tubing should be no longer than 12 in. The chest piece of the stethoscope often has a double head: a flat, closed diaphragm, and an open bell. The diaphragm is used to assess high-frequency sounds and the bell, low frequency. Use the diaphragm to assess breath sounds by applying it firmly to the infant's skin so it moves with the chest wall. Both the diaphragm and the bell are used to assess heart sounds.[8] Do not compress the bell against the chest so firmly that it fills with tissue and acts like a diaphragm (Chapters 6 and 7).

Otoscope

The infant's ears should be assessed for size, shape, and placement. The external auditory canals are examined for patency. Otoscopic examination is not normally done in the newborn period because the ear canals are often filled with vernix. For that reason, use of the otoscope is not described here.

SUMMARY

Each newborn examination is as different as each infant. This text explains basic techniques for examining each body system—to guide examiners with limited experience in performing a thorough newborn physical assessment. Only through experience, however, can practitioners develop clinical expertise. As experience and confidence grows, most examiners develop their own unique approach to physical examination, along with flexibility to adapt their approach to different situations. As long as the necessary equipment is available and basic principles are kept in mind, an infant can be examined in any setting.

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Recording and Evaluating the Neonatal History

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A comprehensive physical examination of the newborn, performed no later than 24 hours after birth by the physician, nurse practitioner, or physician's assistant, is a standard of perinatal care.[1] This applies to normal newborns; the high-risk newborn, however, requires an early and comprehensive history. Ideally, much of the history can be taken prior to the infant's impending birth. The physical examination must be preceded by a thorough review of the history of the current pregnancy; past obstetric history; intrapartum history; maternal medical, family medical, and social histories; and immediate neonatal adaptation history.[2] It is recommended that admitting nursery personnel evaluate the neonate's status and assess risks through a review of the history documented in the antepartum and intrapartum records no later than 2 hours after birth.[1]

Assessment is the gathering of accurate, detailed data, and it includes four components: (a) reviewing the history, (b) reviewing the results of the physical examination, (c) reviewing available laboratory or other data, and (d) formulating an impression (diagnostic differential). The practitioner then develops a plan of care and evaluates the newborn for actual or potential problems.

IMPORTANCE OF THE HISTORY

A comprehensive history is the prerequisite for adequate assessment. The newly born infant is a unique patient; no thorough history has ever been developed for this patient. When a newborn physical examination is performed without knowledge of the complete history, the information necessary for accurate formulation of an impression may be inadequate or incorrect.

The significance of the initial history and its thorough communication to the neonatal healthcare providers cannot be overstated. The newly born infant's history originates from the maternal records, but leads up to the individual neonate's transitional experience and plan of care. It is common for the neonatal healthcare provider to rely on a comprehensive maternal history; this history is often compiled from many sources and, perhaps, by many individuals. Communication with the obstetric healthcare provider, neonatal transport team members, and often the mother herself are essential to developing a thorough history. Written, faxed, or electronic information may be incomplete. The salient pertinent negatives and pertinent positives of a neonatal history are extremely important to developing your initial impression (diagnostic differentials).

Knowledge of the history can also be useful in allaying parental anxiety when performing the initial physical examination. A mother whose ultrasound at 20 weeks revealed the possibility of her fetus having a fluid-filled gastrointestinal (GI) mass, but whose subsequent ultrasounds appeared to be normal, is likely to have an increased level of concern about the status of her neonate's GI tract. Knowledge of this pertinent history allows the examiner to emphasize the normal GI findings, discuss the history of the positive ultrasound findings, and reassure the parents.

The history also alerts the examiner to potential or suspected problems and may indicate the need for more frequent repeat examinations. For example, a cardiac murmur in a neonate with a history of maternal anticonvulsant use throughout pregnancy is of more concern than a murmur in a neonate with a negative history. Embryonic exposure to maternal anticonvulsant therapy increases the risk for structural, congenital cardiac malformations.[3]

The history provides the context in which neonates with identified problems and disorders must be analyzed. Lack of a history or an incomplete history can lead to more extensive testing than would otherwise be necessary or to an incorrect impression or diagnosis. The following case exemplifies the need for obtaining an accurate history.

Jason, a 36-week gestational age, 3-kg neonate, was admitted to the nursery at 1 a.m. following vaginal delivery to a 34-year-old, gravida 4, para 3 mother with a history of preeclampsia. His Apgar scores were 7 and 8 at 1 and 5 minutes, respectively. His admission was uneventful; his physical examination revealed slightly decreased tone and spontaneous activity. Re-evaluation at 1 and 2 hours of life revealed continued poor tone and decreased spontaneous activity, with no other abnormal findings on the physical examination. At 2½ hours of life, an apneic episode was observed and noted. After being notified, the on-call healthcare provider requested that blood for a complete blood count (CBC) and C-reactive protein level be drawn. On the neonate's next examination, the findings were consistent with decreased tone and activity. Blood and spinal fluid were obtained for culture and sensitivity, and antibiotic therapy is started. The maternal chart with antepartum and intrapartum history was subsequently delivered to the nursery; it revealed that the mother had received magnesium sulfate. The neonate's magnesium level was obtained and found to be elevated. Review of the history would have given a more accurate context in which to view this infant's poor tone and activity and would have led to an earlier identification of hypermagnesemia, perhaps avoiding unnecessary testing.

The Supplemental Interview

Sometimes the mother, or both parents may need to be interviewed for information not found in the record. Before approaching the parents, review all available medical records and identify the specific information needed. Remember that the first contact with the mother or parents is one of the most important contacts. Introducing yourself and defining the reason for your questions will facilitate your interview. Be sure the parent(s) is comfortable, and maintain a normal conversational distance. Asking a friendly question, such as what name has been chosen for the baby, helps establish a positive atmosphere.

It is important to make and maintain eye contact and to present questions without reading them. Allow time for the mother to respond to your questions without interrupting her or checking your watch or the clock. Avoid technical language and the tendency to overexplain or lecture as much as possible. If you must take notes, make them brief. Copious note-taking detracts from your listening and observing abilities and may be intimidating.[4] Be an active listener: Show that you understand the responses, and request clarification if appropriate. Periodically, or at the end of the interview, summarize what you have been told. Evaluate answers to questions for content (what was said) and also for affect or tone (how it was said). A parent who avoids answering a question or answers incompletely may be embarrassed or upset by the question or perceives that the question is incriminating (such as in a drug-history interview).[5] When giving parents information about the neonate's health status, have them repeat at the end of the interview what they believe the neonate's health status or problems to be so you can clarify any misunderstandings immediately.

ELEMENTS OF A COMPLETE HISTORY

The historic information gathered must then be organized and presented in a systematic format commonly used by other professionals. The history may be hastily presented by healthcare providers when physical care of the infant is warranted. The physical care of a newly born infant in transition often becomes the priority; however, the comprehensive history is equally important. This is especially true when infants are transported from one institution to another. The comprehensive history is imperative in these situations, and identification of potential problems during and after transport is the common goal. All pertinent information must be included. Elements of a complete history start with the identifying data, followed by the chief complaint, and then the interim history of the neonate or the history of the presenting problem or illness, if one exists. This is followed by the antepartum history; obstetric history; intrapartum history; and the maternal medical, family medical, and social histories. In some institutions, these data may be recorded by electronic medical record; in others, the data may be memorialized in a written or dictated history. After the history is compiled, the physical examination data are recorded, followed by any laboratory or radiology studies obtained. An impression and diagnostic differential of the neonate are then formulated, and a plan of care is outlined.

Identifying Data

The identifying data are the patient's name, birth date, and referral source. Both the referring obstetric care provider and the primary care pediatric provider should be listed so that they will receive a copy of the admission history and physical examination. If the patient is admitted to the neonatal intensive care unit, telephone numbers for the primary care providers should be included so that they can receive updates on the neonate's problems and progress. If the patient is transferred from another facility for care, the name of the referring healthcare facility should also be included.

Chief Complaint

The chief complaint in a neonate is often simply the identification, including age, sex, birth weight and current weight (if >1 day of age), gestational age by dates and examination, and any presenting clinical signs the infant exhibits. In the normal newborn, the chief complaint might simply be stated as: "3-kg female, 41 weeks gestation by dates, appropriate for gestational age, now 2 hours of age." Or it may include identified problems, as in the following example:

1. 2.5-kg male, 42 weeks gestation by dates, small for gestational age, 3 hours of age
2. Hypoglycemia
3. Tachypnea: Pneumonia versus retained lung fluid
4. Suspected perinatal sepsis

Interim History

The interim history or history of present illness chronologically records the neonate's history from the time of delivery until the present time and, in the well newborn, should include data regarding temperature stabilization, feeding, voiding, stooling, and behavioral adaptation. In the neonate with identified problems, the sequence of the newborn's problem or problems is discussed, as are laboratory or x-ray findings, interventions, and responses to treatment. The age of the mother, type of delivery, neonate's birth weight and current weight, and Apgar scores are sometimes included to give a more comprehensive picture. Interim histories are often developed and documented when an infant is hospitalized for an extended length of time. A monthly interim history may be helpful in summarizing and developing the final discharge history and physical for a complicated or extremely premature infant.

Antepartum History

The antepartum history includes more specific historical data on the pregnancy: maternal age, gravidity, and parity; last menstrual period; and estimated date of confinement (EDC) or estimated date of delivery (EDD). The EDD may be revised after serial ultrasound evaluation. The date and gestational age at which prenatal care began, who provided the care, and the number of prenatal visits are recorded. Medical complications and high-risk pregnancy factors (Table 2.1), treatments, and monitoring are recorded. The antepartum history should also include information about exposure of the fetus to radiation; over-the-counter, prescribed, or illicit drugs; and tobacco and alcohol use (Table 2.2).

When reviewing the history of the antepartum period, the examiner should note the results of all tests performed on the mother and/or fetus and understand their implications. Usual prenatal screenings may include the following tests: maternal blood type and Rh; antibody screen; serology; venereal disease research laboratory (VDRL) status; rubella immunity testing; cultures for *gonococcus*, *Chlamydia*, and *Group B Streptococcus*; hepatitis B surface antigen screening; α-fetoprotein; HIV testing; triple/quadruple screening; and ultrasound. Women with specific high-risk factors for tuberculosis are tested. HIV antibody screening is now universally recommended in prenatal care.[9] Those who acquire HIV infection during pregnancy require more extensive evaluation.[9] If no HIV screening information is available, an HIV screen of the mother is recommended in labor or soon after delivery.[9] Abnormal findings on routine screening tests will prompt follow-up or specialized screenings.

A relatively new noninvasive prenatal screening (NIPS) test is now available for women at risk for specific genetic disorders, including many aneuploidies and microdeletions. This new screening test known as NIPS utilizes a cell-free analysis of the fetal DNA found in the maternal blood to detect disorders in women at risk for specific genetic disorders, such as trisomies 13, 18, and 21 and other selected genetic microdeletions.[10]

A schedule for routine, general, and specific laboratory examinations during pregnancy appears in Table 2.3. Indications for antepartum fetal assessment are listed in Table 2.4. Appendix A explains many of the routine and some specialized antepartum screenings and also indicates what the results mean.

Obstetric History

The mother's obstetric history is reviewed for the number of pregnancies, abortions, stillbirths, living children, types of deliveries, dates of birth or abortion, birth weights, and gestational ages at birth. Particular attention should be paid to serial ultrasound findings, biophysical profiles, and any nonstress or contraction stress tests. Any previous premature births, neonatal problems, or subsequent major medical problems of prior children should be noted. The present age and health status of living children are recorded. If any child is deceased, the cause and date of death are included.

Intrapartum History

The intrapartum history is evaluated for duration of labor, whether the labor was spontaneous or induced, medications and anesthetics used during labor and delivery, length of the first and second stages of labor, type of delivery, and whether delivery was spontaneous or required forceps or vacuum assistance. The time and duration of rupture of membranes, the use of amnioinfusion, and the status of the amniotic fluid volume and presence or absence of meconium are ascertained. Laboratory and monitoring data obtained during labor are reviewed and noted, as are any maternal complications that occurred during this period (such as fever, bleeding, or hypertension) and the treatment provided. The presentation of the neonate at

TABLE 2.1: CATEGORIZATION OF HIGH-RISK PREGNANCY FACTORS

Socioeconomic Factors

1. Inadequate finances
2. Poor housing
3. Severe social problems
4. Unwed, especially adolescent
5. Minority status
6. Nutritional deprivation
7. Parental occupation

Demographic Factors

1. Maternal age under 16 or over 35 years
2. Overweight or underweight prior to pregnancy
3. Height <5 feet
4. Maternal education <11 years
5. Family history of severe inherited disorders

Medical Factors

A. *Obstetric History*
   1. Infertility
   2. Ectopic pregnancy or spontaneous abortion
   3. Grand multiparity
   4. Stillborn or neonatal death
   5. Uterine/cervical abnormality
   6. Multiple gestation
   7. Premature labor/delivery
   8. Prolonged labor
   9. Cesarean section
   10. Low-birth-weight infant
   11. Macrosomic infant
   12. Midforceps delivery
   13. Baby with neurologic deficit, birth injury, or malformation
   14. Hydatidiform mole or choriocarcinoma

B. *Maternal Medical History/Status*
   1. Cardiac disease
   2. Pulmonary disease
   3. Metabolic disease—particularly, diabetes mellitus, thyroid disease
   4. Chronic renal disease, repeated urinary tract infections, repeated bacteriuria
   5. GI disease
   6. Endocrine disorders (pituitary, adrenal)
   7. Chronic hypertension
   8. Hemoglobinopathies
   9. Seizure disorder
   10. Venereal and other infectious diseases
   11. Weight loss >5 pounds
   12. Malignancy
   13. Surgery during pregnancy
   14. Major congenital anomalies of the reproductive tract
   15. Mental retardation, major emotional disorders

C. *Current Maternal Obstetric Status*
   1. Late or no prenatal care
   2. Rh sensitization
   3. Fetus inappropriately large or small for gestational age
   4. Premature labor
   5. Pregnancy-induced hypertension
   6. Multiple gestation
   7. Polyhydramnios
   8. Premature or prolonged rupture of membranes
   9. Antepartum bleeding
      a. Placenta previa
      b. Abruptio placentae
   10. Abnormal presentation
   11. Postmaturity
   12. Abnormality in tests for fetal well-being
   13. Anemia

D. *Habits*
   1. Smoking during pregnancy
   2. Regular alcohol intake
   3. Drug use/abuse

GI, gastrointestinal.

Source: From Aumann GM, Baird MM. 1993. Risk assessment for pregnant women. In: Knuppel RA, Drukker JE, eds. *High-Risk Pregnancy: A Team Approach.* Philadelphia, PA: Saunders, 23. Reprinted by permission.[6]

delivery, the Apgar scores, any resuscitative interventions performed, and the response to resuscitation are documented. If resuscitation is required, the timing of the interventions is equally important. To ensure an objective determination of the fetal metabolic condition at the time of birth, umbilical cord blood sampling, including both arterial and venous blood, is indicated if there is any degree of perinatal depression.[12] Histologic

TABLE 2.2: MATERNAL MEDICATIONS AND TOXINS: POSSIBLE EFFECTS ON THE FETUS AND/OR NEWBORN

MEDICATION/ TOXIN	POSSIBLE EFFECT ON FETUS/ NEWBORN
Analgesics and Anti-inflammatories	
Aspirin	Hemorrhage, premature closure of ductus arteriosus, pulmonary artery hypertension
Codeine	Neonatal drug withdrawal reported
Ibuprofen	Reduced amniotic fluid volume when used in tocolysis, theoretic risk for premature ductus arteriosus closure
Indomethacin	Closure of fetal ductus arteriosus, pulmonary artery hypertension
Meperidine	Respiratory depression peaks 2–3 hr after maternal dose
Propoxyphene	Drug withdrawal reported, possible increased risk of anomalies
Anesthetics	
General anesthesia	Respiratory depression of infant at delivery if anesthesia is prolonged before delivery
Lidocaine	High fetal serum levels cause CNS depression, accidental direct injection into fetal head causes seizures
Antibiotics	
Aminoglycosides	Ototoxicity reported after first trimester use of kanamycin and streptomycin
Streptomycin	Damage to the eighth cranial nerve, hearing loss
Vancomycin	Potential for ototoxicity
Cephalosporins	Some drugs in this group displace bilirubin from albumin
Isoniazid	Risk for folate deficiency
Metronidazole	Potential teratogen
Sulfonamides	Some drugs in this group displace bilirubin from albumin
Tetracycline	Yellow-brown staining and caries of teeth
Trimethoprim	Folate antagonism
Anticoagulants	
Warfarin (Coumadin)	Warfarin embryopathy
Anticonvulsants	
Carbamazepine	Neural tube defects, midfacial hypoplasia
Phenobarbital	Withdrawal symptoms, hemorrhagic disease
Phenytoin	Hemorrhagic disease, fetal hydantoin syndrome
Trimethadione	Fetal trimethadione syndrome, cleft lip and palate, cardiac and genital anomalies
Valproic acid	Myelomeningocele, facial and cardiac anomalies
Antineoplastics	
Aminopterin	Cleft palate, hydrocephalus, myelomeningocele, growth restriction
Cyclophosphamide	Growth restriction, cardiovascular and digital anomalies
Methotrexate	Absent digits
Antithyroid Drugs	
Iodide-containing drugs	Hypothyroidism

(*continued*)

TABLE 2.2: MATERNAL MEDICATIONS AND TOXINS: POSSIBLE EFFECTS ON THE FETUS AND/OR NEWBORN (*continued*)

Methimazole	Hypothyroidism, cutis aplasia
Propylthiouracil	Hypothyroidism
Antivirals	
Acyclovir	No known adverse effects reported
Ribavirin	Teratogenic and embryolethal in animals
Zidovudine	Potential fetal bone marrow suppression, reduces perinatal HIV transmission
Cardiovascular Drugs and Antihypertensives	
Angiotensin-converting enzyme inhibitors	Neonatal bradycardia, hypoglycemia
β-blockers (propranolol)	Neonatal bradycardia, hypoglycemia
Calcium channel blockers	If maternal hypotension occurs, could affect placental blood flow
Diazoxide	Hyperglycemia
Digoxin	Fetal toxicity with maternal overdose
Hydralazine	Maternal hypotensive risk with possible effect on placental blood flow
Methyldopa	Mild, clinically insignificant decrease in neonatal blood pressure
Diuretics	
Furosemide	Increases fetal urinary sodium and potassium
Thiazides	Thrombocytopenia
Hormonal Drugs	
Androgenics (danazol)	Masculinization of female fetus
Corticosteroids	Cleft palate reported in animals
DES	DES daughters: Genital tract anomalies, increased rate of premature delivery DES sons: Possible increase in genitourinary anomalies
Estrogens, progestins	Uncertain teratogenic potential, virilization of female fetuses reported with progestins
Tamoxifen	DES-like effect in animal studies
Sedatives, Tranquilizers, and Psychotropic Medications	
Barbiturates (short-acting)	Theoretic risk for hemorrhage and drug withdrawal
Benzodiazepines	Hypotonia, impaired thermoregulation
Lithium	Cardiac anomalies (Ebstein's), diabetes insipidus, thyroid depression, cardiovascular dysfunction
Thalidomide	Limb reduction and other anomalies
Tricyclic antidepressants	Association with limb-reduction defects (causation unproven)
Tocolytics	
Magnesium sulfate	Respiratory depression, hypermagnesemia
Ritodrine	Neonatal hypoglycemia
Terbutaline	Neonatal hypoglycemia
Vitamins and Related Drugs	
A	Excessive doses are teratogenic

(*continued*)

TABLE 2.2: MATERNAL MEDICATIONS AND TOXINS: POSSIBLE EFFECTS ON THE FETUS AND/OR NEWBORN (*continued*)

D	Megadoses may cause hypercalcemia
Isotretinoin	Ear, cardiac, CNS, and thymic anomalies
Menadione (vitamin K3)	Hyperbilirubinemia and kernicterus at high doses
Miscellaneous	
Antiemetics	Doxylamine succinate and/or dicyclomine hydrochloride with pyridoxine hydrochloride reported to be a teratogen (unproven)
Irradiation	Fetal death, microcephaly, growth restriction
Methyl mercury	Mental retardation, microcephaly
Oral hypoglycemics	Neonatal hypoglycemia
Social and Illicit Drugs	
Alcohol	Fetal alcohol syndrome or effects
Amphetamines/ methamphetamines	Withdrawal, prematurity, fetal loss, infant death, low birth weight, and small for gestational age[8]
Cocaine	Decreased birth weight; microcephaly; prematurity; abruptio placentae with possible asphyxia, shock, cerebral hemorrhage, stillbirth
Heroin	Increased incidence of low birth weight and small for gestational age, drug withdrawal, impaired postnatal growth, behavioral disturbances
Marijuana	Decreased fetal growth and increased incidence of acute nonlymphoblastic leukemia in childhood
Methadone	Increased birth weight as compared to heroin, drug withdrawal (worse than with heroin alone)
Phencyclidine (PCP)	Irritability, jitteriness, hypertonia, poor feeding
Tobacco smoking	Decreases birth weight by 175–250 g; increased prematurity rate; increased premature rupture of membranes, placental abruption, and placenta previa; increased fetal death

CNS, central nervous system; DES, diethylstilbestrol.

Source: Adapted from Levy J, D'Harlingue AE. 2013. Recognition, stabilization, and transport of the high-risk newborn. In; Fanaroff AA, Fanaroff JM, eds. *Klaus and Fanaroff's Care of the High-Risk Neonate*, 6th ed. Philadelphia, PA: Saunders, 75–77. Reprinted by permission.[7]

findings of the placenta have been found to be an important part of the intrapartum history when defining and confirming perinatal depression.[13] Harteman and colleagues explored placental pathology with the pattern of brain injury in hypoxic–encephalopathy and found associations with decreased placental maturation and chronic villitis.[13]

Maternal Medical History

The maternal medical history is reviewed for chronic health problems and for diseases or disorders treated in the past and/or during the pregnancy. Potential consequences of maternal medical problems for the fetus and/or newborn are listed in Table 2.5. Infections past, present, and during the pregnancy as well as surgical procedures and hospitalizations that occurred before or during the pregnancy are included. Counseling for psychological or social

TABLE 2.3: RECOMMENDED INTERVALS FOR ROUTINE AND SPECIALIZED ANTEPARTUM TESTS

TIME (WEEKS)	ASSESSMENT
Initial (as early as possible)	CBC Urine culture and baseline protein screening Blood group and Rh type determinations Antibody screen Rubella antibody titer measurement Syphilis screen Cervical cytology—Papanicolau test Hepatitis B surface antigen HIV counseling/testing
Optional labs	Hemoglobin electrophoresis PPD Chlamydia Gonococcus Tay Sachs/other testing as indicated by history Hepatitis C testing when/if history suggests increased risk
8–20 (depending on test and maternal/fetal history)	Ultrasound NIPS DNA analysis Maternal serum α-fetoprotein Amniocentesis Chorionic villus sampling—karyotype, amniotic fluid α-fetoprotein
24–28	Diabetes screening Glucose tolerance testing if diabetes screen abnormal Repeat hemoglobin or hematocrit measurement Repeat antibody test for unsensitized Rh-negative patients
28	Prophylactic administration of Rho(D) immune globulin as indicated
32–36 (when indicated)	Ultrasound Testing for sexually transmitted disease Repeat hemoglobin or hematocrit measurement recommended Group B Streptococcus culture at 35–37 wk

CBC, complete blood count; NIPS, noninvasive prenatal screening; PPD, purified protein derivative.

Source: Adapted from American Academy of Pediatrics, American College of Obstetricians and Gynecologists. 2017. *Guidelines for Perinatal Care*, 8th ed. Elk Grove Village, IL: American Academy of Pediatrics, 347–408.[1]

TABLE 2.4: MATERNAL AND FETAL INDICATIONS FOR ANTEPARTUM FETAL ASSESSMENT

MATERNAL	PREGNANCY RELATED
Antiphospholipid syndrome Hyperthyroidism (poorly controlled) Hemoglobinopathies (hemoglobin SS, SC, or S-thalassemia) Cyanotic heart disease Systemic lupus erythematosus Chronic renal disease Type I diabetes Hypertensive disorders	Pregnancy-induced hypertension Decreased fetal movement Oligohydramnios Polyhydramnios IUGR Postterm pregnancy Isoimmunization (moderate to severe) Previous fetal demise (unexplained or recurrent risk) Multiple gestation (with significant growth discrepancy)

IUGR, intrauterine growth restriction.

Source: Data from American College of Obstetricians and Gynecologists. 1999. Antepartum fetal surveillance. *ACOG Practice Bulletin 9*. Washington, DC: ACOG. Reprinted by permission.[11]

problems should also be noted. Medication use, including herbal supplements and over-the-counter preparations for nonpregnancy-related health or medical problems, should be explored.

TABLE 2.5: POTENTIAL EFFECTS OF MATERNAL MEDICAL CONDITIONS ON THE FETUS AND/OR NEWBORN

MATERNAL CONDITION	POTENTIAL FETAL/NEONATAL EFFECTS
Cardiopulmonary	
Asthma	Increased rates of prematurity, toxemia, and perinatal loss
Congenital heart disease	Effects of cardiovascular drugs (see Table 2.2)
Cystic fibrosis	Prematurity, IUGR, fetal loss
Hypertension, preeclampsia	Premature delivery caused by uncontrolled hypertension or eclampsia; uteroplacental insufficiency, abruptio placentae, fetal loss; IUGR; thrombocytopenia; neutropenia
Endocrine/metabolic	
Diabetes mellitus	Hypoglycemia, hypocalcemia, macrosomia, polycythemia, hyperbilirubinemia, increased risk for birth defects, birth trauma, small left colon syndrome, cardiomyopathy, RDS
Hypoparathyroidism	Fetal hypocalcemia, neonatal hyperparathyroidism
Hyperparathyroidism	Neonatal hypocalcemia and hypoparathyroidism
Hyperthyroidism (Grave's disease)	Fetal and neonatal hyperthyroidism, IUGR, prematurity, congestive heart failure, tachycardia
Obesity	Macrosomia, birth trauma
Phenylketonuria (untreated pregnancies)	Mental retardation, microcephaly, congenital heart defects
Hematologic	
Fetal platelet antigen sensitization	Thrombocytopenia, CNS hemorrhage
Idiopathic thrombocytopenia purpura	Reduced iron stores, lower mental and developmental scores at follow up
Iron deficiency anemia	Potential teratogen
Rh or ABO sensitization	Jaundice, anemia, hydrops fetalis
Severe anemia (hemoglobin < 6 mg/dL)	Impaired oxygen delivery, fetal loss
Sickle cell anemia	Increased prematurity, IUGR, fetal distress
Infections	
Chlamydia	Conjunctivitis, pneumonia
Chorioamnionitis	Increased risk for neonatal sepsis, premature labor and delivery
Cytomegalovirus	IUGR, microcephaly, cytomegalovirus inclusion disease, hearing loss
Gonorrhea	Ophthalmia neonatorum
Hepatitis A	Perinatal transmission
Hepatitis B	Perinatal transmission, chronic hepatitis

(*continued*)

TABLE 2.5: POTENTIAL EFFECTS OF MATERNAL MEDICAL CONDITIONS ON THE FETUS AND/OR NEWBORN (*continued*)

MATERNAL CONDITION	POTENTIAL FETAL/NEONATAL EFFECTS
Hepatitis C	Perinatal transmission, chronic hepatitis
Herpes simplex	Encephalitis, disseminated herpes (risk of neonatal disease is much higher when maternal infection is primary rather than recurrent)
HIV	Risk of infectious transmission
Rubella	Rubella embryopathy, cataracts, cardiac defects
Syphilis	Congenital syphilis, IUGR
Toxoplasmosis	IUGR, microcephaly, hydrocephalus, chorioretinitis, myocarditis
Tuberculosis	Perinatal and postnatal transmission
Varicella	Congenital varicella syndrome, rash, pneumonia, myocarditis, encephalitis
Inflammatory/Immunologic	
Inflammatory bowel disease	Increase in prematurity, fetal loss, fetal, and neonatal growth restriction
Systemic lupus erythematosus	Fetal death, spontaneous abortion, heart block, neonatal lupus, thrombocytopenia, neutropenia, hemolytic anemia, endocardial fibrosis
Neuromuscular	
Maternal seizure disorder requiring anticonvulsants	Teratogenic effects of medications (see Table 2.2)
Myasthenia gravis	Transient neonatal myasthenia, preterm delivery
Myotonic dystrophy	Neonatal myotonic dystrophy
Seizure during pregnancy	Fetal hypoxia
Renal/Urologic	
Chronic renal failure	Prematurity, IUGR, fetal loss
Transplant recipients	Prematurity, IUGR, possible effects of maternal immunosuppressive therapy, and mineral disorders
Urinary tract infection	Prematurity

CNS, central nervous system; IUGR, intrauterine growth restriction; RDS, respiratory distress syndrome.

Source: Adapted from Levy J, and D'Harlingue AE. 2013. Recognition, stabilization, and transport of the high-risk newborn. In *Klaus and Fanaroff's Care of the High-Risk Neonate*, 6th ed., Fanaroff AA, and Fanaroff JM, eds. Philadelphia, PA: Saunders, 73. Reprinted by permission.[7]

Family Medical History

The family medical history is reviewed for ages of the infant's mother, father, and siblings; sexes of the other children; and any diagnosed chronic disorders, disabilities, or known hereditary diseases. A genetic history may be required if the neonate is dysmorphic, and further information should be sought regarding other family members affected with a similar disorder. Information about familial disease or disorders can be elicited by asking if anyone else in the immediate or extended family has the same or a related problem, or if any relative has a genetic abnormality or an unusual disease

or disorder. A finding such as a head circumference several standard deviations above normal with normal weight and length percentiles may raise the possibility of an intracranial abnormality. Review of the family history should include questions regarding any similar finding in the baby's parents when they were children. A grandparent can often provide or confirm this information. The most common cause of a large head in an otherwise normal newborn is benign familial megalencephaly, and one of the parents, usually the father, generally had the same finding in infancy.[14] Preparation of a genetic pedigree chart (Figure 2.1) often helps clarify the relationships of affected family members when the history is positive for similar findings in relatives. The question of consanguinity (mating of individuals related by blood) can be posed by asking the parents if they are related in any way besides by marriage and if there is anyone in the family who was related to both parents before their marriage.

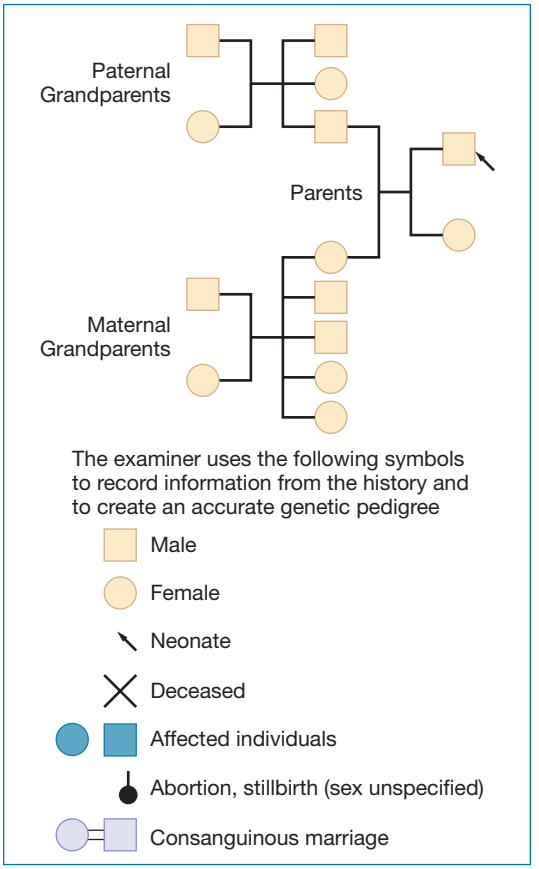

Figure 2.1 Genetic pedigree chart

Social History

The social history includes the mother's marital status, presence or support of the infant's father, and both parents' occupations and education. The family's sources of support in the financial, housing, and care areas are noted. The family's religious affiliation and cultural heritage are included. The family unit should be defined, along with the number of people, family and nonfamily, living in the home. The health status of all household members should also be ascertained. Support agencies with which the family is now working and the family's current social service contacts are important. Members of the family or others who are planning to help with child care after discharge are included. If and when the mother plans to return to work and what arrangements for infant care are planned should be elicited. Information regarding frequent or recent family moves, deaths in the family, and job changes also gives an idea of the stressors affecting the family. This information will assist the examiner in planning for necessary services following discharge or can point to the need for referral to social services.

Remaining Elements

The physical examination is performed, recorded, and/or placed into the electronic medical record. Laboratory or radiology data that have been obtained should be noted. An assessment or impression of the infant's status is then made, including a statement of suspected or potential problems. Here is an example for an infant with problems:

- 3.8-kg preterm female, 35 weeks gestation, born by cesarean section, large for gestational age, infant of a diabetic mother, now 4 hours of age
- Hypoglycemia

- Respiratory distress: tachypnea
- Systolic cardiac murmur at upper left sternal border

An assessment statement for a well newborn with no complications might be similar to the following:

1. 3.5-kg term male, appropriate for gestational age, 4 hours of age
2. Teenage mother with little social support

A plan addressing each of the identified problems or by problems by system should then be developed and documented in the admission history.

SUMMARY

A thorough approach to neonatal history exploration, coupled with analysis of antepartum and intrapartum data, provides a comprehensive view of the neonate's present status and potential health problems. Taking the time to review the history facilitates identifying potential and actual problems in context and, in conjunction with the physical examination findings, formulating priorities of care, and developing a comprehensive plan of care.

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Gestational Age Assessment

Carol Wiltgen Trotter, PhD, RN, NNP-BC

3

A newborn's *gestational age* is defined as the number of completed weeks that have elapsed between the first day of the mother's last menstrual period (LMP) and the day of delivery. Gestational age (or menstrual age) differs from the *postconceptiona*l or *conceptional age*, which is defined as the number of completed weeks from conception until delivery, because ovulation usually occurs about 2 weeks after the first day of the LMP. Defining gestational age using the menstrual age is more accurate because women typically know the date of their LMP and not the date of ovulation.[1] When a preterm infant is conceived using reproductive technologies that allow for identification of the date of conception, the gestational age is actually 2 weeks longer than the conceptional age.[1] Figure 3.1 summarizes the terminology used for age assessment in the perinatal period.

An accurate definition and assessment of gestational age is important to the examiner for two reasons. First, knowledge of the neonate's age and the growth patterns appropriate to that age aid in identification of neonatal risks and in development of management plans. This is particularly true for infants born at or near the limit of viability, as there are numerous medical, social, and ethical issues to consider for the clinician. Accurate assessment of gestational age is critical for determining morbidity and mortality risks that can be used to counsel families regarding treatment decisions.[2] Based on a review of data from 117,236 preterm infants born alive (2013–2014) between 22 and 31 weeks gestation, Pediatrix Medical Group has documented significant differences in mortality for each week of gestation.[3] These differences are highlighted in Figure 3.2 with survival ranging from 11.8% for infants born at 22 weeks gestation to 80.1% for infants born at 25 weeks gestation. In addition, survival without severe intraventricular hemorrhage or severe retinopathy of prematurity for 23-week-gestational-age infants is only 21.8%. Survival without these severe morbidities increases to 41.6% for 24-week-gestational-age infants and to 63.4% for 25-week-gestational-age infants (Figure 3.3).[4] The risks associated with the lowest gestational ages are clearly recognized, and identification of those risks requires accurate assessment of gestational age.

The risks associated with birth at higher gestational ages are also significant. Late preterm infants (34 0/7–36 6/7 weeks gestation) have a different set of problems, and these infants must be identified to improve their management. When compared to the full-term infant, the late preterm infant is at increased risk for temperature instability, hypoglycemia, respiratory distress, apnea, hyperbilirubinemia, and feeding problems.[5,6]

An accurate determination of gestational age is essential for the collection of reliable fetal and infant statistical data that will be used to improve perinatal outcomes.[7] It is also essential when conducting neonatal research and applying the findings to clinical practice. Gestational maturity is an independent predictor of outcome, so gestational age must be controlled for in clinical trials that evaluate new therapies. Accurate gestational age assessment is an important factor when comparing outcomes data from different neonatal care facilities.[1,8]

There are three general methods of determining gestational age in the newborn: (a) calculation of dates based on the mother's

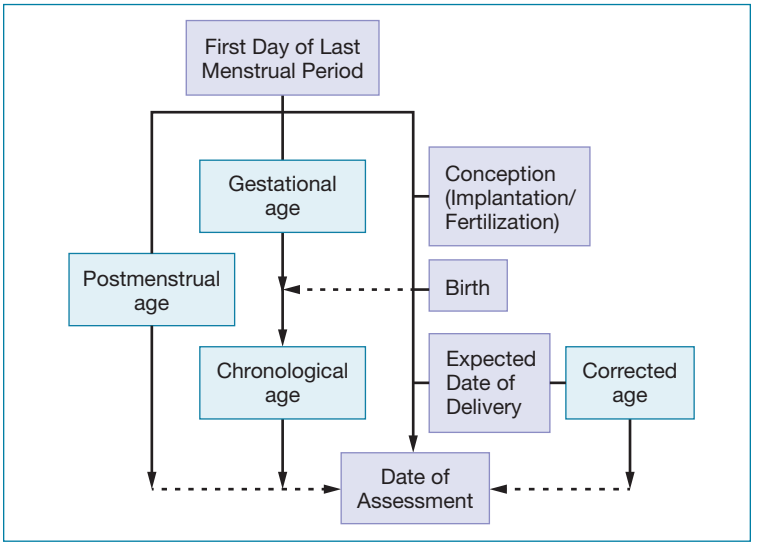

Figure 3.1 Age terminology during the perinatal period

Source: From American Academy of Pediatrics, Committee on Fetus and Newborn. 2004. Age terminology during the perinatal period. *Pediatrics* 114(5): 1363. Reprinted with permission. (Reaffirmed July 2014. http://www.pediatrics.aappublications.org/content/134/3/e920 and published in *Pediatrics* Vol 134, issue 3, September 2014)[1]

Survival by estimated gestational age and birth weight*

Weight Group (gm)	Estimated Gestational Age (wks)										
	22	23	24	25	26	27	28	29	30	31	Overall
250 to 500	6.5%	31.3%	41.2%	52.9%	63.6%						40.5%
501 to 750	16.2%	42.3%	65.1%	77.5%	85.3%	89.2%	88.7%	85.7%			68.7%
751 to 1000			67.9%	85.8%	89.9%	94.0%	97.1%	96.1%	98.7%	97.5%	91.9%
1001 to 1250					91.3%	96.8%	96.6%	98.4%	98.3%	99.3%	97.4%
1251 to 1500						94.6%	96.7%	98.3%	99.4%	99.5%	99.0%
1501 to 1750							96.7%	98.3%	99.0%	99.4%	99.5%
1751 to 2000									98.1%	99.5%	99.9%
2001 to 2250									92.3%	99.5%	99.8%
Overall for EGA	11.8%	40.2%	62.6%	80.1%	88.3%	94.2%	96.0%	97.8%	98.8%	99.4%	

*The outcomes of 117,236 non-anomalous neonates born at, cared for in, and discharged from 278 hospitals in 32 states from 2013 to 2014. Estimated gestational age range was 22 to 42 weeks. Birth weight range was 0.3 to 6.0 kg. For calculations the minimum cell sample size was 20 patients. Data on outcome of infants more than 31 weeks is not presented as their percent survival and percent survival without morbidity approached 100%. These numbers represent an estimate. The likelihood of a good outcome is influenced by many variables, only two of which are estimated gestational age and birth weight.

Figure 3.2 Survival by estimated gestational age and birth weight (%).*

Source: From: Pediatrix Clinical Data Warehouse 2015. Survival by estimated gestational age and birthweight. Sunrise, FL: Pediatrix Medical Group. Reprinted by permission.[3]

Survival without severe IVH or severe ROP, by estimated gestational age and birth weight*

Weight Group (gm)	Estimated Gestational Age (wks)										
	22	23	24	25	26	27	28	29	30	31	Overall
250 to 500	0.0%	13.4%	22.5%	35.3%	50.0%						25.4%
501 to 750	13.5%	23.5%	43.2%	57.1%	70.7%	80.7%	84.5%	85.7%			51.5%
751 to 1000			50.9%	72.7%	77.7%	88.6%	92.0%	93.1%	96.1%	93.7%	83.9%
1001 to 1250					82.0%	89.0%	93.3%	95.3%	96.5%	96.5%	93.4%
1251 to 1500						86.5%	91.4%	94.8%	97.3%	97.9%	96.6%
1501 to 1750							90.0%	97.1%	97.1%	97.9%	97.9%
1751 to 2000									94.8%	97.8%	98.6%
2001 to 2250									92.3%	98.0%	98.4%
2001 to 2250											98.6%
Overall for EGA	7.4%	21.8%	41.6%	63.4%	75.9%	87.5%	91.7%	94.8%	96.8%	97.6%	

*The outcomes of 117,236 non-anomalous neonates born at, cared for in, and discharged from 278 hospitals in 32 states from 2013 to 2014. Estimated gestational age range was 22 to 42 weeks. Birth weight range was 0.3 to 6.0 kg. For calculations the minimum cell sample size was 20 patients. Data on outcome of infants more than 31 weeks is not presented as their percent survival and percent survival without morbidity approached 100%. These numbers represent an estimate. The likelihood of a good outcome is influenced by many variables, only two of which are estimated gestational age and birth weight.

Figure 3.3 Survival without severe IVH or severe ROP, by estimated gestational age and birthweight. IVH, intraventricular haemorrhage; ROP, retinopathy of prematurity

Source: From Pediatrix Clinical Data Warehouse 2015. Survival without severe IVH or severe ROP, by gestational age and birthweight. Sunrise, Florida, Pediatrix Medical Group. Reprinted by permission.[4]

LMP, (b) evaluation of obstetric parameters obtained during the prenatal period, and (c) physical examination of the neonate. Although evaluations based on menstrual dates and obstetric methods are discussed briefly, this chapter focuses on determining gestational age through postnatal physical assessment.

The average pregnancy is usually described as lasting 280 days, or 40 weeks from the first day of the LMP.[9] The accuracy with which the estimated date of confinement (EDC) can be determined based on the LMP depends on the woman having regular menstrual cycles with ovulation occurring on the 14th day after the start of the LMP.[9] If the mother's menstrual cycles are irregular, ultrasound measurements provide the most accurate estimate of gestational age.

Obstetric methods for assessing gestational age include maternal physical examination and ultrasound measurements. Fetal ultrasound measurements are the most powerful methods for assessing gestational age. The best fetal parameters to measure by ultrasound depend on the timing of the exam.

In the first trimester, crown–rump length is the most accurate fetal measurement for determining gestational age.[10] During the second and third trimesters, however, the fetus is too large to use the crown–rump length measurement. Instead, measurements of individual structures are used. The four most common measurements are biparietal diameter, head and abdominal circumferences, and femur length.[9,11] Chervenak and colleagues evaluated 238 pregnancies to determine the accuracy of the four fetal biometric measurements in assigning gestational age. They found head circumference to be the single best predictor, followed by abdominal circumference, biparietal diameter, and then femur length.[12] Head circumference may be more reliable than

the biparietal diameter because normal head shape variants such as dolichocephaly do not alter the head circumference measurement.[11] Advances have been made in improving the accuracy of gestational age estimation by obstetric methods to such an extent that some healthcare professionals believe that formal assessment of gestational age is not required for routine newborn examination. The most accurate guide to gestational age is a combination of menstrual dates and early ultrasound measurements.[13] However, when these data are not known, physical examination is appropriate. When rapid assessment of gestational age is necessary, such as in the delivery room, a quick inspection of a few parameters will suffice. Physical assessment techniques used in the detailed examination of the neonate for gestational age, however, are inspection, palpation, and use of the ophthalmoscope.

HISTORICAL PERSPECTIVE

The gestational age assessment scoring system that provides the basis for many of the tools currently used was published in 1970 by Dubowitz, Dubowitz, and Goldberg.[14] The system is based on assessment of 11 external and 10 neurologic criteria. The external criteria were taken from characteristics defined by Farr and associates.[15] According to Dubowitz, Dubowitz, and Goldberg, and Koenigsberger (as well as secondary sources), the development of neurologic criteria to assess gestational age originated in the work of the French schools in the 1950s under Andre Thomas and later, Madame Sainte-Anne Dargassies.[14,16] During the 1960s, a number of other investigators helped better define the neurologic criteria. Dubowitz, Dubowitz, and Goldberg selected criteria from the data published by Koenigsberger, Robinson, and Amiel-Tison to develop the neurologic portion of their scoring system.[14,16–18]

Since publication of the system devised by Dubowitz, Dubowitz, and Goldberg, many variations of the tool have been used.[19,20] In 1978, Capurro and coworkers published a simplified method of gestational age assessment using seven of the original variables defined by Dubowitz, Dubowitz, and Goldberg. These investigators identified five physical and two neurologic criteria that most accurately determine gestational age.[21] In 1979, Ballard, Novak, and Driver published a tool using six neuromuscular criteria and six physical criteria.[22] In addition, other investigators have published reports of determination of gestational age by measuring hand and foot length,[23–25] cerebellar vermis dimensions,[26] postnatal sonographic femur length,[27] and intermamillary distance.[24] Skin reflectance with an optical fiber spectrophotometer was also measured.[28]

Both the Ballard and the Dubowitz tools have been criticized for overestimating the actual gestational age by 2 weeks.[29,30] Therefore, Ballard and colleagues expanded their tool to achieve greater accuracy and to include the extremely premature neonate.[31] According to its authors, this tool, the New Ballard Score (NBS), overestimates gestational age by 0.3 to 0.6 weeks (2–4 days) at gestational ages less than 37 weeks. However, the NBS estimates of gestational age have been shown to exceed actual gestational age by 1.3 to 3.3 weeks in neonates less than 28 weeks.[32] Because of this discrepancy, Sola and Chow state that there is still no absolute "gold standard" for postnatal assessment of gestational age, especially for neonates less than 28 weeks gestation.[33] Healthcare professionals involved in making clinical decisions for extremely premature neonates must be cautious when applying the results of physical examination data. Because of this variability in accuracy of gestational age assessment, Jacob and Hulman developed an algorithm for gestational age determination (Figure 3.4).[8] They place an emphasis on accurate gestational age assignment for the purpose of comparing outcomes data between institutions. The key component of this algorithm is the obstetrician's assessment of gestational age based on menstrual dates, ultrasound parameters, and maternal physical assessment. The algorithm recommends newborn assessment only if the obstetric parameters are uncertain. The limitations associated with the NBS for gestational age assessment must be kept in mind as it is used throughout this chapter to demonstrate assessment of the neuromuscular and physical criteria associated with various gestational ages.

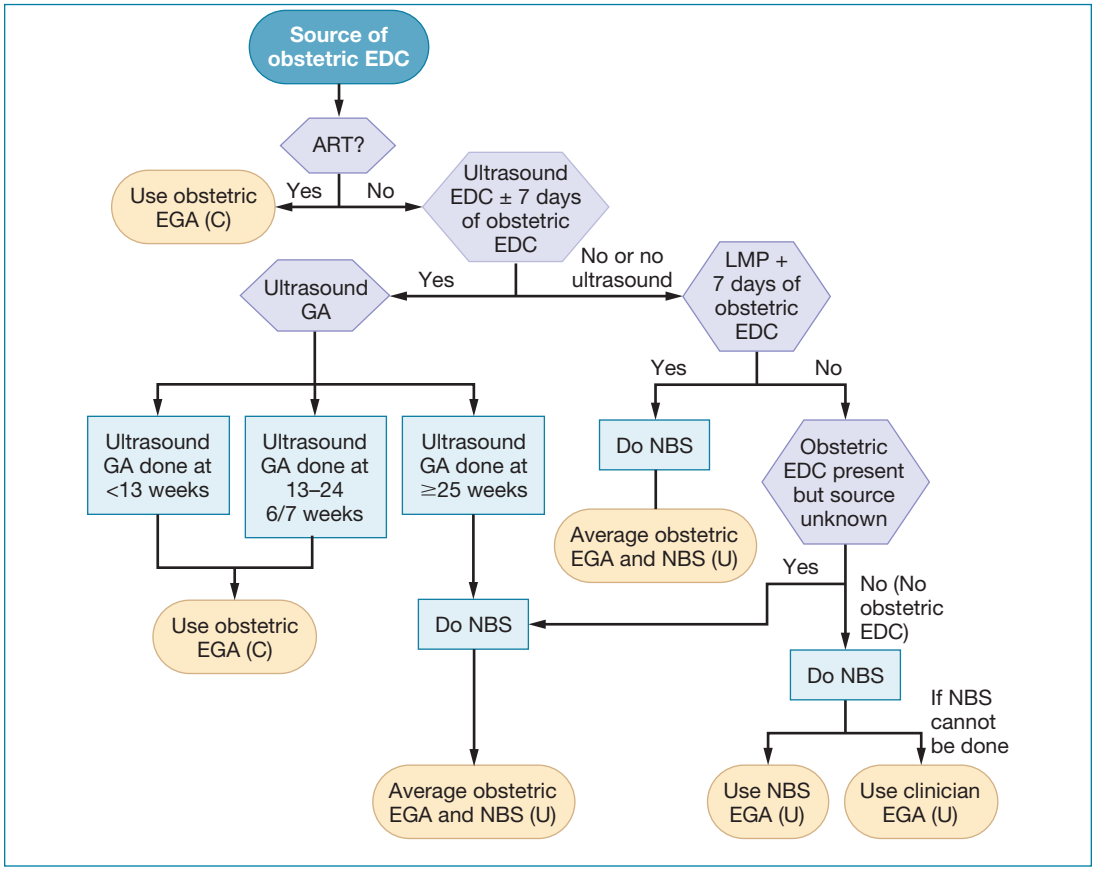

Figure 3.4 Algorithm for the assignment of gestational age. ART, assisted reproductive technology; C, certain; EDC, estimated date of confinement; EGA, estimated gestational age; GA, gestational age; LMP, last menstrual period; NBS, New Ballard Score; U, uncertain.

Source: Adapted from Jacob J, and Hulman S. 2006. A standardized method for assigning gestational age: A tool for measuring gestational age based newborn intensive care outcomes. *American Journal of Perinatology* 23(7): 399. Reproduced with permission of Thieme Medical Publishers via Copyright Clearance Center.[8]

PHYSICAL ASSESSMENT

There are two methods for determining gestational age by physical examination: (a) assessment of the anterior vascular capsule of the lens (AVCL) using the ophthalmoscope and (b) assessment of neuromuscular and physical criteria by inspection and palpation.

Anterior Vascular Capsule of the Lens

The rationale and technique for examining the AVCL to determine gestational age were described in 1977 by Hittner, Hirsch, and Rudolph.[34] The hyaloid system and the tunica vasculosa lentis are transient embryologic vascular systems that invade the developing eye. The purpose of the vascular system is to nourish the eye during active growth. This system can be seen starting at approximately 27 weeks gestation; it then atrophies progressively until it is gone after week 34. Figure 3.5 depicts the developing eye from 5 weeks to term gestation. Early in fetal life, the lens is invaded by the tunica vasculosa lentis (Figure 3.5C). The hyaloid artery supplies the developing lens and the tunica vasculosa lentis until it disappears late in the fetal period. At that time, the tunica vasculosa lentis atrophies.[35]

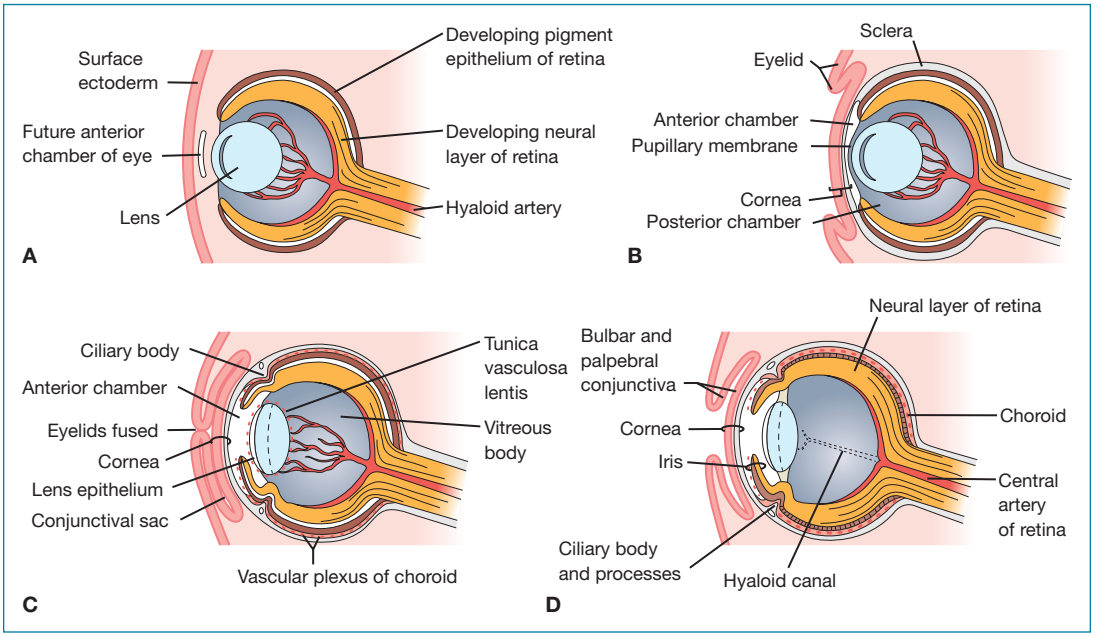

Figure 3.5 The sagittal sections of the eye, showing successive developmental stages of the lens, retina, iris, and cornea

Source: Adapted from Moore KL, Persaud TVN, and Torchia MG. 2016. *The Developing Human: Clinically Oriented Embryology*, 10th ed. Philadelphia, PA: Saunders, 424. Reprinted by permission.[35]

To visualize the vessels, the examiner uses a direct ophthalmoscope set between +6 and +12. These settings allow the examiner's eye to focus on the lens rather than on the retina. To the novice practitioner, the image will initially appear blurry. As the ophthalmoscope is moved closer to the infant's eye (within 6–10 in.), the vascular system will come into view as the examiner focuses on the more anteriorly placed lens.

Figure 3.6 illustrates the grading system for gestational age assessment based on the pattern and presence of vessels noted. This examination must be performed within the first 24 to 48 hours of life because the vascular system atrophies rapidly after that period.[34] Although Dietz, Huppi, and Amato confirmed that the gradual disappearance of the AVCL is not impacted by maternal and neonatal risk factors, Hadi and Hobbs found that maternal chronic hypertension accelerates the atrophy of this system.[36,37] They suggest that chronic intrauterine stress may influence the maturation of this vascular structure.

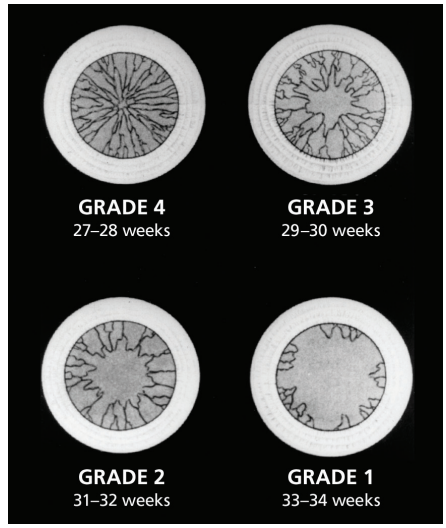

Figure 3.6 Gestational age assessment grading system: Anterior vascular capsule of the lens

Source: From Hittner HM, Hirsch NJ, and Rudolph AJ. 1977. Assessment of gestational age by examination of the anterior vascular capsule of the lens. *Journal of Pediatrics* 91(3): 456. Reprinted by permission.[34]

Neuromuscular and Physical Criteria

Ballard and colleagues have defined six neuromuscular and six physical criteria for evaluating gestational age in the newborn.[31,38] The criteria and the scoring tool are depicted in Figure 3.7. A discussion of each criterion follows, and photographs depicting assessment of the criteria accompany the text. Descriptions of the technique for assessment were taken from Dubowitz, Dubowitz, and Goldberg and from Ballard unless otherwise noted.[15,31,38,39]

Maturational Assessment of Gestational Age (New Ballard Score)

Name_____ **Date/time of birth**_____ **Sex** _____

Hospital no._____ **Date/time of exam** _____ **Birth w eight** _____

Race_____ **Age when examined** _____ **Length**_____

Apgar score: 1 minute _____ **5 minutes** _____ **10 minutes**_____ **Head circ.** _____

Neuromuscular Maturity **Examiner**_____

Neuromuscular Maturity Sign	Score –1	0	1	2	3	4	5	Record Score Here
Posture								
Square Window (Wrist)	>90°	90°	60°	45°	30°	0°		
Arm Recoil		180°	140°–180°	110°–140°	90°–110°	<90°		
Popliteal Angle	180°	160°	140°	120°	100°	90°	<90°	
Scarf Sign								
Heel to Ear								
							Total Neuromuscular Maturity Score	

Score
Neuromuscular____
Physical_____
Total_____

Maturity Rating

Score	Weeks
–10	20
–5	22
0	24
5	26
10	28
15	30
20	32
25	34
30	36
35	38
40	40
45	42
50	44

Physical Maturity

Physical Maturity Sign	Score –1	0	1	2	3	4	5	Record Score Here
Skin	sticky friable transparent	gelatinous red translucent	smooth pink visible veins	superficial peeling and/or rash, few veins	cracking pale areas rare veins	parchment deep cracking no vessels	leathery cracked wrinkled	
Lanugo	none	sparse	abundant	thinning	bald areas	mostly bald		
Plantar Surface	heel-toe 40–50 mm:-1 <40 mm:-2	>50 mm no crease	faint red marks	anterior transverse crease only	creases anterior 2/3	creases over entire sole		
Breast	imperceptible	barely perceptible	flat areola no bud	stippled areola 1–2 mm bud	raised areola 3–4 mm bud	full areola 5–10 mm bud		
Eye/Ear	lids fused loosely: -tightly: -21	lids open pinna flat stays folded	sl. curved pinna; soft; slow recoil	well-curved pinna; soft but ready recoil	formed and firm; instant recoil	thick cartilage ear stiff		
Genitals (Male)	scrotum flat, smooth	scrotum empty faint rugae	testes in upper canal rare rugae	testes descending few rugae	testes down good rugae	testes pendulous deep rugae		
Genitals (Female)	clitoris prominent & labia flat	prominent clitoris & small labia minora	prominent clitoris & enlarging minora	majora & minora equally prominent	majora large minora small	majora cover clitoris and minora		
							Total Physical Maturity Score	

Gestational Age (weeks)
By dates_____
By ultrasound_____
By exam_____

Figure 3.7 Gestational age assessment scoring system: Neurologic and physical criteria (New Ballard score)

Source: From Ballard JL, et al. 1991. New Ballard score, expanded to include extremely premature infants. *Journal of Pediatrics* 119(3): 418. Reprinted by permission.[31]

Neuromuscular Criteria

Posture. It is an indicator of resting muscle tone. The infant is first allowed to assume a baseline position of comfort and is then observed in the supine position. A score is assigned based on the degree of flexion of the arms, knees, and hips. The degree of hip adduction/abduction is also noted. The neonate demonstrates increasing flexion and hip adduction with advancing gestational age; lower extremity flexion precedes flexion of the upper extremities (Figures 3.8–3.10).

Square window. The infant's fingers are extended, and the hand is flexed to the forearm by applying gentle pressure on the dorsum of the hand. Enough pressure is applied to get as full a flexion as possible without rotating the wrist. The angle between the forearm and the palm is measured. The angle decreases with advancing gestational age (Figures 3.11–3.13).

Arm recoil. This criterion focuses on passive flexor tone of the biceps muscle. The neonate's arms are flexed for 5 seconds while he is in the supine position. The arms are then fully extended by pulling the hands and quickly releasing them. The examiner should avoid maintaining the extremity in the extended position for too long, thereby fatiguing the flexors. Degree of arm flexion and the strength of the recoil are scored. A sluggish response with little or no flexion receives a low score. A brisk, fully flexed response receives a high score. According to Ballard, a score of 4 requires that the fist come in contact with the face (Figures 3.14 and 3.15).[39]

Popliteal angle. The popliteal angle maneuver evaluates passive flexor tone in the knee. The neonate should be in the supine position with the pelvis on the mattress. The untested portion of the extremity should be resting quietly on the supporting surface,

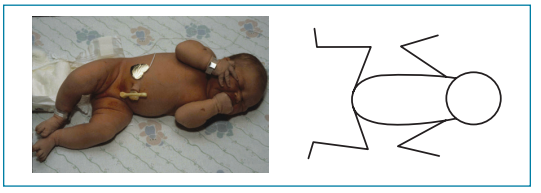

Figure 3.8 Full-term infant. Posture score = 4

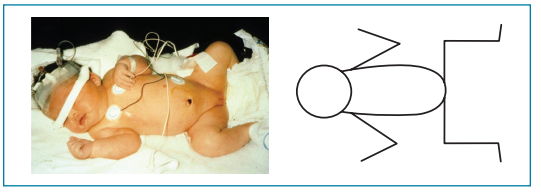

Figure 3.9 Preterm large-for-gestational-age female infant. Posture score = 3

Figure 3.10 Preterm infant with no arm flexion. Posture score = 1

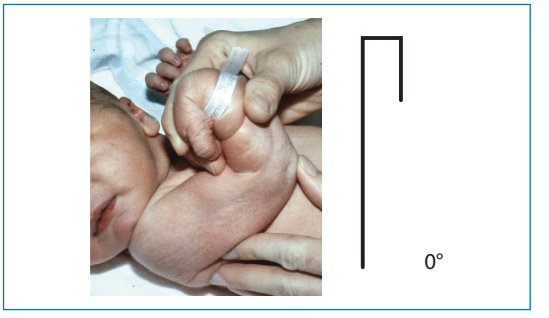

Figure 3.11 Square window in term neonate. Score = 4

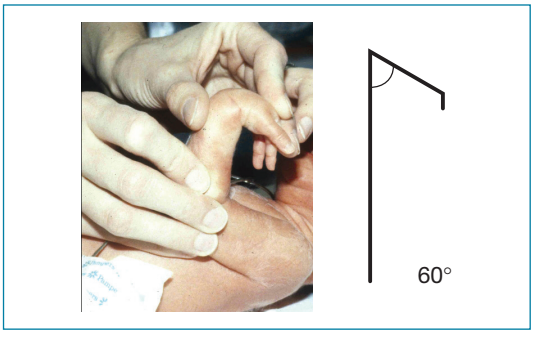

Figure 3.12 Square window in preterm infant. Score = 1

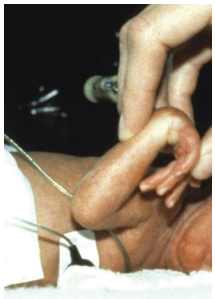


Figure 3.13 Square window in preterm infant. Score = 0

A. Arms are flexed for five seconds and then extended.
B. After arms are extended, they are released, and the degree of arm flexion is scored. Score = 4.

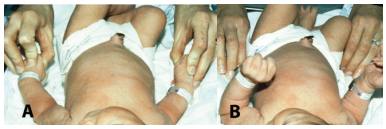

Figure 3.14 Eliciting arm recoil in a term infant

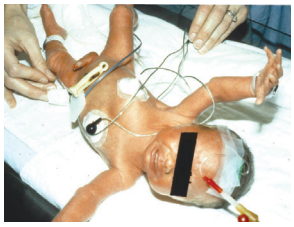
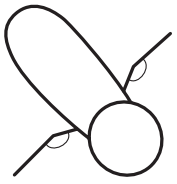

Figure 3.15 Arm recoil maneuver elicited Moro response in this preterm infant. Score = 2

and the thigh should be supported from the side without interfering with the flexor being tested.[39] With the thumb and index finger of one hand, the examiner holds the infant's knee and thigh adjacent to the chest/abdomen. At the same time, the examiner grasps the foot and extends the leg gently with the other index finger. The popliteal angle (the angle between the lower leg and thigh, posterior to the knee) is measured. This angle decreases with advancing gestational age (Figures 3.16–3.18).

Scarf sign. With the neonate supine and the head in the midline position, the

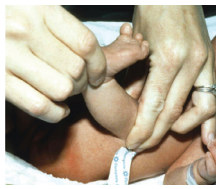
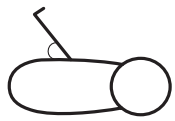

Figure 3.16 Popliteal angle in term infant. Score = 5

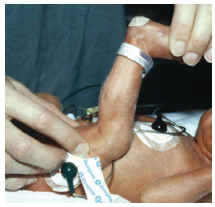
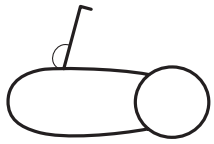

Figure 3.17 Popliteal angle in preterm infant. Score = 2

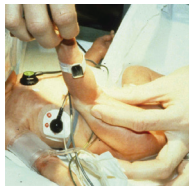
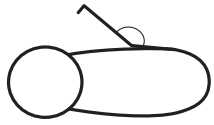

Figure 3.18 Popliteal angle. Score = 1

examiner grasps the infant's hand and pulls the arm across the chest and around the neck. The arm should be gently pulled across the chest and posteriorly as far as possible around the opposite shoulder. The examiner scores this item based on the relationship of the elbow to the midline of the body when the arm is pulled across the chest. The neonate demonstrates increasing resistance to this maneuver with advancing gestational age (Figures 3.19 and 3.20).

Heel to ear. This maneuver evaluates passive flexor tone surrounding the pelvic girdle. With the infant supine and the pelvis flat on the table, the examiner gently grasps one foot with the thumb and index finger and draws the foot as near to the ear as possible without forcing it.[18] The thigh should be positioned along the side of the body. When significant

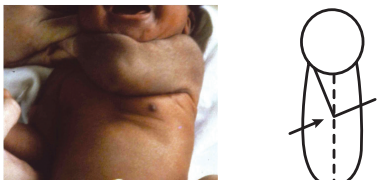

Figure 3.19 Scarf sign in term infant. Elbow drawn to midline of the body. Score = 2

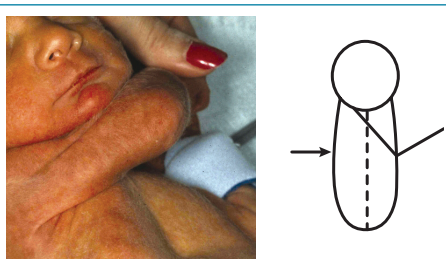

Figure 3.20 Scarf sign in preterm infant with elbow past midline. Score = 1

resistance is detected, the examiner notes the distance between the foot and the ear, as well as the degree of knee extension. The location of the heel relative to the body landmarks is scored. The neonate demonstrates increasing resistance to this maneuver with advancing gestational age (Figures 3.21 and 3.22).

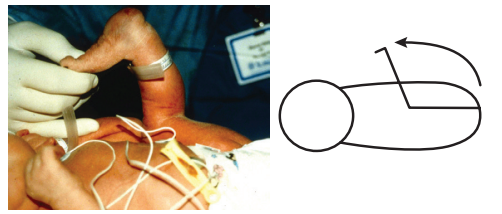

Figure 3.21 Heel to ear. Score = 2

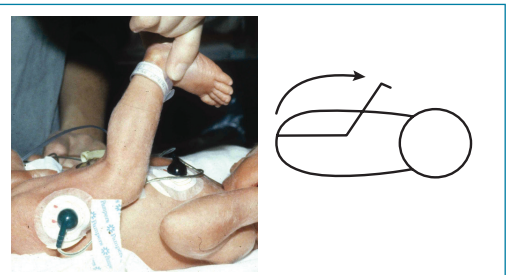

Figure 3.22 Heel to ear maneuver in preterm infant. Score = 1

Physical Criteria

Skin. The examiner assesses skin texture, color, and opacity. As the neonate matures, more subcutaneous tissue develops, and there is a gradual loss of the vernix caseosa. Veins become less visible, and the skin becomes more opaque (Figures 3.23–3.25).

Lanugo. It is the fine, downy hair present on the body of the neonate. Its production begins toward the end of the 12th week. Lanugo is most abundant over the back (particularly between the scapulae), although it will be noted over the face, legs, and arms as well. Thinning of lanugo begins on the back, and at term most of the lanugo on the back is gone (Figures 3.26–3.28).

Plantar surface. In the extremely premature neonate, the examiner assesses the

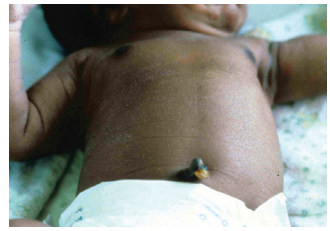

Figure 3.23 Skin of term infant. Score = 4

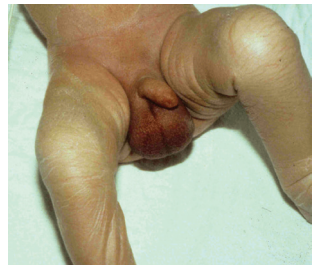

Figure 3.24 Skin of postterm infant. Score = 5

plantar surface of the foot for length and, as the neonate matures, for creases. Foot length is measured from the tip of the great toe to the back of the heel. Creases should appear

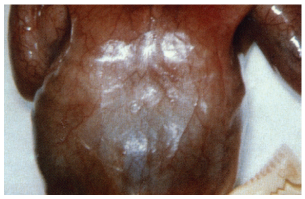

Figure 3.25 Friable, transparent skin of extremely preterm infant. Score = 1

Source: From Clark DA. 2000. *Atlas of Neonatology.* Philadelphia, PA: Saunders, 2. Reprinted by permission.[40]

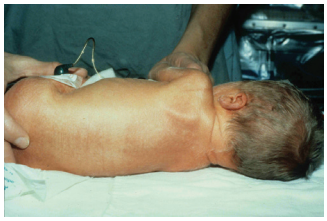

Figure 3.26 Lanugo over shoulders and cheek. Score = 3

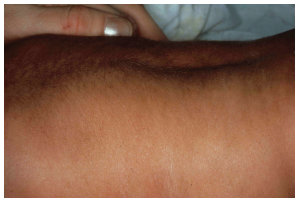

Figure 3.27 Lanugo over back of preterm infant. Score = 2

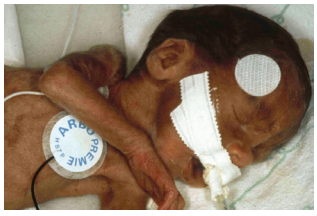

Figure 3.28 Lanugo noted over arm, shoulder, and face of preterm infant. Score = 1

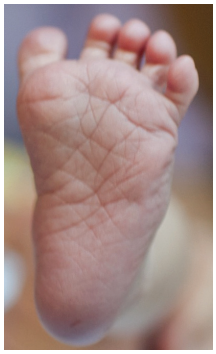

Figure 3.29 Sole creases covering the foot of a term neonate. Score = 4

Courtesy of Nancy Borowick.

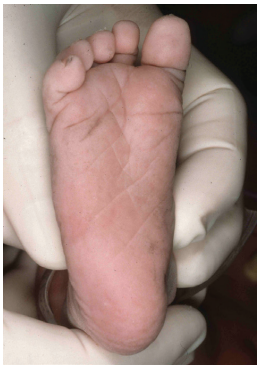

Figure 3.30 Sole creases covering the anterior two-thirds of the foot. Score = 3

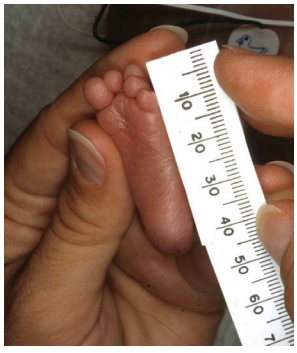

Figure 3.31 Faint red marks over the soles of the feet of a preterm infant. Score = 1

between 28 and 30 weeks gestation and should cover the entire plantar surface at or near term (Figures 3.29–3.31).

Breast. This is assessed by observing the size of the areola and for stippling, and it is palpated to determine the amount

of breast tissue. Breast tissue is measured in millimeters. Breast tissue increases and areola development progresses with advancing gestational age, but the nutritional status of the fetus may affect the amount of breast tissue present (Figures 3.32–3.34).

Ear/Eye. The ear is assessed by observing its form, by palpating for the amount of cartilage present in the pinna, and for the recoil of the pinna when it is folded and released (Figures 3.35–3.37). In a mature neonate, the pinna will snap back rapidly when released. In a very immature neonate, there is a lack of cartilage in the pinna, so it may even remain folded when released. For an infant born at a very early gestation, the eyes are evaluated

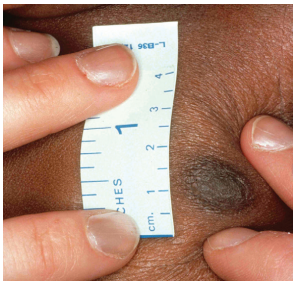

Figure 3.32 Evaluation of breast tissue in postterm infant. Score = 4

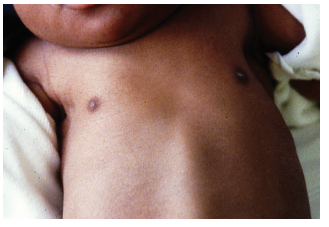

Figure 3.33 Breast tissue in a term neonate. The breast bud is 3–4 mm, and the areola is raised. Score = 3

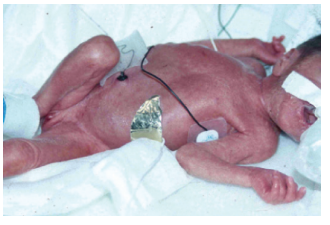

Figure 3.34 Breast tissue in a preterm neonate. There is no breast bud, and the areola is flat. Score = 0

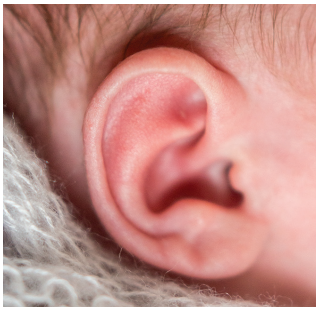

Figure 3.35 Fully formed ear of term infant. Score = 4

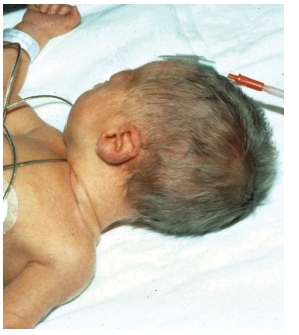

Figure 3.36 Ear of preterm infant with a partially curved pinna. Score = 1

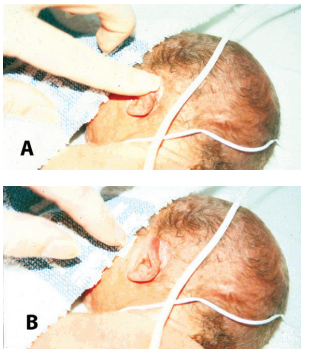

Figure 3.37 Eliciting recoil of pinna

based on fusion of the eyelids, which occurs at approximately 9 to 10 weeks gestation. The eyelids open during the 26th to 28th week. Ballard and associates consider loosely-fused lids (–1) as closed but able to be separated by gentle manipulation and tightly fused lids (–2) as unable to be separated with gentle manipulation.[31] They collected data on this criterion at gestational weeks 20–28. These data are depicted in Figure 3.38. All eyelids were fused at less than 23 weeks and open at 28 weeks. Between 23 and 27 weeks gestation, there is variability between the numbers of infants with tightly fused, loosely fused, and open eyelids. According to Ballard, the variability in eyelid fusion at each gestational age may be due to certain stress-related factors.[39]

Genitalia. The male genitalia are assessed for the presence of the testes, the degree of descent of the testes into the scrotum, and the development of rugae—creases that appear over the scrotum. Testicular descent from the peritoneal cavity begins at approximately 30 weeks gestation, and both testicles should be palpable in the inguinal canal by 34 weeks gestation. Rugae become more prominent as the scrotal sac thickens with advancing gestation (Figures 3.39–3.41).

The female genitalia are assessed with the hips abducted to about 45 degrees. Scoring is based on the prominence of the clitoris as well as the development of the labia minora and majora. With advancing gestational age, the labia majora and minora become more developed so that at term they completely cover the clitoris (Figures 3.42 and 3.43).

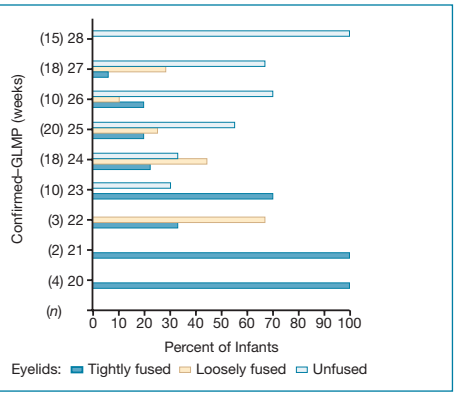

Figure 3.38 Eyelid fusion at gestational weeks 20 to 28 by C-GLMP. C-GLMP, confirmed-gestational age by last menstrual period

Source: From Ballard JL, et al. 1991. New Ballard score, expanded to include extremely premature infants. *Journal of Pediatrics* 119(3): 422. Reprinted by permission.[31]

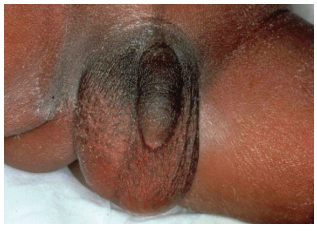

Figure 3.39 Male genitalia in postterm infant. Score = 4

Figure 3.40 Deep rugae of male genitalia. Score = 4

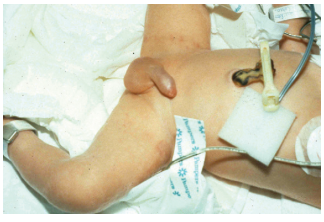

Figure 3.41 Male genitalia in preterm neonate. Score = 0

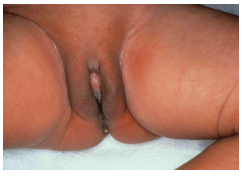

Figure 3.42 Female genitalia in term infant. Score = 3

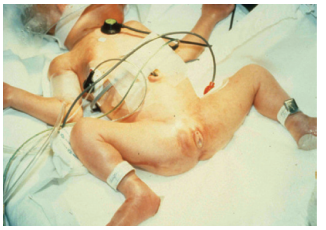

Figure 3.43 Female genitalia in preterm infant. Score = 0

The labia majora contain fat; therefore, the nutritional status of the neonate will impact the size. The labia majora in neonates who have been nutritionally deprived in utero may be small relative to the clitoris and labia minora, causing the examiner to score the infant as less mature. The opposite is true for neonates who have been exposed to overnutrition in utero. According to Ballard, the criteria should be scored as observed, not as the examiner thinks it should be scored because of other influences (e.g., intrauterine nutritional status).[39] The total score will most likely be balanced out when the neuromuscular and physical scores are added together.

Scoring the Physical Examination

After the infant has been examined and a score assigned to each criterion, the score for each category, neuromuscular and physical, is determined. These two scores are added to obtain the final maturity rating score. As shown in Figure 3.7, the final maturity rating score (in the shaded area) is matched with the corresponding gestational age in weeks (in the unshaded column to the right). If the maturity rating score is 15, for example, the infant is assigned a gestational age of 30 weeks. The NBS authors do not give guidelines for what gestational age to assign when the maturity score falls between the numbers listed. In my experience, a gestational age that most closely approximates the maturity rating is chosen. For example, if the maturity rating score is 23, a gestational age of 33 weeks is assigned. It is also appropriate to consider the gestational age determined by dates and obstetric measurements in making a final determination of gestational age.

Infants born before 37 completed weeks of gestation are considered preterm. Infants born between the beginning of week 38 and the completion of week 41 of gestation are considered term. Infants born at 42 weeks gestation or later are considered postterm.

Timing of the Physical Examination

There are no consistent, specific guidelines for optimal timing of the gestational age examination. Ballard and associates state that in extremely premature neonates, prompt examination at a postnatal age of less than 12 hours is essential to ensure validity of the examination.[31] Ballard, Novak, and Driver previously recommended examining all infants between 30 and 42 hours of age.[22] This short wait allows for stabilization and adjustment of the neonate to extrauterine life. Koenigsberger states that the neurologic examination is of little value during the first 48 hours of life because tone and reflexes change rapidly with extrauterine adjustment.[16] Amiel-Tison suggests repeating the examination performed on the first day of life again at 2 to 3 days of age because tone changes in the days following birth. Amiel-Tison also states that the optimal time for neuromuscular evaluation is 1 hour before feeding (when the infant is neither too sleepy nor too agitated) because both sleepiness and agitation affect tone.[18] When conducting their study, Dubowitz, Dubowitz, and Goldberg performed all assessments within 5 days of delivery. They found that when they did multiple assessments on 70 neonates, the assessments were as reliable during the first 24 hours as during the subsequent 4 days of life.[14]

Much of the concern regarding gestational age assessment and timing of the examination relates to neuromuscular adjustment following birth. This is further complicated by conditions altering neuromuscular function, such as asphyxia or medications

administered during the pre- and postnatal periods. However, Ballard states that the items used to evaluate passive tone on the NBS are not affected by the perinatal events that profoundly affect active tone.[38] This may be the reason that Dubowitz, Dubowitz, and Goldberg did not find differences in the gestational age assessments performed on the first day of life and those done at 4 days of life.[14]

Still, it is clear that controversy exists as to the optimal timing for the gestational age examination. In general, it is advisable that the initial examination be performed within the first 48 hours of life. Follow-up examinations may be indicated if the findings from the first examination disagree with gestational age assessments based on menstrual dates or obstetric methods. However, most healthcare professionals will assign the gestational age based on dates and obstetric measurements. When using the NBS tool, if the neuromuscular condition of the neonate is unreliable at birth, the examiner may assess the physical criteria, multiply the score by a factor of two, and then assign a gestational age based on this total score. Although the validity of this approach has not been addressed, it is based on the fact that Dubowitz, Dubowitz, and Goldberg, when analyzing their data, found that external characteristics scored collectively gave a better age index than did the neurologic criteria scored collectively.[14] However, the total score using both parameters was the most accurate.

EVALUATION OF GROWTH INDICES

When the gestational age of the neonate has been determined, the examiner plots the gestational age by the neonate's weight, length, and Occipital-frontal circumference (OFC) on growth charts such as those seen in Figure 3.44. For example, an examiner using the weight chart would define a neonate as being appropriate for gestational age (AGA) if his weight falls between the 10th and 90th percentiles. A small-for-gestational-age (SGA) neonate is one whose weight falls below the 10th percentile for gestational age. A large-for-gestational-age (LGA) neonate is one whose weight falls above the 90th percentile for gestational age. These classifications are depicted in Figure 3.44. On the basis of the relationship between weight and gestational age, each neonate will fall into one of nine categories: preterm SGA, AGA, or LGA; term SGA, AGA, or LGA; or postterm SGA, AGA, or LGA. These classifications are important because neonatal risk factors are identified based on intrauterine growth patterns and the gestational age of the neonate. Problems of neonates who are SGA are summarized in Table 3.1.

A low birth weight infant is defined as one with a birth weight of less than 2,500 g. A very low birth weight infant is one weighing 1,500 g or less. An extremely low birth weight infant weighs 1,000 g or less.[2,44,45] An intrauterine growth restricted (IUGR) neonate is one who has not grown at the expected in utero rate for weight, length, or OFC. Generally, an IUGR infant demonstrates restricted fetal growth subsequent to a pathophysiologic process occurring during the perinatal period. Table 3.2 summarizes the clinical features of the IUGR infant. The term IUGR has also been used interchangeably with SGA, which is not appropriate. Practically speaking, IUGR infants are often SGA, but this is not always the case. Some neonates may be growth restricted, yet not fall below the 10th percentile. For example, a 36-week-gestational-age infant with a birth weight of 2,000 g is well below the 50th percentile for weight on the graph depicted in Figure 3.44, which indicates a significant restriction of growth. However, this infant does not fall below the 10th percentile for weight so cannot be identified as SGA using this graph. Additionally, not all infants identified as SGA have experienced a pathologic process. They may simply be infants who are constitutionally small based on genetics, race, or ethnicity and represent one of the normal outliers for the newborn population.[47] There are two types of IUGR patterns: symmetric and asymmetric. Generally, symmetric growth restriction involves the head,

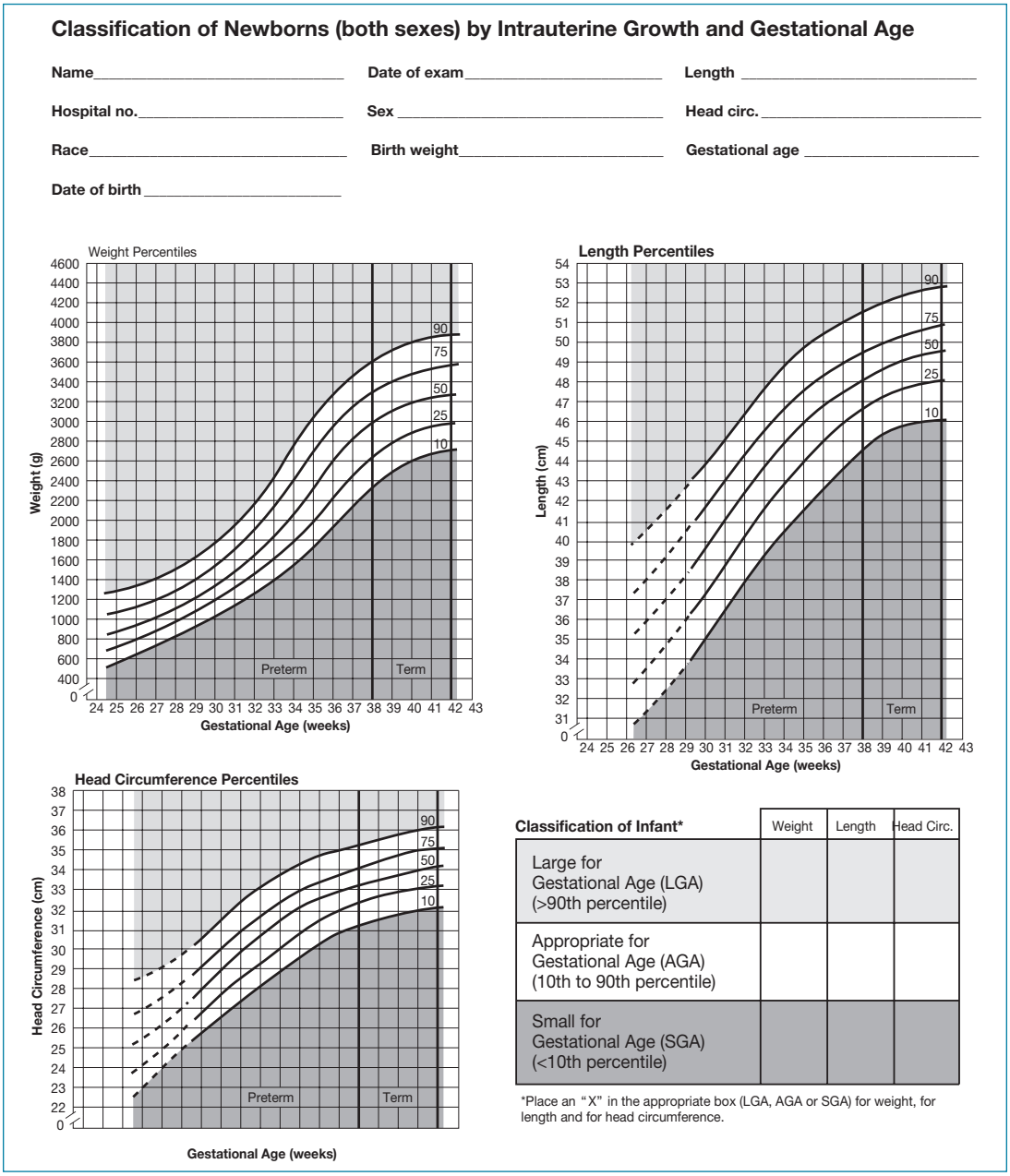

Figure 3.44 Intrauterine growth/gestational age charts

Source: From Ross Laboratories publication 10–91; (0.05) A-58560. Constructed from Battaglia FC, and Lubchenco LO. 1967. A practical classification of newborn infants by weight and gestational age. *Journal of Pediatrics* 71(2): 159–163[41]; Lubchenco LO, Hansman C, and Boyd E. 1966. Intrauterine growth in length and head circumference as estimated from live births at gestational ages from 26 to 42 weeks. *Pediatrics* 37(3): 404[42]. Reprinted by permission.

length, and the infant's weight. Asymmetric growth restriction involves just the infant's weight. Table 3.3 summarizes the characteristics of these two types of infants.[46] Generally, symmetric IUGR is caused by factors that produce a diminished overall growth rate and typically exert their effects early in gestation. Examples include congenital viral

TABLE 3.1: PROBLEMS OF NEONATES WHO ARE SMALL FOR GESTATIONAL AGE

PROBLEM	PATHOGENESIS	ASSESSMENT/PREVENTION/ TREATMENT
Fetal death	Placental insufficiency, chronic fetal hypoxia	Biophysical profile Vessel velocimetry Cordocentesis Maternal O_2 Early delivery
Asphyxia	Acute fetal hypoxia superimposed on chronic fetal hypoxia, acidosis Placental insufficiency ↓ cardiac glycogen stores	Antepartum and intrapartum monitoring Efficient neonatal resuscitation
Meconium aspiration pneumonia	Hypoxic stress	Pharyngeal–tracheal aspiration
Fasting hypoglycemia	↓ hepatic glycogen ↓ gluconeogenesis ↓ counterregulatory hormones Cold stress Asphyxia–hypoxia	Early oral or intravenous alimentation or both
Alimented hyperglycemia	"Starvation diabetes"	Glucose infusion not to exceed 8 mg/kg/min except with hypoglycemia Consider insulin if transient or permanent neonatal diabetes mellitus; consider insulin resistance at receptor or postreceptor level
Polycythemia/ hyperviscosity	Placental transfusion Fetal hypoxia Erythropoietin	Neonatal partial exchange transfusion
Temperature instability	Cold stress Poor fat stores Catecholamine depletion Hypoxia, hypoglycemia Reduced fasting O_2 consumption	Neutral thermal environment Early alimentation
Dysmorphology	TORCH Syndrome complexes Chromosome disorders	Disease-specific therapy or prevention
Teratogen exposure		Disease-specific therapy or prevention

(*continued*)

TABLE 3.1: PROBLEMS OF NEONATES WHO ARE SMALL FOR GESTATIONAL AGE (*continued*)

PROBLEM	PATHOGENESIS	ASSESSMENT/PREVENTION/ TREATMENT
Pulmonary hemorrhage (rare)	Hypothermia, polycythemia Hypoxemia/DIC	Avoid cold stress, hypoxia Endotracheal administration of epinephrine PEEP
Immunodeficiency	"Malnutrition" effect TORCH	Unknown Specific therapy if available
Decreased bone mineral density	Possible substrate deficiency or altered vitamin D metabolism	Appropriate postnatal oral calcium and vitamin D intake

DIC, disseminated intravascular coagulation; PEEP, positive end-expiratory pressure; TORCH, toxoplasmosis, other (e.g., congenital syphilis and viruses), rubella, cytomegalovirus, and herpes simplex virus association.

Source: From Kliegman RM. 2011. Intrauterine growth restriction. In *Neonatal-Perinatal Medicine: Diseases of the Fetus and Infant*, 9th ed., Martin RJ, Fanaroff AA, and Walsh MC, eds. Phildelphia, PA: Mosby, 267. Reprinted by permission.[43]

TABLE 3.2: CLINICAL FEATURES OF INTRAUTERINE GROWTH RESTRICTION

Relatively large head compared with whole body
Shrunken abdomen with "scaphoid" appearance (must be distinguished from diaphragmatic hernia)
Loose skin, sometimes dry, peeling, with the appearance of "hanging," occasionally meconium stained
Long fingernails, especially in term and postterm infants with severe IUGR, occasionally meconium stained
Face with shrunken appearance or wizened
Widened or overriding cranial sutures, anterior fontanel larger than usual
Thin umbilical cord, sometimes meconium stained

Source: From Subhani M. 2005. Intrauterine growth restriction. In *Intensive Care of the Fetus and Neonate*, 2nd ed., Spitzer AR, ed. Philadelphia, PA: Mosby, 139. Reprinted by permission.[46]

infections, single-gene defects, and chromosomal disorders. These conditions have a significant impact on cell replication and overall growth potential. Infants with asymmetric IUGR are typically those with normal OFC and length measurements, but who have a relatively low weight. These infants appear thin and wasted, with a head that is disproportionately large when compared with body size. This pattern of growth restriction is associated with poor placental perfusion or nutritional deficits to the fetus during the third trimester. Conditions predisposing the neonate to asymmetric growth restriction include maternal preeclampsia, poor caloric intake during pregnancy, and chronic fetal distress.

LGA infants are frequently born to diabetic mothers with poor glucose control. Maternal hyperglycemia results in fetal hyperglycemia and hyperinsulinemia. The high insulin levels act as a fetal growth hormone, causing macrosomia and resulting in hypoglycemia postnatally. The term macrosomia is frequently used interchangeably with LGA. A macrosomic infant is one who weighs more than 4,000 g at birth. Risks associated with macrosomic or LGA infants are typically related to difficult deliveries and the postnatal hyperinsulinemia that occurs after prolonged maternal hyperglycemia.

SUMMARY

Assessment of the neonate's gestational age is an essential component of the neonatal physical examination. Knowledge of gestational

TABLE 3.3: SYMMETRIC AND ASYMMETRIC INTRAUTERINE GROWTH RESTRICTION

	SYMMETRIC	ASYMMETRIC
Etiology	Usually intrinsic to the infant	Usually extrinsic to the infant
Affected gestation	Quite early, in first trimester	Most in third trimester, some in late second trimester onward
Body affected	Yes	Yes
Bone growth affected	Yes	No
Biparietal diameter	Low profile type	Low flattening type
Brain affected	Yes, symmetrically to body size	No (known as "brain sparing")
Ponderal index	Normal	Low
Genetic disorders	Yes	No
Risk for hypoglycemia	Low	High
Risk for perinatal asphyxia	Low	High
Blood flow in internal carotid artery	Normal	Redistribution
Maternal and fetal arterial waveform velocity	Normal	Decreased
Glycogen and fat content	Relative	Decreased
Fetal distress	No	Yes
Examples	Chromosomal disorders, genetic disorders, infections (e.g., TORCH)	Chronic fetal distress, preeclampsia, chronic hypertension

TORCH, toxoplasmosis, rubella, cytomegalovirus, herpes simplex

Source: From Subhani M. 2005. Intrauterine growth restriction. In *Intensive Care of the Fetus and Neonate*, 2nd ed., Spitzer AR, ed. Philadelphia, PA: Mosby, 139. Reprinted by permission.[46]

age and appropriate growth patterns assists the examiner in identifying potential risks to the neonate. With experience and a cooperative neonate, the examiner should be able to perform the gestational age examination in 5 minutes. Infants born with altered neuromuscular states may require an additional gestational age examination at a later date.

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Skin Assessment

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4

Assessment of the skin is an important element of the newborn physical examination. Valuable information regarding the newborn's health and well-being can be obtained by observing the color, integrity, and characteristics of the skin. Close examination can aid the practitioner in determining gestational age, nutritional status, functioning of organ systems, and the presence of cutaneous or systemic disease.

Newborn rashes, birthmarks, and lesions can be a source of questions and anxiety for parents. Those caring for newborns must be well versed in normal and abnormal variations, for both educational and diagnostic purposes.

A complete examination of the skin involves both inspection and palpation. Most skin variations can be noted by inspection alone, but palpation is essential to avoid missing less obvious problems. Palpation is also important for determining the thickness, turgor, and consistency of the skin.

ANATOMY AND PHYSIOLOGY

The skin serves a number of basic functions in the newborn. These include physical and immunologic protection, thermoregulation, sense perception, and self-cleaning. An understanding of the skin's structure is essential to careful examination and recognition of irregularities in appearance or function.

The skin consists of three layers: the epidermis, the dermis, and the subcutaneous layer (Figure 4.1). The epidermis, or outermost layer, is subdivided into five layers. The top layer is the stratum corneum, consisting of dead cells that are constantly being brushed off and replaced. Although the term newborn has the same number of layers as adult skin, it is thinner, and does not function as well.[1,2] The epidermis is only about 0.01 mm to 0.05 mm thick. Close adherence of the stratum corneum cells to one another prevent chemicals and microorganisms from entering the body and prevent significant insensible water loss through the skin in the term newborn, child, and adult.[1,2]

The lower layers of the epidermis contain keratin-forming cells and melanocytes. Melanocytes are pigment-producing cells that provide protection from ultraviolet radiation. Different numbers of melanocytes are found in various regions of the skin, with more in the facial area than on the trunk.[2] The amount of pigmentation in the skin depends on the development of the melanocytes, with more advanced melanocytes producing and dispersing more pigment. Melanin production is immature in the newborn, increasing the newborn's sensitivity to sunlight.[2]

The dermis lies directly under the epidermis. The epidermis and dermis are attached by means of a basement membrane referred to as the *dermal–epidermal junction*. This junction consists of various structures that anchor and attach the epidermis to the dermis. This membrane is well established in the fetus in the third trimester, although the anchoring is less secure than in the adult.[1,2]

The dermis consists of fibrous and elastic tissues, sweat glands, sebaceous glands, and hair shafts, as well as blood vessels and cutaneous nerves. The fibrous, elastic tissues are what give the dermis its strength and elasticity. Sweat glands and blood vessels

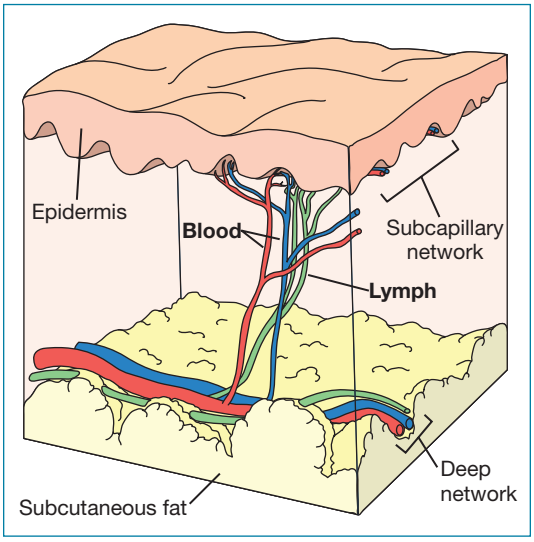

Figure 4.1 Cross section of human skin and the anatomic relationships among the various structures

Source: Adapted from Nasemann T, Sauerbrey W, and Burgdorf W. 1983. *Fundamentals of Dermatology.* New York, NY: Springer-Verlag, 11. Reprinted by permission.[3]

help to maintain body temperature, and cutaneous nerves protect the skin from injury and provide the sensations of touch, temperature, and pain.

The third layer of the skin consists of subcutaneous fat. This fat layer serves as insulation, protection for internal organs, and calorie storage. It is less developed in the preterm or low-birth-weight newborn.

The skin of the newborn is similar to that of the adult in its basic structure. As with any other organ, however, the less mature the newborn, the less mature the function of the skin.

The more immature the skin, the thinner and more permeable it is. Fibrils that connect the dermis and the epidermis are more fragile in the newborn's term and preterm skin than in adult skin, and the stratum corneum is thinner.[1,2] Sweat glands, although present at birth, do not reach full adult functioning until the second or third year of life.

Fetal skin is covered in utero with vernix caseosa, a greasy white or yellow material composed of sebaceous gland secretions, proteins, and exfoliated skin cells. Vernix becomes thicker during the third trimester and provides protection for the skin in utero. It gradually decreases as the fetus approaches 40 weeks gestation.[2,4]

The fetus is also covered with a fine, soft, and downy type of hair called *lanugo* while in utero. Lanugo first appears at approximately 20 weeks gestation and covers most of the body, including the face. Most of it disappears by 40 weeks gestation.

BEGINNING THE EXAMINATION

Several factors influence the appearance of the neonate's skin and affect the examiner's ability to note normal and abnormal variations. A consistent, systematic approach to examining the skin increases the likelihood of gathering all the information.

A family and maternal history and an account of the labor and delivery are useful to the examiner because they may highlight items that should be inspected with extra care. For example, a forceps- or vacuum-assisted delivery may lead to disruption of skin integrity. A family history of neurofibromatosis would alert the examiner to search for café au lait patches.

Before beginning the examination, consider the newborn's environment. The amount and type of light and the temperature of the room will affect the appearance of the skin. Adequate lighting, preferably bright natural light, is important. Ideally, the newborn should be examined under a radiant heat source to allow for complete undressing and visualization. This allows the examiner to observe color and vascular changes.

To begin the examination, undress the newborn and inspect the general color, consistency (smooth, peeling), thickness, and opacity of the skin. Note the distribution of hair and any obvious markings or anomalies. Then begin a closer inspection of the newborn. It is important to follow the same pattern every time to avoid missing subtle deviations. Many examiners find it easiest to begin at the head and progress downward toward the feet. While looking closely at the skin, note the size, color, and placement

TABLE 4.1: DERMATOLOGIC TERMINOLOGY

When reporting skin irregularities, correct terminology facilitates accurate description of observations. The following terms are commonly used to define skin irregularities.

Bulla	A vesicle >1 cm in diameter containing serous or seropurulent fluid
Crust	A lesion consisting of dried serous exudate, blood, or pus on the surface of the skin
Cyst	A raised, palpable lesion with a fluid- or semisolid-filled sac
Ecchymosis	A large area of subepidermal hemorrhage, initially bluish black in color, then changing to greenish brown or yellow; it does not blanch with pressure
Lesion	An area of altered or abnormal tissue
Macule	A discolored, flat spot <1 cm in diameter that is not palpable
Nodule	An elevated, palpable lesion with indistinct borders; some of the lesion is palpable below the skin outside the elevated area
Papule	An elevated, palpable lesion, solid and circumscribed, <1 cm in diameter
Patch	A macule >1 cm in diameter
Petechia	A small, purplish, hemorrhagic spot on the skin, the size of a pinpoint
Plaque	An elevated, palpable lesion with circumscribed borders >1 cm, or a fusion or coalescence of several papules
Purpura	A small hemorrhagic spot larger than a petechia, 1–3 mm in size
Pustule	An elevation of the skin filled with cloudy or purulent fluid
Scale	An exfoliation of dead or dying bits of skin; can also result from excess keratin
Vesicle (blister)	An elevation of the skin filled with serous fluid; <1 cm in diameter
Wheal	A collection of fluid in the dermis that appears as a reddened, solid elevation of the skin

Note: It is important to describe the color of the lesion and its distribution. Lesions may be arranged in distinct patterns, such as linear or circular, or as distinct separate lesions.

of any discolorations, markings, or variations (Table 4.1). The infant should be turned over for a complete examination of the back. Inspect all skin crevices, including the axillae and the groin.

As stated earlier, palpation is an important aspect of the examination. It permits the examiner to assess the underlying dermis, thickness of the skin and subcutaneous fat, presence of edema, and any irregularities in texture or consistency. Palpation is also necessary to determine the size or configuration of certain lesions or variations that may be observed during the examination.

Blanching of lesions can be assessed by pressing the pad of the thumb or finger against the lesion for a few seconds and observing for color changes when the pressure is removed. Hydration and nutrition can also be assessed by examination of the skin. Poor skin turgor or loose, hanging skin indicates dehydration or poor nutritional status. It is important to remember that poor skin turgor is a late sign of dehydration. Excessive fluid, or edema, can be noted by pressing the pad of the thumb into the skin and looking for pitting. Edema may be noted especially in dependent areas or on the scalp following delivery. Most newborns will have some edema around the face and eyes as a result of excess fluid volume after delivery.

To determine the amount of subcutaneous fat, the examiner pinches a skin fold between thumb and forefinger. Loose skin folds will be present in the term infant who has a decreased amount of fat—for example, the infant who has intrauterine growth restriction. Loose skin may also be seen in the postterm infant who has lost some weight in the week or 2 before delivery.

RECOGNIZING COMMON VARIATIONS IN NEWBORN SKIN

A number of changes occur in newborn skin during the first few days and weeks after birth. Most are benign, transient lesions that do not require therapeutic intervention. It is important to be familiar with these normal changes and to be able to distinguish them from signs of serious disease.

Color Variations

Acrocyanosis

Acrocyanosis refers to bluish discoloration of the palms of the hands and the soles of the feet. Most infants present with acrocyanosis at birth. It may persist for up to 48 hours after delivery and is exacerbated by low environmental temperatures. It is benign in the otherwise normal infant. Circumoral (around the mouth) cyanosis may also be present during the first 12 to 24 hours after birth. Acrocyanosis that persists beyond the first few days after birth, or circumoral cyanosis lasting longer than 24 hours should be investigated.

Plethora

Plethora describes the ruddy, or red, appearance present in some newborns. It may be indicative of a high hemoglobin level. The infant who is plethoric at birth and demonstrates clinical signs, such as hypoglycemia or respiratory distress, should have hemoglobin and hematocrit checked by heelstick or central venipuncture. A newborn with a central hematocrit greater than 65% is considered polycythemic and should be watched closely for hypoglycemia, cyanosis, respiratory distress, or jaundice.[5]

Jaundice

Jaundice is the term used to describe a yellow coloring of the skin and the sclera. The color is caused by the deposit of bile pigment that is a result of hyperbilirubinemia, or excess bilirubin in the blood. The presence of jaundice in a newborn should be noted, with particular attention paid to the age of the newborn and the degree of jaundice present. As a general rule, jaundice first appears in the head or face, and then progresses head to toe as the bilirubin level rises. Because it is difficult, if not impossible, to estimate serum bilirubin levels on the basis of clinical appearance, transcutaneous or serum bilirubin levels must be obtained to determine the presence of hyperbilirubinemia.

The bluish mottling or marbling of the skin seen in response to chilling, stress, or overstimulation is called *cutis marmorata.* It is caused by the constriction of capillaries and venules and usually disappears when the newborn is warmed. Persistent cutis marmorata may be seen in newborns and infants with trisomy 21, trisomy 18, and Cornelia de Lange syndrome.[6,7]

Harlequin Color Change

Seen only during the newborn period, harlequin color change appears to be more common in the low-birth-weight infant.[8] When the newborn is lying on one side, a sharply demarcated red color is seen in the dependent half of the body, with the superior half appearing pale (Figure 4.2). If the newborn is rotated to the other side, the color reverses.

The color change may last anywhere from 1 to 30 minutes and disappears slowly if the newborn is returned to the back or abdomen. This phenomenon occurs in both healthy and ill newborns and is of no pathologic significance.[8,9] It has been attributed to an immature autonomic vasomotor control.[9] This phenomenon can be observed in the first 3 weeks of life.

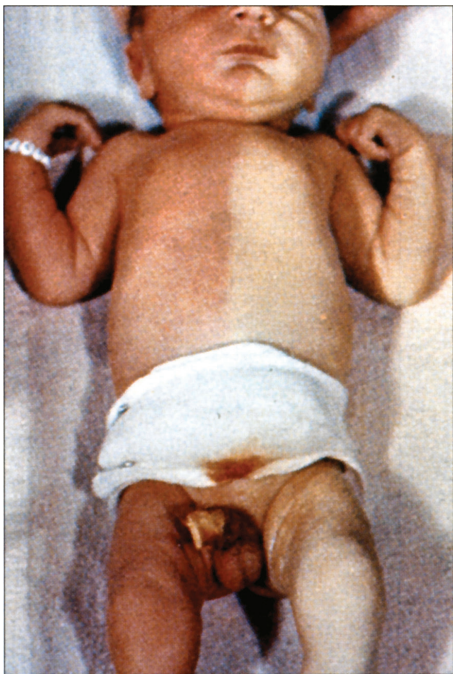

FIGURE 4.2 Harlequin color change

Source: From Solomon LM, and Esterly NB. 1973. *Neonatal Dermatology*. Philadelphia, PA: Saunders. Reprinted by permission.[10]

Common Newborn Skin Lesions

A benign rash, *erythema toxicum* is found in up to 70% of term newborns.[9] The rash consists of small white or yellow papules or vesicles with erythematous bases. It may be found on any part of the body, but it is most commonly seen on the face, trunk, and extremities (Figure 4.3). This rash often disappears and then reappears moments or hours later on a different part of the body. It may last anywhere from a few hours to several days. The peak incidence is from 24 to 48 hours of life, but it can continue to occur up to 3 months of age. The cause is unknown; however, it may be exacerbated by handling or chafing of linen. No treatment is necessary. In fact, use of lotions or creams may make the rash worse.[9]

Diagnosis is generally made by visual recognition of the eruption. A definitive diagnosis can be made by observation of the presence of numerous eosinophils in a smear of an aspirated papule.

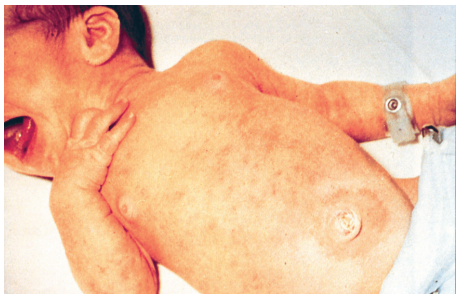

Figure 4.3 Erythema toxicum

Courtesy of Barbara Quissell, MD, Presbyterian/St. Luke's Medical Center, Denver, CO.

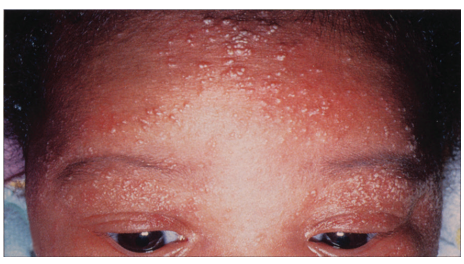

Figure 4.4 Milia

Source: From Cohen BA. 2013. *Pediatric Dermatology*, 4th ed. Philadelphia, PA: Saunders, 22. Reprinted by permission.[11]

Milia

Milia are multiple yellow or pearly white papules about 1 mm in size (Figure 4.4). They are usually found on the brow, cheeks, and nose of up to 40% of newborns.[9] When found in the midline of the palate, they are called *Epstein pearls*; those found along the lingual and buccal parts of the dental ridges, away from the midline, are referred to as *Bohn nodules*. Milia are epidermal cysts caused by accumulation of sebaceous gland secretions. They resolve spontaneously during the first few weeks of life.

Sebaceous Gland Hyperplasia

Sebaceous gland hyperplasia is characterized by numerous tiny (<0.5 mm) white or yellow papules found on the nose, cheeks, and upper lips. These enlarged sebaceous glands are caused by maternal androgenic stimulation

and occur in approximately 50% of newborns.[9] They will spontaneously decrease in size after birth and require no treatment.

Miliaria (Heat Rash)

Miliaria is caused by obstruction of the sweat ducts as a result of an excessively warm, humid environment. This rash is seen primarily over the forehead, on the scalp, in skin creases on the neck or thighs, or in the groin area. Miliaria is classified as one of three types, depending on its severity.

Miliaria crystallina consists of clear, thin vesicles, 1 to 2 mm in diameter that develop in the epidermal portion of the sweat glands. The vesicles rupture about 24 to 48 hours after appearance, leaving a white scale.

Prolonged obstruction of the ducts of the sweat glands, leading to release of sweat into adjacent tissue, is termed *miliaria rubra* and is accompanied by a prickly sensation (prickly heat). Miliaria rubra appears as small erythematous papules about 2 to 4 mm in diameter. This rash is found most often in the neck, axilla, and groin areas, where sweat is likely to accumulate. Continued occlusion results in leukocyte infiltration of the papule. If the condition is not resolved, it can lead to a secondary staphylococcal infection of the deeper dermal portions of the sweat glands. This condition, termed *miliaria profunda*, is extremely rare and seen mostly in tropical locations.[12]

Treatment of miliaria consists of eliminating precipitating factors, such as excessive heat and humidity. Eliminating overheating and keeping the infant dry often cause the lesions to resolve within a few hours.

Sebaceous Nevus

Sebaceous nevus is a small yellow or yellowish-orange papule or plaque most often found on the scalp or face, but may be seen on other parts of the body. It consists of immature hair follicles and sebaceous glands. The sebaceous nevus is devoid of hair when found on the scalp because of the rudimentary hair follicles found in the lesion. It may remain unchanged until puberty, when it enlarges and becomes thicker due to stimulation of the sebaceous glands found in the lesions. There is some risk for secondary tumors developing in these lesions, including basal cell or other neoplasms; therefore, surgical removal may be recommended.[8,9,12] Multiple sebaceous nevi may indicate epidermal nevus syndrome (Solomon syndrome), which can include skeletal, ocular, and cerebral anomalies.[8,9,12]

Common Pigmented Skin Lesions

Hyperpigmented Macule (Mongolian Spot)

The most common pigmented lesion in the newborn, the hyperpigmented macule is seen in up to 90% of African American, Asian, and Hispanic infants and up to 10% of Caucasian infants.[13] These large macules or patches are seen most commonly over the buttocks, flanks, or shoulders (Figure 4.5). They are gray or blue-green in color. Hyperpigmented macules may be mistaken for bruising because

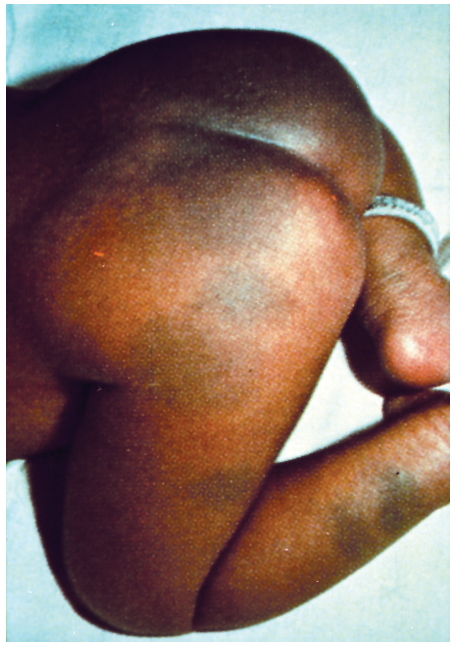

Figure 4.5 Hyperpigmented macule

Courtesy of Barbara Quissell, MD, Presbyterian/St. Luke's Medical Center, Denver, CO.

of their color and location. It is important to document the size and distribution of the lesion to avoid later suspicion of nonaccidental trauma.

Hyperpigmented macules are caused by melanocytes that infiltrate the dermis. The macules tend to fade gradually over the first 3 years of life and will become less apparent as the infant's skin darkens. Some may persist into adulthood.

Transient Neonatal Pustular Melanosis

Transient neonatal pustular melanosis begins with superficial, vesiculopustular lesions (Figure 4.6), often causing some alarm when present at birth. These vesicles rupture within 12 to 48 hours, leaving small pigmented macules. The macules are often surrounded by a ring of very fine white scales. Any stage or combination of stages (vesicles, pustules, or scaling of ruptured vesicles) may be present at birth. These small hyperpigmented macules may remain for up to 3 months after birth. Transient neonatal pustular melanosis is benign, requiring no treatment. Aspiration of the vesicles reveals numerous neutrophils and almost no eosinophils. This skin lesion is found in up to 5% of African American infants and about 0.2% of Caucasian infants.[9,14]

Congenital Melanocytic Nevus (Pigmented Nevus)

A dark-brown or black macule, the congenital melanocytic nevus is caused by proliferation of melanocytes within the epithelial structures that sometimes extend into the subcutaneous fat. The pigmented nevus is most commonly seen on the lower back or buttocks, but may occur anywhere on the body and is seen in all ethnicities.[15,16] The skin may be smooth over the lesion or may be nodular or irregular. Lesions range in size from less than 1 cm to more than 20 cm in diameter (Figure 4.7). Lesions may be with or without tufts of hair. Small- to moderate-sized lesions (<20 cm in diameter) are generally benign, but may pose a small risk for developing malignant melanoma and should be either excised or watched closely.[15,16] Two percent to 5% of larger lesions (>20 cm in diameter) can demonstrate malignant changes.[16] These larger lesions should be observed closely for changes in size or shape, which is common during childhood and may require skin biopsy.[16] Pigment-specific laser therapy may be useful in reducing the size of some lesions, but surgical excision may be required.[16–18] Repigmentation may occur with either laser ablation or surgical excision because some nevomelanocytes may remain in the dermis after the procedure.[18]

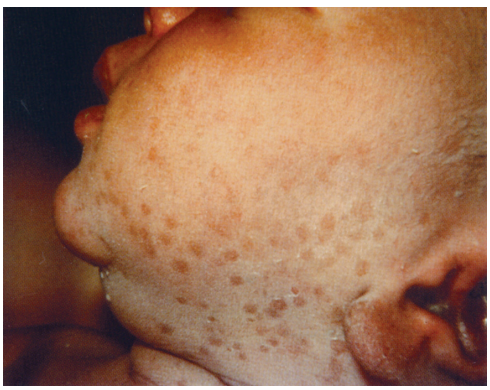

Figure 4.6 Multiple papules present in neonatal pustular melanosis

Source: From Clark DA. 2000. *Atlas of Neonatology.* Philadelphia, PA: Saunders, 261. Reprinted by permission.[19]

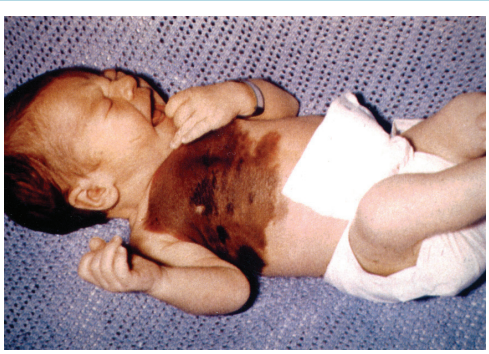

Figure 4.7 Congenital melanocytic nevi

Courtesy of Peter Honeyfield, MD, Presbyterian/St. Luke's Medical Center, Denver, CO.

Café au Lait Patch

A café au lait patch is a tan or light-brown, oval-shaped macule with a well-defined border (Figure 4.8). When less than 3 cm in length and less than three to five in number, they are of no pathologic significance. Up to 33% of normal children have one or more café au lait patches, with fair-skinned children often having multiple patches. The presence of six or more patches greater than 0.5 cm in length may indicate cutaneous neurofibromatosis, although they may occur in children without the disease.[12,20]

Neurofibromatosis (von Recklinghausen disease) is an autosomal dominant disorder in which tumors of various sizes form on peripheral nerves. Cranial nerves may also be affected. Ninety percent of patients with neurofibromatosis have café au lait patches.[20] Neurofibromas, small skin-colored nodules, may be present at birth or may not appear until adolescence in children with neurofibromatosis.

Tuberous Sclerosis

Tuberous sclerosis is a hereditary disorder characterized by cutaneous and central nervous system tumors resulting in seizures, developmental delays, and behavioral problems.[21,22] It presents with hypopigmented, white macules that may be thumb or leaf shaped. They are sometimes referred to as *ash leaf macules*. They may number up to 100 and are found anywhere on the newborn's skin, most often on the trunk and buttocks. Infants who present with unexplained seizures should be examined closely for these lesions. Most patients with tuberous sclerosis will have these macules at birth or soon afterward. Because of their light color, the lesions may not be readily apparent in fair-skinned infants. A Wood (ultraviolet) light may be helpful in illuminating white or hypopigmented lesions. Up to 5% of the population has one hypopigmented lesion.[13,21] If more than three are present, further evaluation is indicated.[21,22]

Skin Lesions Secondary to Trauma

Forcep Marks

Forcep marks may be seen on the cheeks, scalp, and face of newborns born following the use of forceps (Figure 4.9). The marks are generally red or bruised areas where the forceps were applied. The skin may also be abraded. The newborn should be examined for other complications of birth trauma, such as facial palsy, fractured clavicle, or skull fracture.

Subcutaneous Fat Necrosis

Subcutaneous fat necrosis consists of a subcutaneous nodule or nodules that are hard, nonpitting, and sharply circumscribed

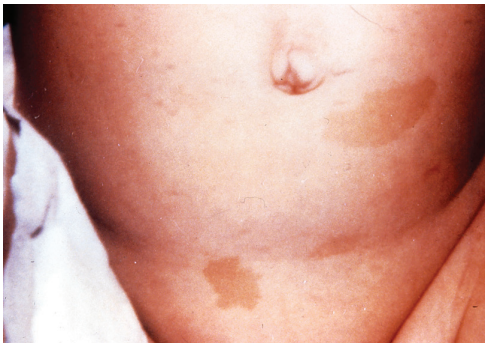

Figure 4.8 Café au lait patches

Courtesy of Eva Sujansky, MD, associate professor of pediatrics, University of Colorado Health Sciences Center , Aurora, CO.

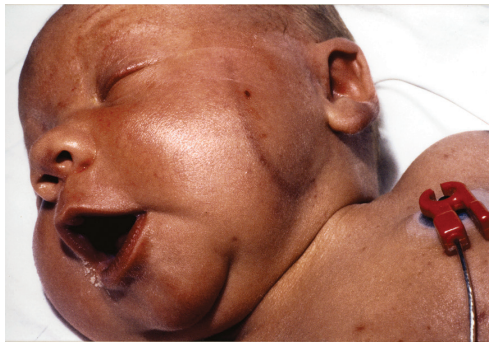

Figure 4.9 Forcep mark

Courtesy of Peter Honeyfield, MD, Presbyterian/St. Luke's Medical Center, Denver, CO.

(Figure 4.10). They may have a red or purplish color. They are more common in term newborns and are thought to be caused by trauma, cold, or asphyxia. It has been reported in some infants undergoing therapeutic hypothermia.[23,24] The nodules appear during the first weeks of life and are generally 1 to 2 cm in diameter. They may grow slightly larger over several days, and then resolve on their own over several weeks.[23,24] They may also be delayed or reoccur 2 to 3 weeks after resolution of the original nodule.[24] Occasionally, hypercalcemia may occur up to 6 months after presentation of the initial lesion. Serum calcium levels should be monitored, and intervention with fluids, calcium-wasting diuretics, and glucocorticoids may be necessary.[23,24]

Sucking Blister

A vesicle or bulla may appear on the lip, finger, or hand of a newborn as a result of vigorous sucking, either in utero or after birth. A sucking blister (Figure 4.11) may be intact or ruptured and requires no treatment.

Scalp Lesion

Scalp abrasions or lacerations may occur as a result of the insertion of a scalp electrode or trauma during delivery. Also, application of a suction cup for vacuum-assisted delivery may result in bruising or abrasions. Scalp pH measurements are used less often than in the past, but may result in lacerations to the fetal scalp. Treatment of lacerations or abrasions consists of keeping the area clean and dry and observing for secondary infections. Facial bruising may also be significant following delivery.

Vascular Skin Lesions

Nevus Simplex

The *nevus simplex*, also called *salmon patch* or *stork bite*, is the most common of the vascular birthmarks. It is seen in over 40% of newborns.[9] Nevus simplex is an irregularly bordered pink macule composed of dilated, distended capillaries (Figure 4.12). The lesion is most often found on the nape of the neck, the upper eyelid, the bridge of the

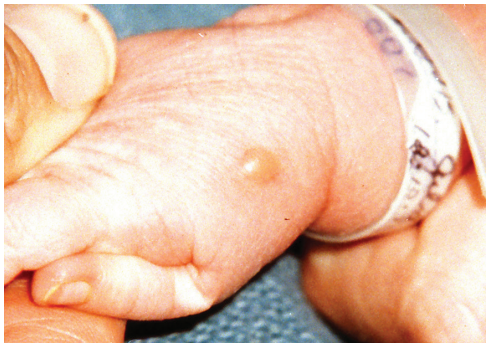

Figure 4.11 Sucking blister

Courtesy of Peter Honeyfield, MD, Presbyterian/St. Luke's Medical Center, Denver, CO.

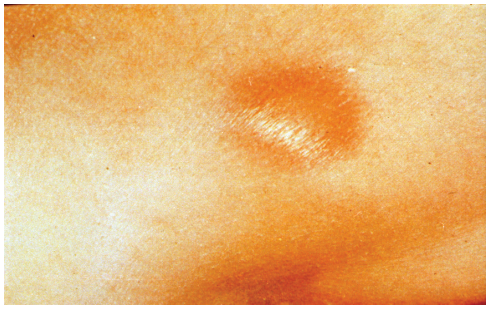

Figure 4.10 Subcutaneous fat necrosis

Courtesy of Barbara Quissell, MD, Presbyterian/St. Luke's Medical Center, Denver, CO.

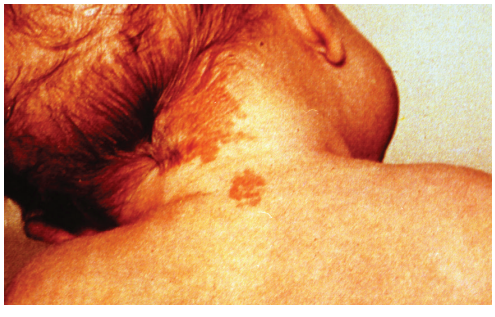

Figure 4.12 Nevus simplex

Courtesy of Barbara Quissell, MD, Presbyterian/St. Luke's Medical Center, Denver, CO.

nose, or the upper lip. Nevi simplex blanch with pressure and frequently become more prominent with crying. They generally fade by the second year of life, although those on the nape of the neck may persist.

Port Wine Nevus (Nevus Flammeus)

The port wine nevus is a flat pink or reddish-purple lesion consisting of dilated, congested capillaries directly beneath the epidermis. (In African American newborns, it is a jet-black color.) It has sharply delineated edges and does not blanch with pressure (Figure 4.13). The port wine nevus does not grow in size or spontaneously resolve. It may be small or may cover almost half of the body. It is usually unilateral, but may occasionally cross the midline. Unfortunately, the lesion most often occurs on the face, but it may appear on other parts of the body.[9]

Initially, the best treatment is to cover the lesion with a water-repellent cosmetic cream. Flashlamp-pulsed dye laser therapy can be successful in eliminating or reducing port wine nevi. Pulsed dye laser works by photocoagulation and destruction of the blood vessels in the lesion without damaging surrounding healthy skin. The treatment is more effective in infants and young children because the lesion tends to become darker and thicker with time.[25,26] Despite reports of good results in the newborn period, up to 10% of port wine stains have been noted to recur 7 months to 15 years after treatment.[25–27]

Most port wine nevi are isolated, but those that follow a pattern similar to the branches of the trigeminal nerve (the forehead and upper eyelid) may be associated with Sturge–Weber syndrome. This disorder causes a proliferation of endothelial cells, particularly in the small blood vessels. Intracerebral calcifications and atrophic changes may be present.[28] Children with Sturge–Weber syndrome may present with seizures, developmental delays, hemiparesis, and glaucoma. The cause of this syndrome is unknown. It does not appear to be hereditary.

Infantile Hemangioma

Formerly called a *strawberry hemangioma*, the superficial infantile hemangioma is a bright-red, raised, lobulated tumor that occurs on the head, neck, trunk, or extremities. It is soft and compressible, with sharply demarcated margins (Figure 4.14). The tumor may also occur in the throat (deep), causing airway obstruction.

Infantile hemangiomas occur in up to 10% of newborns and are more common in female and premature infants.[12,29] The lesion is caused by dilated capillaries, with associated endothelial proliferation in the dermal and subdermal layers. Most infantile hemangiomas have both superficial and deep components. Twenty percent to 50% are present at birth; the others are usually apparent by 6 months of age.[12,30] It is not uncommon for an infant to have more than one of these lesions.

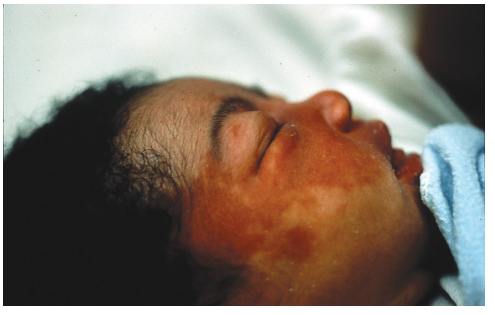

Figure 4.13 Port wine nevus

Source: From Clark DA. 2000. *Atlas of Neonatology.* Philadelphia, PA: Saunders, 267. Reprinted by permission.[19]

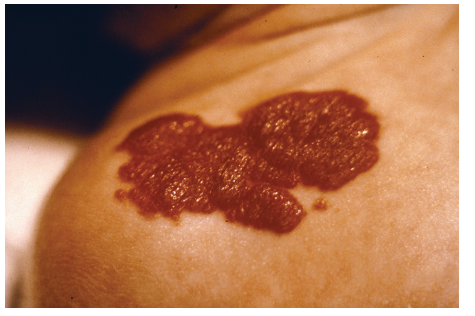

Figure 4.14 Infantile hemangioma

Source: From Clark DA. 2000. *Atlas of Neonatology.* Philadelphia, PA: Saunders, 265. Reprinted by permission.[19]

The infantile hemangioma grows rapidly for approximately 6 months and then gradually begins to regress spontaneously. Complete regression may take several years. Complications of infantile hemangioma include bleeding, ulceration, infection, or compression of vital organs or orifices. The preferred treatment is to allow natural spontaneous regression. If the lesion is interfering with vital functions (such as the airway or vision), treatment with systemic corticosteroids may be helpful. Propranolol has also been shown to successfully induce regression of infantile hemangiomas.[30] Pulsed dye lasers may reduce the extent of vascular proliferation and may increase healing in ulcerated hemangiomas or those likely to cause disfigurement.[29]

Cavernous Hemangioma

Cavernous hemangioma is similar to the infantile hemangioma, but consists of larger, more mature vascular elements lined with endothelial cells and involving the dermis and subcutaneous tissues. The overlying skin is bluish-red in color. On palpation, the cavernous hemangioma is soft and compressible, with poorly defined borders (Figure 4.15).

A cavernous hemangioma usually increases in size during the first 6 to 12 months of the infant's life, and then involutes spontaneously. Treatment is unnecessary unless the lesion is interfering with vital functions. In those cases, treatment with systemic corticosteroid therapy has been shown to cause some shrinkage.[8,31]

Two syndromes are related to cavernous hemangiomas. The first, Kasabach–Merritt syndrome, is a cavernous hemangioma associated with sequestration of platelets, thrombocytopenia, and consumption of fibrinogen and coagulation factors. Mortality can be as high as 20% to 30%, usually resulting from infection, severe hemorrhage, or iatrogenic complications.[28,31] It has been suggested that these tumors are not hemangiomas, but rather lymphatic malformations that have an underlying venous connection.[28,31] Treatment consists of supportive therapy, such as platelet transfusions and administration of corticosteroids. Angiogenesis inhibitors may restrain the growth of these tumors by inhibiting the growth of new blood vessels surrounding them.[31]

The second, Klippel–Trenaunay–Weber syndrome, consists of a vascular nevus with hypertrophy of the bone and soft structures of an extremity or other organs (Figure 4.16). The hypertrophy is probably the result of excess blood flow and malformed vessels.[31] This is a rare congenital anomaly, seen more frequently in males.[32] Prognosis depends on the severity of limb hypertrophy and other organ involvement.[32]

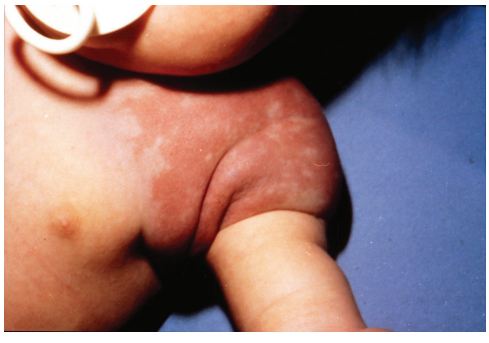

Figure 4.15 Cavernous hemangioma

Courtesy of Barbara Quissell, MD, Presbyterian/St. Luke's Medical Center, Denver, CO.

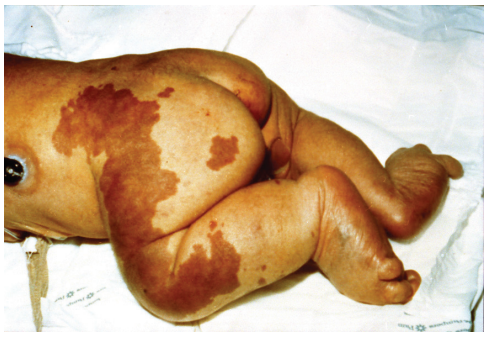

Figure 4.16 Klippel–Trenaunay–Weber syndrome

Courtesy of Peter Honeyfield, MD, Presbyterian/St. Luke's Medical Center, Denver, CO.

Infectious Lesions

Candida Oral Infection (Thrush)

A common infection in the newborn, thrush is an oral fungal infection caused by the organism *Candida albicans.* It appears as patches of white material scattered over the tongue and mucous membranes. The material is adherent and cannot be scraped off with a tongue depressor. Thrush is treated with an oral antifungal solution.[8]

Candida Diaper Dermatitis

Diaper dermatitis is common in newborns and generally needs no treatment other than frequent diaper changes and the diaper area kept clean and dry. Barrier ointments, such as petroleum or zinc oxide, help keep urine and feces away from the skin. Diaper dermatitis can be caused by *C. albicans.* This rash is a moist erythematous eruption with small white or yellow pustules. Small areas of skin erosion may also be seen. It is found primarily over the buttocks and perianal region, occasionally spreading to the thighs. Treatment consists of keeping the area clean and dry and applying an antifungal cream several times per day. If the rash is persistent or severe, an oral antifungal solution may be recommended to prevent reinfection resulting from Candida passing through the gastrointestinal tract.[33] Infants may present with both an oral Candida infection and Candida diaper dermatitis.

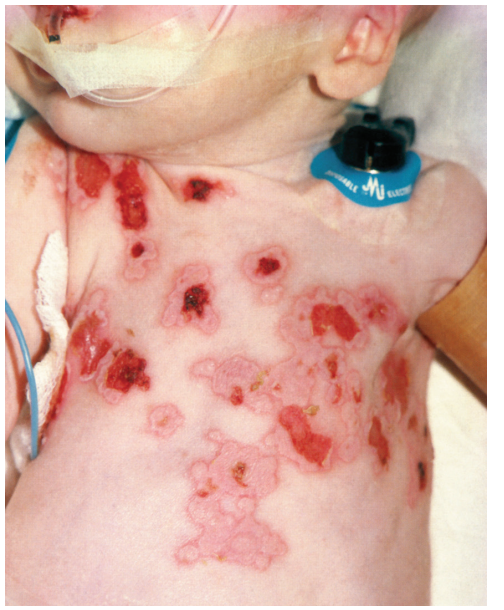

Figure 4.17 Blisters on trunk seen with neonatal herpes

Source: From Weston WL, Lane AT, and Morelli JG. 2007. *Color Textbook of Pediatric Dermatology*, 4th ed. Philadelphia, PA: Mosby, 130. Reprinted by permission.[36]

Herpes

Neonatal herpes is one of the most serious viral infections in the newborn, with a mortality rate of up to 40% in those newborns with disseminated disease.[34,35] The rash appears as vesicles or pustules on an erythematous base (Figure 4.17). Clusters of lesions are common. The lesions ulcerate and crust over rapidly. Fifty percent to 70% of newborns with neonatal herpes eventually develop this characteristic rash, but not always before they exhibit other clinical signs.[34,35] Absence of vesicles does not rule out the presence of the disease.[34,35] Treatment includes use of an intravenous antiviral agent such as acyclovir. Topical ophthalmic antiviral therapy should be used if there is ocular involvement.[35] Other signs, such as seizures, should be treated as they occur. Precautions for blood and body secretions must be observed.

Staphylococcal Scalded Skin Syndrome

Staphylococcal scalded skin syndrome begins with a generalized, tender erythema, followed by bullous eruption and peeling of the epidermis, often in large sheets (Figure 4.18). The eruption is caused by a *Staphylococcus aureus* infection, which produces a toxin damaging to epidermal cell walls.[37] Treatment includes systemic antistaphylococcal antibiotics and isolation to prevent spread of the infection. There may be extensive insensible water loss, so fluids and electrolytes should be closely monitored.

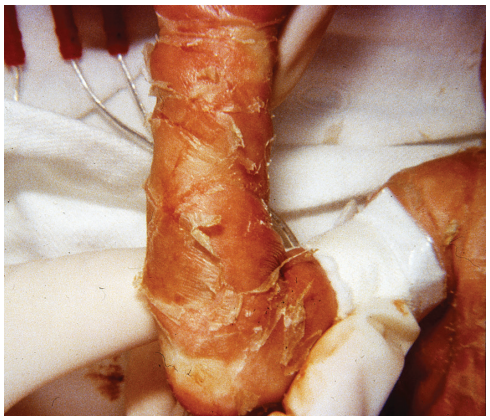

Figure 4.18 Scalded skin syndrome secondary to staphylococcal infection

Courtesy of Dr. David A. Clark, Albany Medical Center, Albany, NY, and Wyeth-Ayerst Laboratories. Philadelphia, PA. Reprinted by permission.

Other Dermatologic Congenital Viral Infections

Infections caused by cytomegalovirus, rubella virus, or enterovirus may present with dermatologic findings. Most often, these consist of jaundice and petechiae or purpura seen on the head, trunk, and extremities. Often described as "blueberry muffin" spots, these lesions are caused by thrombocytopenia and dermal erythropoiesis. One associated finding is hepatosplenomegaly. Treatment depends on the specific viral agent present.[34,35]

Miscellaneous Skin Lesions

Aplasia cutis congenita (*ACC*) is a congenital abnormality characterized by the absence of some or all layers of the skin (Figures 4.19 and 4.20). It most often appears as an ulceration or scarred area on the scalp (on the parietal bones or near the sagittal suture), but it can occur on other parts of the body.

ACC may be an isolated defect, or it can be associated with other anomalies, such as midline defects and chromosomal disorders (trisomy 13).[6] The cause is unclear, but ACC may result from vascular disruptions, midline developmental disruptions, trauma, or uterine or amnionic abnormalities (amniotic disruption sequence). Treatment consists of keeping the area clean and dry. Use of antibacterial dressings may be helpful. Large defects may require surgery (see also Chapter 5).

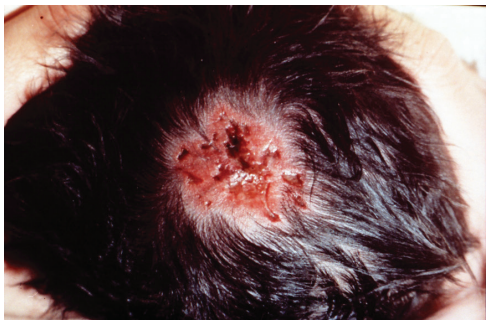

Figure 4.19 Aplasia cutis congenita

Courtesy of Peter Honeyfield, MD, Presbyterian/St. Luke's Medical Center, Denver, CO.

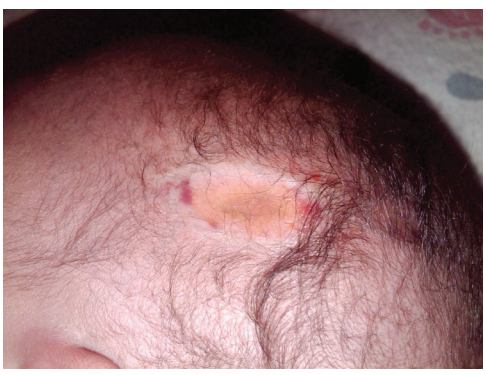

Figure 4.20 Aplasia cutis congenita

Courtesy of Mary Ellen Honeyfied, DNP, NNP, Sedalia, CO.

INSPECTING THE NAILS

The examination of the skin is not complete without close attention to the nails on the hands and feet. The nail consists of hard keratin. Damage to the nail matrix can be caused by trauma, inflammation, or genetic abnormalities. This damage can appear as pits, ridges, aplasia, or hypertrophy.[38] Most defects in the neonatal period are congenital rather than traumatic in origin.

Absence or Atrophy of Nails

Absence or atrophy of nails is seen in a number of congenital syndromes, including trisomy 13, trisomy 18, and Turner syndrome.[6,39] Inherited ectodermal dysplasias and skeletal anomalies may also be seen with absent or dystrophic nails.[38]

Hypertrophy of Nails

Hypertrophic nails are rarely seen in the newborn period. They may occur in diseases such as congenital hemihypertrophy or familial onychogryposis.

Abnormally Shaped Nails

Spoon- or racquet-shaped nails may occur as a result of a congenital or hereditary anomaly. They may be associated with anomalies of the hair or skin. Spoon-shaped nails may also be a temporary finding in an otherwise healthy newborn.[35]

SUMMARY

Careful assessment of the skin gives the examiner insight into the overall health of the newborn as well as any underlying pathology. Although numerous other anomalies may occur in newborn skin, the most common variations presented here should aid the healthcare professional in performing a complete and thorough newborn skin examination.

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Head, Eyes, Ears, Nose, Mouth, and Neck Assessment

Patricia J. Johnson, DNP, MPH, RN, NNP

5

Examination of the infant's head and neck requires visual inspection, obtaining measurements, palpation, use of an ophthalmoscope, brief auscultation, and, in rare cases, transillumination. After initial close observation, the examination should proceed from the top of the head down to the neck. Attention should be given to the infant's state, to facilitate optimal assessment with minimal discomfort. Portions of the examination may need to be postponed until the infant is in a more relaxed state. It is best to examine the eyes when the infant is in a quiet alert state and the oropharynx when the infant is crying.

Initial observations should include the infant's state; color of the skin and mucous membranes; size and symmetry of the head and face; and obvious deformations, malformations, or evidence of birth trauma. Minor anomalies of the head and neck are common. The initial inspection will provide important clues to possible congenital abnormalities as well as evidence of birth trauma and will often reveal important race-specific variations.

CRANIUM

Head Shape

The shape of the infant's head usually relates to molding of the skull during labor and delivery (Figure 5.1). An infant delivered by cesarean section will usually have a well-rounded head. The breech position may cause the head to be molded posteriorly into an egg shape, with a prominent occiput. Prolonged diagonal pressure may cause the head to appear "out of round," or asynclitic, when viewed from above. Distortion of the skull due to positional, external pressures in utero or during labor can be expected to self-correct. Parents can be reassured that molding usually resolves within a few days to a few weeks after birth.[1–4]

Skull

Inspection and careful palpation of the infant's skull are necessary to identify bones, sutures (fibrous joints), and fontanels (Figure 5.2). Sutures separate bones, and fontanels occur where two sutures meet. The metopic suture extends midline down the forehead between the two frontal bones and intersects with the coronal suture, which separates the frontal and parietal bones. The anterior fontanel (AF) is formed at the intersection of the metopic, coronal, and sagittal sutures. The size of the AF varies from 0.6 to 3.6 cm across, and in African American infants, it is commonly larger—from 1.4 to 4.7 cm.[5] Fontanels are measured diagonally from bone to bone rather than from suture to suture (Figure 5.3). Because there is a wide variation in fontanel size at birth, the measurement may serve only as a baseline for serial comparison. Individual measurements are not useful as a single examination finding, have no clinical significance, and have limited reproducibility. The AF is normally described as flat and soft if assessed with the infant in a quiet state and held in the sitting position. A tense or bulging fontanel may be a sign of increased intracranial pressure or may occur when the

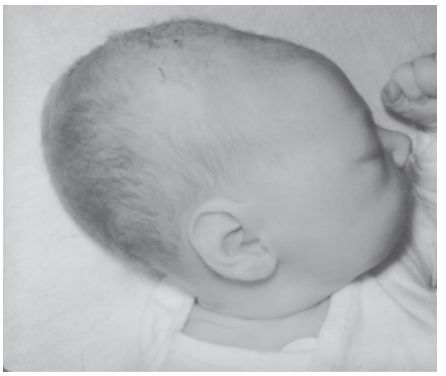

Figure 5.1 Head molding in the newborn infant

Courtesy of Dr. David A. Clark, Albany Medical Center, Albany, NY.

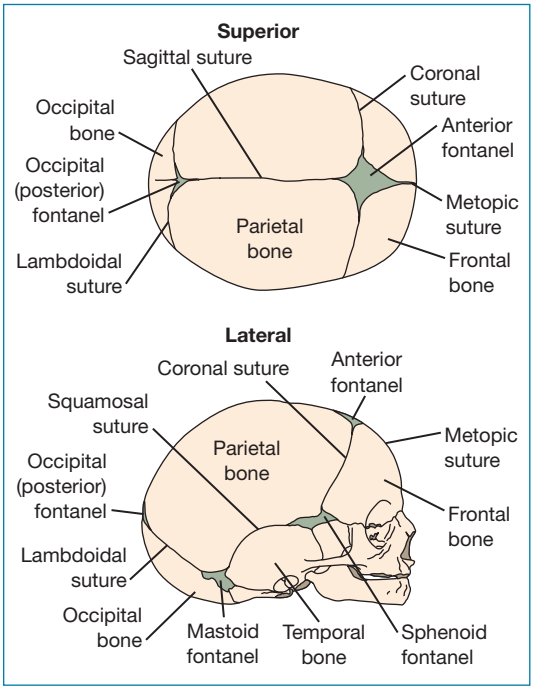

Figure 5.2 Sutures, fontanels, and bones of the neonatal skull

Source: Adapted from Scanlon JW, et al. 1979. *A System of Newborn Physical Examination.* Baltimore, MD: University Park Press, 47.[1]

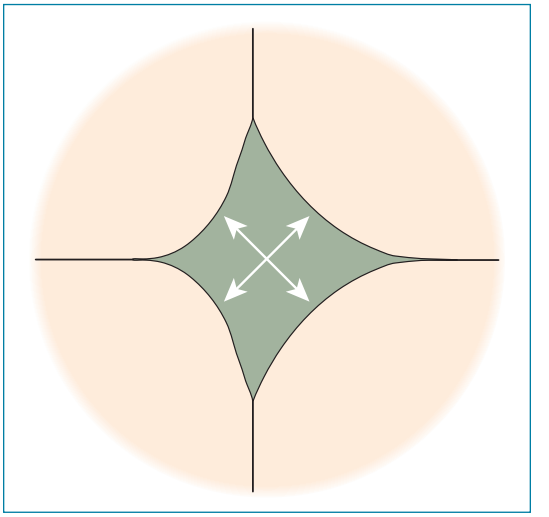

Figure 5.3 Measurement of the anterior fontanel: Bone to bone

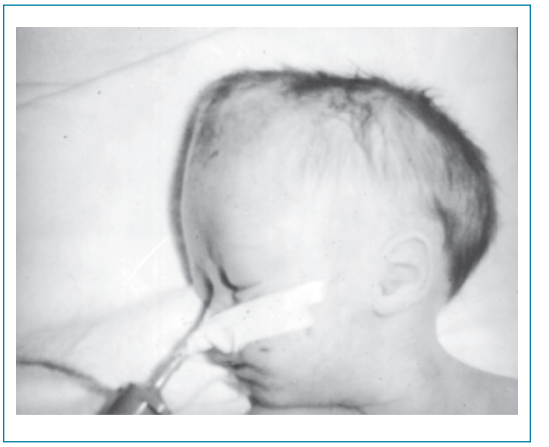

Figure 5.4 Bulging anterior fontanel

Courtesy of Dr. David A. Clark, Albany Medical Center, Albany, NY, and Wyeth-Ayerst Laboratories, Philadelphia, PA. Reprinted by permission.

infant is crying (Figure 5.4).[6,7] If the examiner is uncertain whether the fontanel is flat or bulging, it should be palpated with the infant in an upright, sitting position. A sunken fontanel is a sign of severe dehydration and is rarely seen in the newborn nursery. However, it may indicate excessive or acute decompression in an infant with a newly inserted external ventricular reservoir, drain, or ventricular peritoneal (VP) shunt for treatment of hydrocephalus. A very large AF can be associated with hypothyroidism.[8] The AF normally closes by 6 to 24 months of age.[1,2]

Auscultation over the fontanels and lateral skull bones is performed during examination of the head and neck. Auscultation

of the fontanels is specifically indicated in infants with multiple hemangiomas or heart failure.[5] In the otherwise normal infant, auscultation of a bruit (murmur-like sound) over the fontanels or the lateral skull can be a normal finding. Evidence of a bruit over the fontanel in an infant with suspected cardiac failure is associated with an intracranial arteriovenous malformation, which may be the cause of the congestive heart failure.[1,5,9]

The sagittal suture extends midline between the two parietal bones to the posterior fontanel (PF). This fontanel is formed where the sagittal suture meets the lambdoidal suture, which extends posterolaterally to separate the occipital from the parietal bones. The PF is usually small, 0.5 cm in diameter in Caucasian newborns and 0.7 cm in African American newborns, and closes by approximately 2 to 3 months of age.[1,2,5] If palpable, the PF should be soft and flat. A third "fontanel" may occur along the sagittal suture between the AF and PF. This is really a defect of the parietal bone and not a true fontanel. It can be a normal variant or associated with Down syndrome or congenital hypothyroidism.[5,10]

The squamosal suture extends above the ear to separate the temporal bone from the parietal bone. This suture and the sphenoid and mastoid fontanels are usually apparent only when there is increased intracranial pressure, as with severe hydrocephalus, but may be palpable in the premature infant with rapid brain growth.[1]

Mobility of sutures is assessed by placing the thumbs on the bones on either side of the suture and gently pressing down alternately with one thumb and then the other. Normal sutures can be described as approximated and mobile. Sutures may be split (separated) up to 1 cm. More widely split sutures may indicate increased intracranial pressure and require further investigation. With molding, the edge of the bone on one side of the suture will feel as if it is on top of the edge of the opposing bone; these sutures are described as overriding (Figure 5.5). It is common for the lambdoidal sutures to be overriding, with the parietal bone on top of the occipital due to molding after birth or with a minor decrease in hydration.

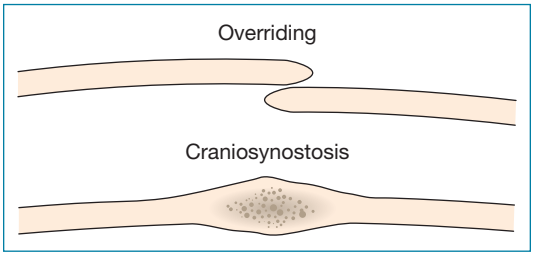

Figure 5.5 Cross-section of sutures

It is important to differentiate an overriding suture from one that feels ridged and immobile. The suture that peaks and is immobile implies premature fusion of the suture, or craniosynostosis.[1,11] Premature closure of a suture stops bone growth perpendicular to the suture, but allows continued parallel growth and compensatory expansion at the functional sutures, leading to abnormal head shape. Abnormal head shape resulting from craniosynostosis may be noted at birth, or the premature fusion may not be suspected until later in infancy (Figure 5.6).[12] Fused coronal sutures limit forward growth of the skull and lead to a broad skull (brachycephaly). Early closure of the sagittal suture limits lateral growth and results in a long, narrow head (scaphocephaly). Plagiocephaly is an asymmetric skull resulting from closure of the sutures on one side. The premature infant's head may develop the shape termed *dolichocephaly*, which is flattened side to side without craniosynostosis. Viewing the head from above may facilitate assessment of skull shape. Craniosynostosis may be primary (isolated), associated with a genetic syndrome, such as Apert or Crouzon, or the result of a metabolic disorder such as hyperthyroidism.[3,11,13] The incidence of isolated craniosynostosis in the United States is reported to be 0.4 to 1/1,000 live births.[11,13]

Palpation of the skull may reveal areas of soft or thinning bone, called *craniotabes.* Pressing on the bone elicits collapse with recoil of the underlying bone and a snapping sensation similar to pressing on a ping-pong ball. Craniotabes, an incidental finding evident upon palpation of parietal bones near the sagittal suture, is most often due to external pressure from prolonged vertex engagement

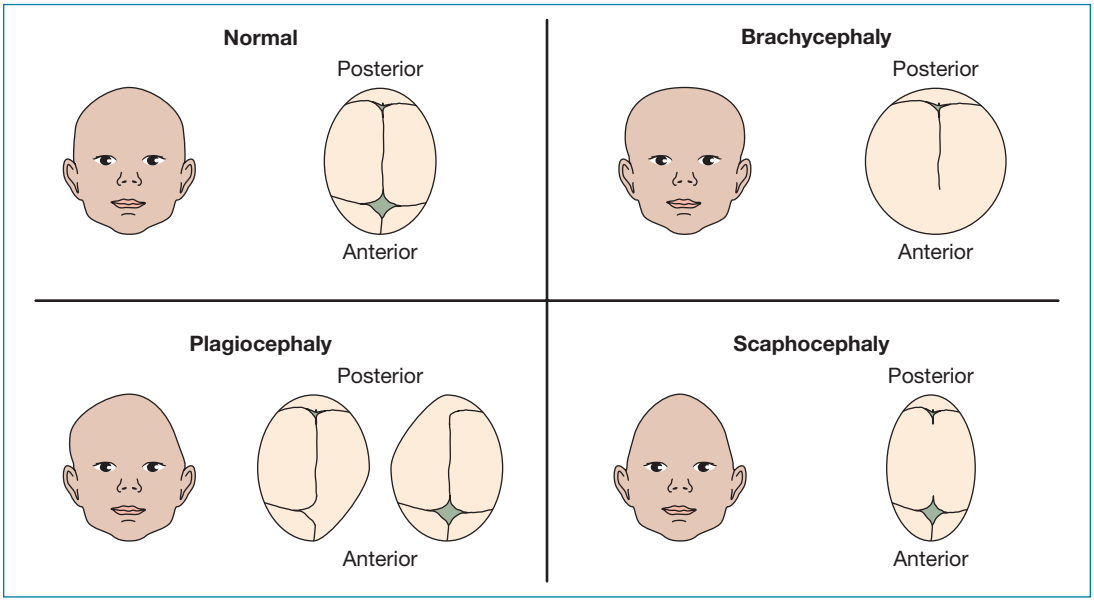

Figure 5.6 Normal skull and shapes that result from craniosynostosis

Source: From Disabato J, and Wulf J. 1989. Nursing strategies: Altered neurologic function. In *Family-Centered Nursing Care of Children*, Foster RL, Hunsberger MM, and Tackett JJ, eds. Philadelphia, PA: Saunders, 1731. Reprinted by permission.[12]

or pressure of the fetal head on the uterine fundus with breech position. It is also seen in up to 30% of normal newborns, especially in premature newborns, and is associated with pathologic conditions, including rickets, osteogenesis imperfecta, congenital syphilis, and vitamin D deficiency.[14,15] Craniotabes may also be caused by internal pressure on the fetal head from hydrocephalus. When due to external pressures and not a metabolic or underlying disease process, craniotabes can be expected to resolve in a few weeks. If caused by hydrocephalus, resolution will depend on the degree and persistence of internal pressure causing the characteristic bone thinning.[1,2]

While palpating the skull, the examiner also inspects the scalp and head for evidence of birth trauma and other abnormalities. The most common form of trauma to the head is caput succedaneum (caput). This is edema of the presenting part of the scalp caused by pressure that restricts the return of venous and lymph flow during labor and delivery. It can be accentuated by vacuum-assisted delivery. The edema pits on pressure, usually crosses suture lines, and has poorly defined edges. Caput is noted immediately after birth and resolves within a few days, which can help differentiate it from a cephalhematoma (Figure 5.7).[1,2,4]

A cephalhematoma results from the collection of blood between the periosteum and the skull (Figure 5.8). It may not be evident at birth because of an associated caput. A cephalhematoma has clearly demarcated edges confined by suture lines. With time, it may liquefy and become fluctuant on palpation; it may take weeks or months to resolve completely. The most common locations for a cephalhematoma are the parietal and occipital bones. Associated depressed skull fractures are very rare.[1,2,4,6]

A subgaleal hemorrhage is the third and potentially most serious lesion resulting from birth trauma. It is most common with instrumented vaginal delivery, especially vacuum-assisted delivery. However, it has been reported with cesarean births or any maneuver during delivery that produces a shearing force to the scalp resulting in tearing of the large emissary veins. With a subgaleal hemorrhage, there is bleeding into the galea aponeurotica or subaponeurotic

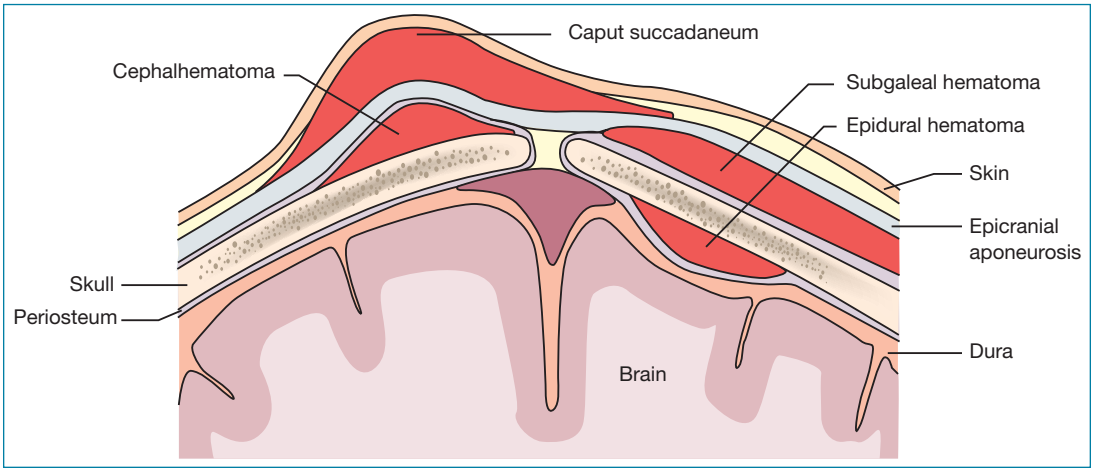

Figure 5.7 Areas of birth trauma

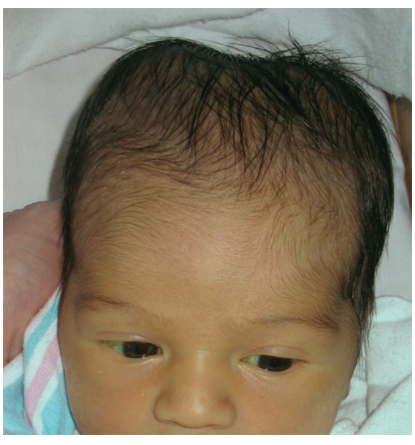

Figure 5.8 Bilateral cephalhematoma

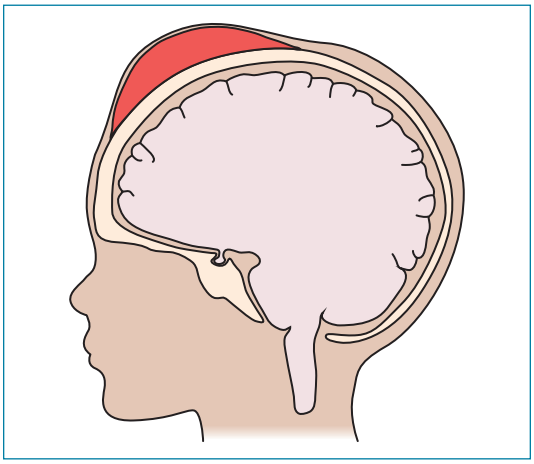

Figure 5.9 Subgaleal hemorrhage, sagittal view

space, which extends from the orbital ridges to the nape of the neck and laterally to the ears (Figure 5.9). This potential space produces a large compartment capable of containing the total blood volume of an infant, and therefore a subgaleal hemorrhage has a 5% to 22% mortality rate if blood loss is extensive and undiagnosed.[16] It presents with generalized scalp edema, usually with ecchymosis, and often with bilateral or unilateral periorbital and periauricular edema. The ballotable fluid mass crosses the sutures and can be manually repositioned from the eyebrows to the nape of the neck, differentiating it from a large caput. If progressive, it can result in severe anemia, hypotension, and death unless diagnosed early. Effective treatment may involve volume resuscitation, blood replacement, and treatment of presenting clotting abnormalities.

Trauma to the scalp may include puncture from a scalp electrode; lacerations from fetal blood sampling or uterine incision; and bruises, abrasions, or subcutaneous

fat necrosis from an instrument (forceps or vacuum) delivery.[2,6,17] The lesion should be described by its appearance, size, and location near sutures, fontanelles, or underlying bones.

Aplasia cutis congenita (see Figure 4.19) is an uncommon scalp lesion usually found on the parietal bones or near the juncture of the sagittal and lambdoidal sutures. This lesion is a hairless, circumscribed area of 1 cm or more, and its surface is often shiny, cicatricial, and flat or keloid in appearance. Occasional blistering or ulceration is evident with serosanguineous exudate or fresh granulation. In most infants, the defect is an isolated finding that resolves with residual scar formation and absence of hair growth. There is a risk for underlying defect and associated major defects (e.g., trisomy 13) in some infants with this lesion, necessitating careful evaluation of the infant for major abnormalities.[18] Other skin lesions are described in Chapter 4.

Malformation of the skull associated with incomplete neural tube closure results in an encephalocele. Central nervous system tissue can protrude from a defect anywhere on the skull, most commonly midline and in the occipital area.[3]

Scalp Hair

Examination of the scalp should include assessment of the hair for quantity, texture, distribution, and hair whorls. Low hairline, increased quantity of hair, and brittleness may be associated with congenital anomalies. The slope of each hair follicle appears to be associated with the stretch of the skin during periods of rapid brain growth, particularly from weeks 10 to 16 of gestation.[8,19] One or two hair whorls can be normal. An abnormally placed whorl, absence of hair whorl, or unusual hair growth may be associated with aberrant brain growth and mental retardation.[8] Hair consistency, color, and growth patterns commonly reflect developmental, familial, and racial variations with no clinical significance.[19] Unusual hair growth patterns or presentation should be carefully described.

Alopecia is an abnormal deficiency of hair that is diffuse or focal. Diffuse alopecia is more commonly due to a genetic anomaly in the hair follicles or is syndromic. Focal alopecia is often traumatic or associated with underlying scalp lesions. Hirsutism, or excess hair growth, may be genetic, syndromic, metabolic, drug-induced, or an isolated finding.

Head Size

Measurement of the occipital–frontal circumference (OFC) is fundamental for adequate assessment of head size. Taking this measurement may be unsettling for the infant and can be deferred to the end of the exam. As such, the inspection and palpation of the head would be completed prior to measuring the OFC. A nonstretchable paper tape measure should be used to obtain three measurements of the OFC; the largest of these should be recorded. Accuracy of this measurement depends on encircling the head at the widest occiput prominence and anteriorly 1 to 2 cm above the glabella space at the largest frontal prominence. The average OFC at 40 weeks gestational age is 35 cm. The OFC ranges from 33 to 37 cm between the 10th and 90th percentile.[20] Before closure of the fontanels, the measurement of the OFC is an indirect measure of intracranial contents, including the brain, cerebrospinal fluid, cerebral blood volume, and bone.[21] The OFC is often misleading immediately after birth due to cranial molding, scalp edema, or hemorrhage under the periosteum. Subsequent measurement(s) should be obtained several days after birth once the initial distortions resulting from the birth process resolve. The OFC should be plotted on a standard growth chart and the gestation-specific percentile in which the measurement falls noted. The percentile of the OFC, along with those for weight and length for gestational age, are necessary to diagnose symmetric versus asymmetric growth restriction and micro- or macrocephaly.[1,20]

Microcephaly is defined as an OFC less than 2 standard deviations (*SD*) below the mean for gestational age or less than the 3rd

percentile.[21] Microcephaly is usually associated with microencephaly and is caused by reduced brain growth.[21,22] With microcephaly, the sutures often become prematurely fused because the expansive force of brain growth that enlarges the cranial vault is lacking. Microcephaly can be an isolated finding, or it may be associated with a genetic syndrome or congenital infection.[1–3,22] As with the other growth parameters, OFC in the infant born prematurely tends to be lower than expected for gestational age. Discrepancies may be due to inaccurate dating or to biologic differences and/or maternal or neonatal pathology that results in growth restriction, which occurs more frequently than growth acceleration.[23] In addition, gender and racial variations have been reported. Females tend to have smaller head circumferences than males in most ethnic groups, and African American infants as well as most non-White groups have smaller head circumferences than White infants at the same gestation. These growth variations indicate the need for gender- and race-specific population growth references, which are currently not available, to minimize the potential of mislabeling infants as small for gestational age.[24–26]

Macrocephaly is diagnosed when the OFC is above the 90th percentile despite appropriate weight and length for gestational age. Macrocephaly may be familial, caused by hydrocephalus, or associated with dwarfism or osteogenesis imperfecta.[1,27] Familial macrocephaly is more often macroencephaly or large brain volume without hydrocephalus. Confirmation requires obtaining measurements of the parents' heads and plotting them on a Weaver curve.[28]

Transillumination of the skull is not part of the routine newborn physical exam, but it may be helpful when the infant's head has an unusual shape or size, or the neurologic examination is abnormal. A transilluminator or a flashlight with a rubber cuff may be placed flat against the infant's head in a dark room (see Figure 1.3). A ring of light more than 2 cm larger than the light source implies increased fluid or decreased brain tissue in the cranium. A false-positive transillumination may occur with a large caput because the scalp edema will transmit a halo of light. More definitive studies are necessary for diagnosis.[1,2] If the infant presents with a high level of suspicion for hydrocephalus, transillumination is often replaced by one of the more definitive studies, such as cranial ultrasound, CT, or MRI.

FACE

Examination of the face should begin with observation of the relationships among all the facial components: ears, eyes, nose, and mouth. The forehead of a newborn takes up the upper half of the face, reflecting the large cranial volume needed for rapid brain growth. In childhood, the growth of the mid- and lower face exceeds that of the upper face, eventually resulting in the face and skull shape of the adult.[3]

The shape and symmetry of the face, as well as evidence of trauma, should be noted. Face or brow presentation or the presence of a nuchal cord at delivery may cause facial bruising, petechiae, and progressive edema. Unusual flattening of facial features may occur as a result of prolonged intrauterine compression from oligohydramnios. Forceps application can cause bruises, abrasions, or subcutaneous fat necrosis.[2] The location and extent of any trauma should be carefully described. Other skin lesions that may be seen on the face are described in Chapter 4.

Many malformation syndromes have very distinctive facial features. Unusual facial characteristics may serve as aids in the diagnosis of a particular syndrome or may be familial.[8] Some syndromic facial features may not be evident in the prematurely born infant. Phenotypic presentation may be progressively evident when the infant's corrected gestational age (CGA) nears 40 weeks. Some features present later in infancy or childhood.

Facial movements during crying are assessed for symmetry. Damage to the facial nerve (seventh cranial nerve) prior to or during delivery can result in paralysis of the affected side of the face. Extensive damage can involve the entire side of the face

innervated by the damaged nerve and cause drooping of the muscles on the affected side of the face. More common, the damage involves only the lower branch (mandibular branch) of the facial nerve, which controls the muscles around the lips. Evidence of this damage presents as a characteristic decreased movement on the affected side of the face when the infant cries, because the weakened facial muscles of the affected side allow the mouth to be pulled to the unaffected side ("drooping mouth" appearance; Figure 5.10). These infants often have loss of forehead wrinkling and nasolabial folds and partial closing of the eye on the affected side, differentiating them from infants with congenital asymmetric crying facies (ACF). If the nerve damage is caused by pressure, then the facial effects resolve within hours to weeks after birth. Congenital facial palsy is often spontaneous, but is also associated with difficult or instrumented delivery requiring forceps. A persistent palsy may be an indication of an underlying, more central abnormality. ACF, for example, is a congenital absence or hypoplasia of the depressor anguli oris muscle (DAOM) on one side. The DAOM controls frowning, with resulting asymmetry of the face with crying. There is an association with other anomalies, and further evaluation is warranted, especially for cardiovascular abnormalities.[7] (Chapter 11 provides guidelines for a complete neurologic assessment.)

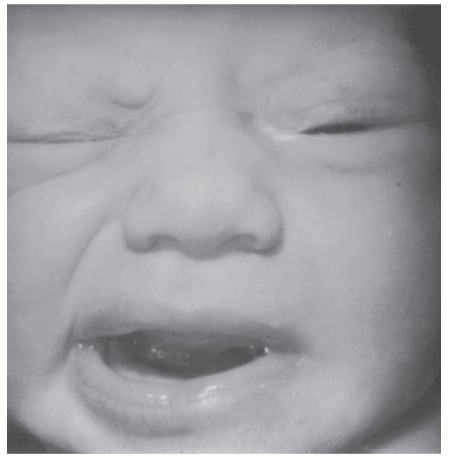

Figure 5.10 Facial asymmetry

Courtesy of Dr. David A. Clark, Albany Medical Center, Albany, NY, and Wyeth-Ayerst Laboratories. Philadelphia, PA. Reprinted by permission.

Ears

Abnormal formation or placement of the ears can be associated with chromosomal anomalies and syndromes; however, a wide number of minor structural variations fall within the normal range. Ear anomalies are usually nonspecific and supportive of a diagnosis rather than presence of the anomaly being diagnostic in and of itself.[1,2,4,8] To describe abnormalities, the examiner should be familiar with the normal anatomy and descriptive nomenclature of the ear (Figure 5.11).[4] In a term infant, the pinna should be well formed, with cartilage that recoils easily after folding (Chapter 3). Temporary asymmetry of the ears from unequal intrauterine pressure on the sides of the head is common.[1] Especially hairy ears, involving both the pinna and lobes, are familial, syndromic, or associated with infants of mothers with poorly controlled diabetes mellitus.

Minor malformations, such as pits and skin tags, may be familial or associated with other anomalies (Figure 5.12).[4,8] These minor malformations are usually located anterior to the tragus and are thought to be embryologic remnants of the first branchial cleft or arch. A preauricular sinus may be blind, or it may communicate with the internal ear or brain. Chronic infection could necessitate surgical removal of the entire sinus tract. Darwinian tubercle is a normal variant, appearing as a small nodule on the upper helix. A very poorly formed external ear should alert the examiner to a possible chromosomal anomaly or syndrome.[4,8]

The position of the external ear on the head can be assessed by extending a line from the inner to the outer canthus of the eye toward the ear (Figure 5.13). If the insertion of the ear falls below this line, it is low set. It is important to assess both ears because one may be posteriorly rotated and give the appearance of being low set.[4,6,8]

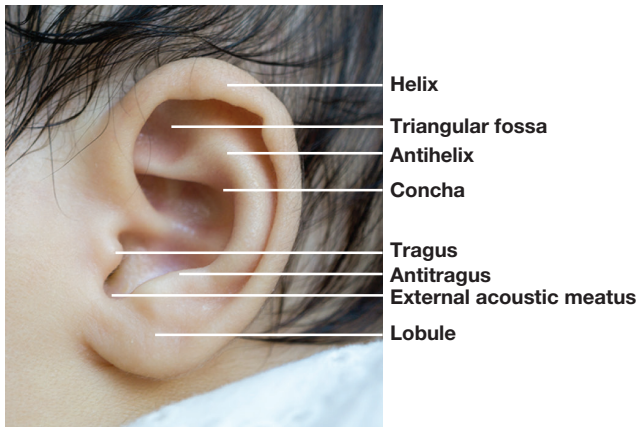

Figure 5.11 Normal external newborn ear (pinna)

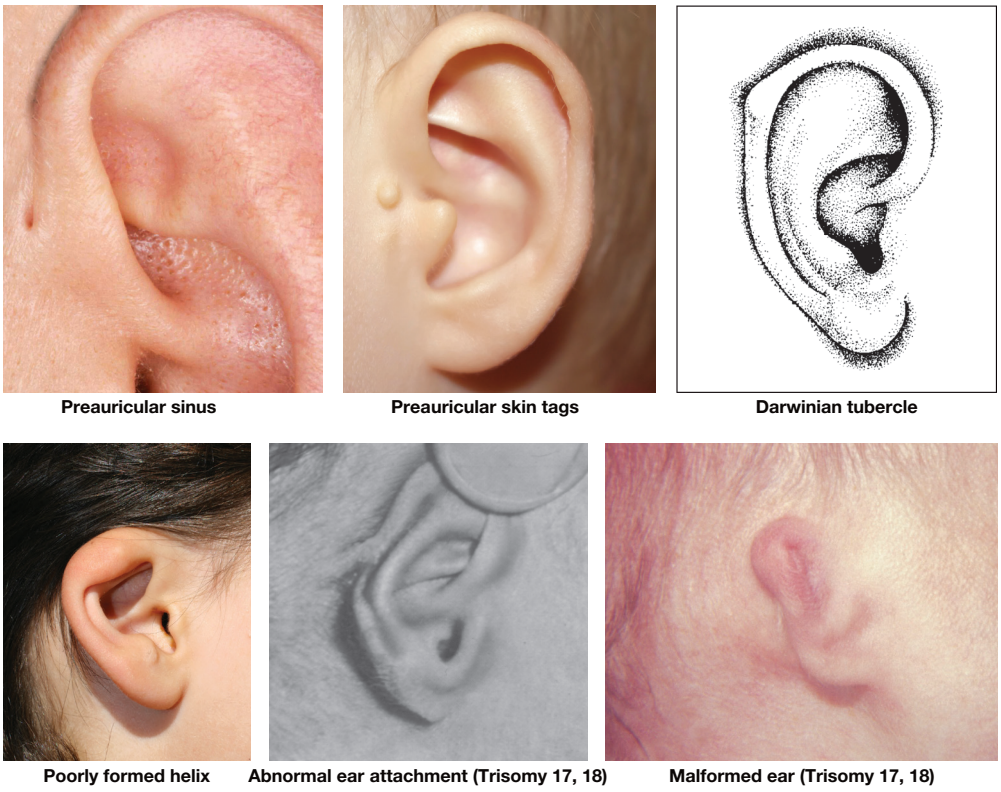

Figure 5.12 Variations and minor malformations of the ear

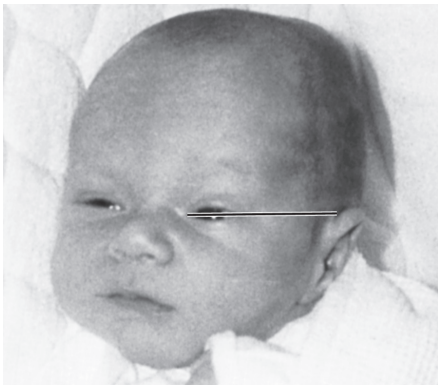

Figure 5.13 The normal ear position

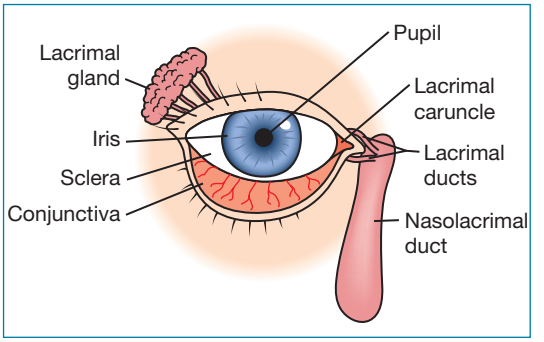

Figure 5.14 Anatomy of the eye

The ear should be inspected visually to assess the presence and patency of the auditory canal.[1,2,6] The newborn infant's external auditory canal is short and may contain vernix, intrapartum blood, or meconium debris, making otoscopic examination difficult. Otoscopic examination is not part of the routine newborn examination; it can be done later at the first well-baby visit.

Significant hearing loss is one of the most common major abnormalities present at birth.[29] Assessment of an infant's hearing during an examination, however, can be difficult. The infant should startle, cry, or stiffen at the sound of a loud noise or alert to the sound of a voice, but may be responding to air movement rather than noise and can quickly habituate to a repeated stimulus.[1,2] Any infant with a small or abnormally developed ear is at increased risk for hearing loss in that ear. Current recommendations endorse universal hearing screening of all newborns, with a goal of appropriate intervention by 6 months of age.[29]

Eyes

Eyes are usually a major area of interest when parents are viewing their newborn infant. It is important for the examiner to be familiar with the normal anatomy and descriptive nomenclature of the eyes (Figure 5.14). Parents will need reassurance about common eye trauma, such as bruises or edema of the eyelids and hemorrhages seen around the iris that can occur with a normal vaginal delivery. Conjunctival or subconjunctival hemorrhage results from rupture of a capillary in the mucous membrane that lines the eyelids and is reflected onto the eyeball (conjunctiva). It is seen as a bright-red area on the white part of the eye (sclera) near the iris and usually resolves within 1 week to 10 days without residual effects. Inflammation of the conjunctiva can be caused by debris from delivery or prophylaxis of the eyes with erythromycin ointment.[1] Malformation of the eyelids is uncommon, but coloboma (an absence or defect of some ocular tissue, including the eyelid) may be seen. Eyelid eversion with marked chemosis (swelling) of the palpebral conjunctiva is a rare, usually bilateral abnormality seen more commonly in African American newborns at birth and may be seen with Down syndrome. This abnormality requires involvement of ophthalmology, but is often resolved without surgery.[30] Ptosis, a paralytic drooping of an eyelid when the lids are fully open, may also be seen. Nevus simplex is a common vascular birthmark seen on the eyelids and glabella (area above the nose and between the eyebrows).[4] (See Chapter 4 and Figure 4.12.)

Tear formation does not usually begin until 2 to 3 months of age. The nasolacrimal duct is not fully patent until 5 to 7 months of age, and purulent or mucoid eye drainage is common. Unless accompanied by conjunctival inflammation with redness or swelling, this drainage can generally be treated by

lacrimal massage and gentle cleansing with water and a cotton ball.[1,2,6] A rare obstruction of the nasolacrimal duct will develop cysts caused by distention of the lacrimal sac and duct. This is referred to as *dacryocystocele* and presents with a bluish bulge below the medial canthus. These infants require ophthalmology involvement due to risk of infection and recurrance but may respond to palpation of the lacrimal sac with mucoid discharge.[31]

Abnormal placement of the eyes or small palpebral fissures (eye openings) can alert the examiner to the presence of a syndrome or chromosomal anomaly. The distance between the outer canthi can be divided into equal thirds, with one normal palpebral fissure length fitting into the inner canthal distance (Figure 5.15).[32] Hypertelorism exists if the eyes are more widely spaced; hypotelorism is present if the eyes are more closely spaced. Small palpebral fissures can give the appearance of hypertelorism.[1,2,8,33]

If the outer canthus of the eye is higher than the inner canthus, the eye is said to be upslanting; if the outer canthus is lower than the inner canthus, the eye is downslanting. The epicanthal fold is a vertical fold of skin on either side of the nose that covers the lacrimal caruncle and, although a characteristic finding in Asians, occurs in 20% of non-Asians, usually disappearing by 10 years of age as the growth of the nasal bridge catches up to that of the medial canthal skin.[34] Epicanthal folds with upslanting palpebral fissures are common in infants of Asian descent. They may suggest Down syndrome in other ethnic groups, especially if associated with hypertelorism.[1,8]

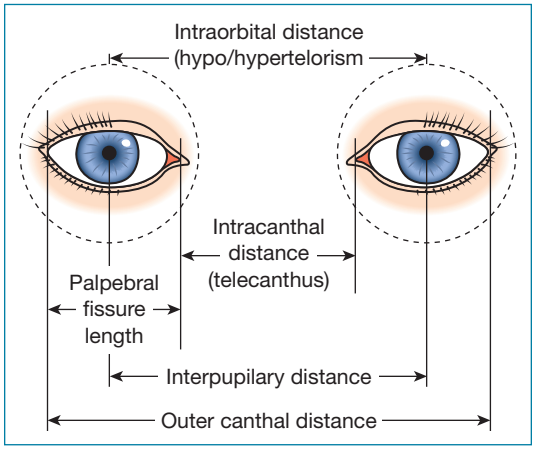

Figure 5.15 Eye measurements

Source: Adapted from Cheng KP, and Biglan AW. 2007. Ophthalmology. In *Atlas of Pediatric Physical Diagnosis*, 5th ed., Zitelli BJ, and Davis HW, eds. Philadelphia, PA: Mosby, 727. Reprinted by permission.[32]

An eyebrow normally extends above the eye in a curve approximately the length of the palpebral fissure. Eyebrows that meet at the glabella and abnormally long or tangled eyelashes are associated with some syndromes, such as Cornelia de Lange.[8] Examination of the eyes is much easier if the infant opens them spontaneously. At the beginning of the examination, the examiner can give an auditory stimulus, change the infant's position from supine to upright, gently swing the infant in an arc on the examiner's arm, or dim the lights. If the eyes are forcefully opened, the eyelids will often evert, making visualization impossible. The eye exam will be more difficult if attempted within several hours after eye prophylaxis.

A full ophthalmoscopic exam is not practical, so the examiner must be alert to obtain the information needed in a limited time.[2,6] The ophthalmoscope should first be adjusted for focus and then for supply of a small, round, white beam of light. The examiner directs the light at the infant's pupils from a distance of about 6 in. to assess equality of pupil size, and pupillary reflex (constriction with bright light). The notation "PERL" can be used if "pupils are noted to be of equal size and react to light" by constricting. The examiner also seeks to elicit the retinal reflex (red reflex). When a bright light is directed at the newborn's lens, a clear red, orange, or yellowish color is reflected from the retina back to the examiner. Opacity of the lens or cornea interrupts the reflection (leukokoria or white retinal reflex); lack of the red reflex could imply congenital cataract, retinoblastoma, or glaucoma (Figure 5.16).[1,2,35] In dark-skinned infants, the red reflex pales from orange toward pale yellow to gray in the darkest pigmented infants. The lens vessels of the premature infant can be examined to help determine gestational age

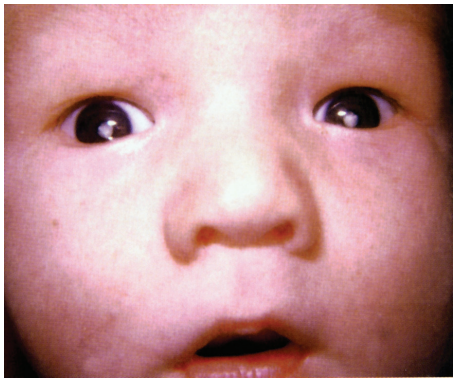

Figure 5.16 Congenital cataracts

Source: From Clark DA. 2000. *Atlas of Neonatology.* Philadelphia, PA: Saunders, 280. Reprinted by permission.[36]

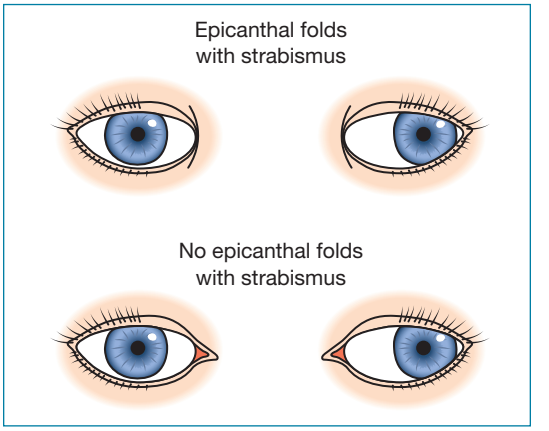

Figure 5.17 Strabismus

Source: Adapted from Alexander MM, and Brown MS. 1979. *Pediatric History Taking and Physical Diagnosis for Nurses*, 2nd ed. Philadelphia, PA: Mosby, 97. Reprinted by permission of the authors.[37]

(Chapter 3). A keyhole-shaped pupil, also known as coloboma of the iris, may be associated with other anomalies, or it can be an isolated finding.[1,2,6]

The iris of a newborn infant is generally dark gray, blue, or brown at birth and will acquire final pigment color at about 6 months of age. Brushfield spots are white specks scattered linearly around the entire circumference of the iris. They are associated with Down syndrome, but may be seen as a normal variant.[8] The sclera of a term infant is generally white to bluish white. A blue sclera is associated with osteogenesis imperfecta. The sclera may become jaundiced with hyperbilirubinemia. In a normal gaze, no sclera should be visible above the iris. The "sunset sign" is often seen in infants with hydrocephalus, where there is lid retraction and a downward gaze.[1,2,6]

Observation of eyeball movements during neurologic assessment is discussed in detail in Chapter 11. Nystagmus is a rapid, searching movement of the eyeballs. Limited horizontal nystagmus may be elicited with rotational eye movement, but disappears by 3 to 4 months of age. Spontaneous horizontal, vertical, or torsional nystagmus and persistent nystagmus are always aberrant and likely associated with visual and/or neurologic abnormalities. Strabismus results from muscular incoordination and gives the appearance of crossed eyes (Figure 5.17). Pseudostrabismus from a flat nasal bridge or epicanthal folds usually resolves by 1 year of age and is differentiated from strabismus by the presence of symmetric corneal light reflexes. Unusual protrusion (exophthalmos, proptosis), enlargement or bulging of the eyeball is associated with hyperthyroidism, tumors, hemangiomas, and congenital glaucoma and can be associated with damage to the eye with hemorrhage into its orbit.[1,4,33,38]

Nose

The size and shape of the nose may be racially characteristic and familial, but the infant's nose is generally smaller and flatter than an adult's. The nose should be symmetric and placed vertically in the midline. Nasal flaring is abnormal and is a sign of respiratory distress. Sneezing is common and normal unless excessive or continuous. Nasal stuffiness can be normal in the newborn period; however, chronic breathing difficulty or chronic nasal discharge is abnormal.[1,2,6,39] A very low nasal bridge with a broad base may be associated with Down syndrome.[2,8]

Deviation of the nasal septum to one side may be a deformation from position in utero, or it may be caused by a dislocated septum. If the septum will not easily straighten and the nares remain asymmetric

when the tip of the nose is pushed to midline, the septum is dislocated and will require treatment (Figure 5.18).[1,40] The examiner can elevate the tip of the nose slightly to view the nasal septum, the floor of the nose, and the turbinates.[40]

Nasal patency can be assessed by watching the infant breathe in a quiet state. Because infants are obligate nose breathers, bilateral choanal atresia (obstruction of the posterior nasal passages) will cause the infant to be cyanotic at rest and pink when crying and breathing through the mouth. If obstruction is suspected, a soft #5 French catheter can be gently passed through each nostril to assess patency. If an infant has noisy nasal respirations, a piece of cold metal can be held under the nose to observe for mist formation under both nares. If the turbinates are swollen from previous suctioning, passing a catheter to assess patency may only make the edema worse.[1,2]

Nasal occlusion can also be acquired from accumulation of secretions resulting in noisy respirations, feeding difficulties, respiratory distress, or apnea. Excessive bulb suctioning may aggravate the occlusive tendency; normal saline irrigation or intermittent installation of normal saline drops may facilitate resolution of obstruction from secretions.

With increased use of nasal continuous positive airway pressure (NCPAP) and nasal intermittent positive pressure ventilation (NIPPV) in premature infants and infants with respiratory distress, erosive trauma to the nasal labia, nasi columella, and nares is more common. Preventative and protective skin coverings can help, but monitoring the area for ulceration will minimize long-term disfigurement and scarring.

Major abnormalities of the nose, including clefts, single nares, masses, and partial or complete hypoplasia, are usually syndromic or associated with major central nervous system anomalies.

Mouth

The lips and mucous membranes of a healthy term infant should be pink. Mild circumoral cyanosis is normal during transition and with crying during the first few days.

Abnormal shape and size of the oral opening, lips, philtrum (midline groove between the nose and upper lip), and mandible may be associated with a syndrome. A small oral opening, known as microstomia, may be seen in some syndromes.[8] Macrostomia is seen with storage diseases such as the mucopolysaccharidoses. Lip thickness is related to racial and familial characteristics, and many

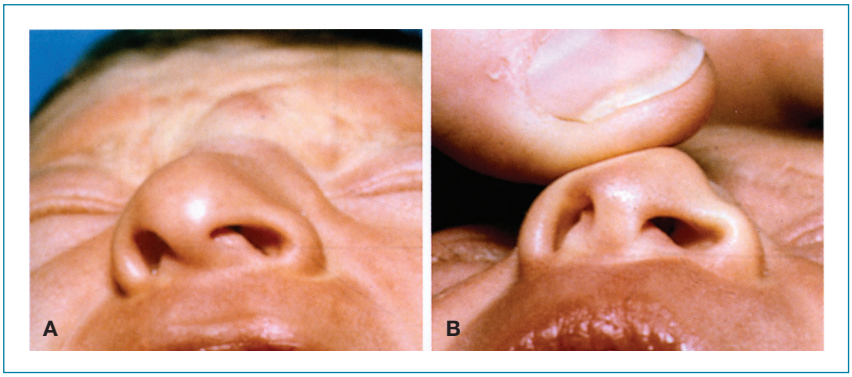

Figure 5.18 Nasal deformity. This infant incurred dislocation of the triangular cartilage of the nasal septum during delivery. Inspection of the nose reveals deviation of the septum to the right and asymmetry of the nares **(A)**. When the septum is manually moved toward the midline, the asymmetry persists, confirming the dislocation **(B)**.

Source: From Brozanski BS, Riley MM, and Bogen DL. 2012. Neonatology. In *Atlas of Pediatric Physical Diagnosis*, 6th ed., Zitelli BJ, McIntire SC, and Norwalk AJ, eds. Philadelphia: Mosby, 57. Reprinted by permission.[17]

infants have calluses on the lips from sucking in utero. Unusually thick or thin lips are abnormal and require further evaluation for trauma or an underlying mass. A thin upper lip with a smooth philtrum and short palpebral fissures may be seen in fetal alcohol syndrome. Cleft lip may be unilateral or bilateral and may be small or extend to the floor of the nose. Micrognathia, or abnormally small lower jaw, is seen in Pierre Robin sequence and some syndromes (Figure 5.19).[1,2,8,13]

During the examination, assess the cry for quality, strength, pitch, and hoarseness or stridor. Inspect the symmetry of facial movements during the cry, and assess rooting and sucking reflexes (Chapter 11).

Visual examination inside the mouth is easiest when the infant is crying. Use of a tongue depressor may stimulate a strong protrusion reflex, making assessment difficult. The mouth may be opened by gently pressing down on the chin. Abnormalities of the oropharynx are unusual, but a quick look is necessary to ensure absence of a structural anomaly or tumor. If possible, the uvula should be visualized at the back of the soft palate. A bifid uvula is often associated with submucosal cleft, notched hard palate, and muscular diastasis of the soft palate. It may be associated with other congenital anomalies.[2,8,41]

The tongue should fit well into the floor of the mouth and appear symmetric. A large tongue (macroglossia) impedes closure of the mouth and is associated with Beckwith–Wiedemann syndrome, hypothyroidism, and mucopolysaccharidosis. The tongue may *appear* large when the lower jaw is small (micrognathia). This is seen in Pierre Robin sequence, where there is hypoplasia of the mandible, the tongue is placed further back than normal (glossoptosis), and the palate is cleft.[8]

The frenulum (frenulum linguae) attaches the underside of the tongue to the floor of the mouth, usually midway between the tongue's ventral surface and tip. A very thick or prominent frenulum, or "tongue tie," is ankyloglossia. The incidence of ankyloglossia in the newborn has been reported to be 3% to10%.[42,43,44] If the frenulum limits movement of the tongue or pulls the tongue to a "V" at the tip, it is abnormal and may limit suck effectiveness primarily for breast feeding. This may be an indication for frenotomy (clipping of the frenulum) or if severe may require frenuloplasty (surgical removal of frenulum).[42,44,45]

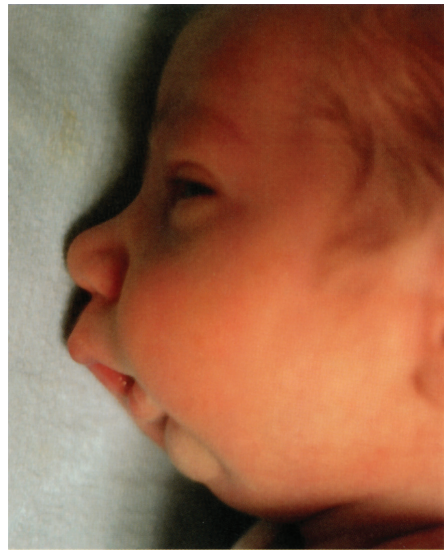

Figure 5.19 Pierre Robin sequence (micrognathia)

Source: From Clark DA. 2000. *Atlas of Neonatology.* Philadelphia, PA: Saunders, 144. Reprinted by permission.[36]

White patches on the tongue and mucous membranes may be from residual milk, and leukoplakia is normal in darkly pigmented infants. If the white coating cannot be easily removed with a tongue blade or cotton swab, it may be lesions of a candidal infection (oral thrush). A translucent or bluish swelling under the tongue is a mucocele or ranula. These mucous or salivary gland retention cysts usually resolve spontaneously.[2,4]

A gloved finger inserted into the infant's mouth with the finger pad up may be used to assess for continuity of the hard and soft palates, strength and coordination of suck, and for a gag reflex. A gloved finger can also be used to run along the gum line to assess for lumps or masses. Small whitish-yellow clusters of Epstein pearls may be seen at the junction of the hard and soft palates

and on the gums. Epstein's pearls are epithelial inclusion cells and usually disappear by a few weeks of age. They are histologically the same as Bohn nodules found on the gums and milia seen on the skin.[1,2,4]

The gums should be pink and smooth. Natal teeth are present at birth, and neonatal teeth erupt shortly after birth.[46] Natal teeth are rarely seen in Caucasian infants, but are a common variant in Native American infants.[47] Teeth or eruption cysts with teeth appearing after birth (neonatal teeth) are usually seen in the lower incisor region. Natal and neonatal teeth are usually immature caps of enamel and dentine with poor root formation and may be very mobile. They may interfere with breastfeeding, may cause ulceration of the infant's tongue and pain with feeding, and there is a presumed risk of aspiration. For these reasons, removal is generally recommended, after consultation with a dentist and the family. If the tooth is firmly implanted, removal may result in hemorrhage and may impact future dentition, and the dentist may choose to leave it in place and do follow-up examinations, including x-rays, to assess for any complications.[2,4,46,48]

Excessive oral secretions requiring frequent suctioning are abnormal in the newborn infant. The etiology could be esophageal atresia or poor swallow due to a neurologic abnormality. Strength and coordination of swallow should be assessed at every feeding. If there is concern, patency of the esophagus is assessed by passing a large-bore (usually #10 French) orogastric tube; an x-ray is done to confirm placement.[2]

NECK

The infant's neck is normally short, but severe shortness may be syndromic. To observe the shape and symmetry of the neck, elevate the shoulders, allowing the head to fall back slightly. Asymmetry is most likely the result of in utero positioning.

The neck must be visualized and palpated anteriorly, laterally, and posteriorly. The thyroid gland is difficult to palpate unless it is enlarged. Goiter, caused by intrauterine deprivation of thyroid hormone, is very unusual.[2]

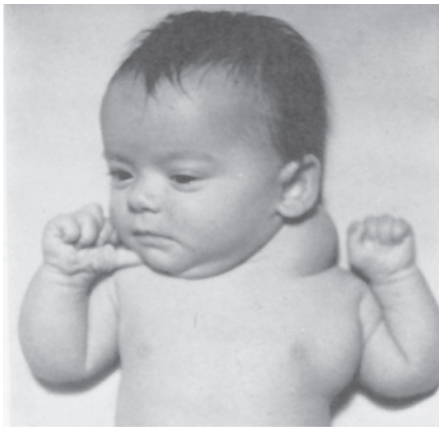

Figure 5.20 Cystic hygroma, with extension to the axilla

Source: From Koop CE. 1976. *Visible and Palpable Lesions in Children.* New York, NY: Grune & Stratton, 35. Reprinted by permission of the author.[49]

Cystic hygroma is the most commonly seen neck mass (Figure 5.20). It is caused by development of sequestered lymph channels, which dilate into large cysts. Cystic hygroma is soft and fluctuant, transilluminates well, and is usually seen laterally or over the clavicle. Its size can range from only a few centimeters to massive, and it may cause severe feeding difficulties or airway compromise. Very small lesions may regress spontaneously, but surgical resection is usually required.[1]

A mass high in the neck may be a thyroglossal duct cyst or a branchial cleft cyst. A branchial sinus may also be seen anywhere along the sternocleidomastoid muscle. It may communicate with deeper structures, and infection may necessitate surgical removal of the entire sinus tract.[50]

During palpation of the neck, note redundant skin or webbing of the neck. This is associated with Turner (Figure 5.21), Noonan, and Down syndromes.[1,6,8]

The clavicles should be palpated to assess for fracture, especially if there is a history of shoulder dystocia or macrosomia. Some infants present with nonrespiratory tachypnea caused by discomfort from the

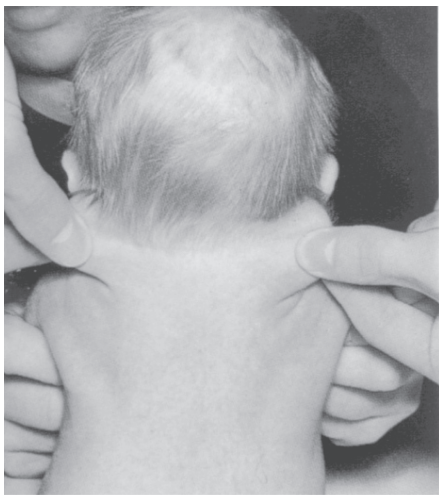

Figure 5.21 Webbed neck—Turner syndrome

Source: From Milner RDG, and Herber SM. 1984. *Color Atlas of the Newborn.* Oradell, NJ: Medical Economics, 102. Reprinted by permission of Blackwell Scientific Publications.[51]

fracture. Movement of the bone ends and crepitus may be felt soon after birth, or a fracture may not be evident for weeks until callus has formed and can be palpated as a mass over the clavicle. Fractured clavicles in the newborn heal spontaneously without treatment.

Assessment of neck reflexes, range of motion, and tone is described in Chapters 10 and 11.

SUMMARY

Careful examination of the head and neck is important because abnormalities presenting at birth in these regions are often indicative of other anomalies or a specific syndrome. Examination of the eyes and mouth requires the infant's cooperation, and the examiner needs to be alert for opportune times. States are now adopting the current recommendation for universal hearing screening for all newborns. The states with statutes or other regulatory language related to universal newborn hearing screening can be found at www.infanthearing.org/legislation.[52]

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Chest and Lung Assessment

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6

Physical assessment of the lungs begins with a careful review of the infant's history. Physical findings vary greatly with the infant's gestational and postnatal age. Other important factors from the infant's prenatal, intrapartum, and postnatal history are outlined in Table 6.1.

Begin the physical examination of the chest with observation so as not to disturb the infant before assessing breath sounds. Auscultation and palpation follow. Depending on the infant's physical condition, rest may be necessary between parts of the examination. Make provisions to ensure the infant does not become cold during the examination; cold stress will precipitate or further aggravate respiratory distress.

LANDMARKS AND STRUCTURE

The chest cavity is surrounded by the sternum, 12 thoracic vertebrae, and 12 pairs of ribs (seven true vertebrocostal pairs and five false, or vertebrochondral, dyads; Figure 6.1). The ribs in the neonate are much more cartilaginous than in the adult, accounting in part for increased chest-wall compliance and for retractions seen in the infant with respiratory distress. The lower boundary of the thorax is formed by the diaphragm, normally a convex muscular sheath. The diaphragm inserts on the sternum, the first three lumbar vertebrae, and the lower six ribs.[1]

Other palpable thoracic landmarks include the suprasternal notch, found on the upper aspect of the sternum, and the xiphoid process, which protrudes below the sternum. The clavicles and scapulae complete the bony structure of the chest.

The chest cavity consists of three spaces: the mediastinum and the right and left pleural cavities. The mediastinum contains the heart, esophagus, trachea, mainstem bronchi, thymus, and major blood vessels. The three lobes of the right lung and two lobes of the left lung are encased in serous membranes, which make up the visceral and parietal pleura (Figure 6.2).

Reference Lines

To aid in accuracy, use the following reference lines to describe the location of a physical finding during the examination of the chest and lungs (Figure 6.3):

- Anterior axillary line: Extends from the anterior axillary fold
- Midclavicular line: Vertical line drawn through the middle of the clavicle
- Midsternal line: Bisects the suprasternal notch
- Nipple line: Horizontal line drawn through the nipples

HISTORY

Prior to examining the newborn, review the maternal and newborn history. Check for any anomalies noted on the maternal ultrasound. In addition, note the infant's gestational age and any relevant maternal medical issues that may impact the infant's respiratory

TABLE 6.1: HISTORY TO BE COLLECTED

PRENATAL/INTRAPARTUM HISTORY
Gestational age
Maternal drug ingestion
Fetal distress
Maternal health (e.g., diabetes, fever)
Prolonged rupture of membranes
Meconium-stained fluid
Delivery mode
Apgar scores
POSTNATAL HISTORY
Corrected age
Duration of mechanical ventilation
History of RDS or CLD
History of pneumonia
Difficulty feeding
Apnea

CLD, chronic lung disease; RDS, respiratory distress syndrome.

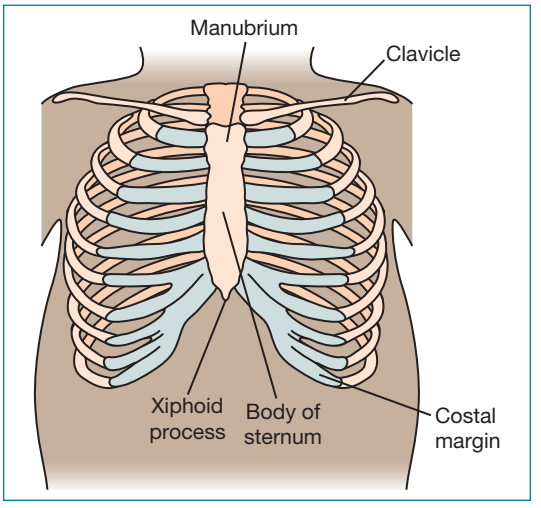


Figure 6.1 Bony structure of the chest

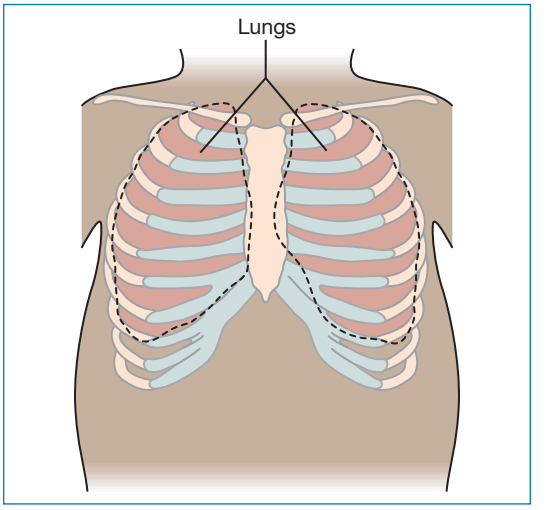


Figure 6.2 Internal structure of the chest

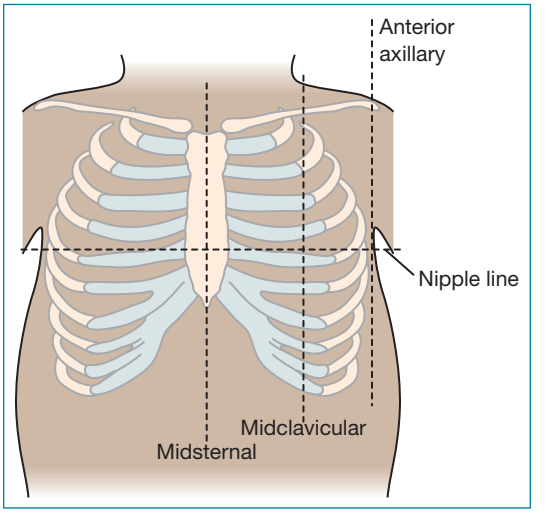


Figure 6.3 Reference lines

status. Depending on the age of the infant, note whether or not there has been any report of respiratory distress, increased work of breathing, or cyanosis. Note the infant's feeding pattern and any changes in color or respiratory effort associated with feeding.

INSPECTION

General

Begin the inspection with an overall assessment of the infant's color, tone, and activity. These findings provide clues to oxygenation and respiratory status.

Color

Observe the color of the infant's skin and mucous membranes. Normally the lips and mucous membranes should be pink and well perfused. Acrocyanosis (bluish coloration of the hands and feet) after birth is common and may persist during transition (up to 24 hours) following delivery.[2]

Color deviations might include cyanosis (either generalized or central [lips, tongue, and mucous membranes]) and acrocyanosis or mottling in the infant beyond transition. Other abnormalities in color, such as ruddiness and paleness, are discussed in Chapters 3 and 7.

Tone and Activity

Observe the infant's muscle tone and level and type of activity. Normal findings include flexed posture and active movement of all four limbs when awake. (Note the ability to attain and maintain flexion is decreased with prematurity.) Deviations include hypotonia and inactivity.

Respirations

Rate

Count the infant's respirations for 1 full minute. Normal findings are 30 to 60 breaths per minute, with wide variations. If the room is very warm or cool, the infant's respiratory rate can vary. Most infants experiencing temperature stress will be tachypneic (respirations >60 breaths per minute), but occasionally they may demonstrate bradypnea (respirations <40 breaths per minute). Because of the increased likelihood of retained fetal lung fluid, infants delivered by cesarean section have a higher incidence of transient tachypnea and respiratory distress.[3]

Although respiratory rates vary, tachypnea persisting beyond 2 hours of age may indicate underlying lung pathology (e.g., transient tachypnea of the newborn, respiratory distress syndrome [RDS], meconium aspiration, or pneumonia), hyperthermia, or pain. Bradypnea and shallow respirations are associated with central nervous system (CNS) depression secondary to factors such as maternal drug ingestion, asphyxia, or birth injury.

Quality

Observe the general appearance of the infant. Relaxed, symmetric, diaphragmatic respirations are normal. The newborn infant uses the diaphragm as the primary muscle of respiration. For the diaphragm to work efficiently, the rib cage must be stabilized by the intercostal muscles and the abdomen by the abdominal muscles. Coordination of this activity is still developing in term infants; it is less well developed in preterm infants, especially during rapid-eye-movement sleep, when respiratory instability may be seen.[1] To compensate for chest-wall instability, the infant's diaphragm is situated higher in the chest and is more concave in shape than the adult diaphragm, allowing for more efficient contractions.[1]

Work of Breathing

During normal respiratory efforts in the neonate, the lower thorax pulls in, and the abdomen bulges with each respiration. Deviations include asymmetric chest movement and excessive thoracic expansion. Paradoxical or "seesaw" respirations, where the chest wall collapses and the abdomen bulges on inspiration, suggest poor lung compliance and loss of lung volume.[4]

Infants are generally nose breathers, although most term infants will breathe through their mouth if the nares are occluded.[5] Nasal flaring, grunting, or moaning may be noted immediately after birth as the neonate attempts to clear fetal lung fluid from the lungs. The sounds are created by exhalation against a partially closed glottis in an attempt to increase the functional residual capacity in the lungs and stabilize the alveoli. The presence of fetal lung fluid or lung pathology may decrease lung compliance and, coupled with a more cartilaginous rib cage, can result in visible retractions of the chest wall. Retractions may be seen above or below the sternum, under the rib

cage (subcostal), or between the ribs (intercostal). Suprasternal retraction, especially if accompanied by gasping or stridor, is a sign of upper airway obstruction (laryngeal webs or cysts, tumors, or vascular rings). Severe respiratory distress immediately after birth may signal an underlying condition such as diaphragmatic hernia or pneumothorax. Figure 6.4 illustrates the most common sites for retractions. Beyond the immediate newborn period, flaring, grunting, and retractions suggest respiratory pathology (e.g., transient tachypnea of the newborn, RDS, atelectasis, or pneumonia).

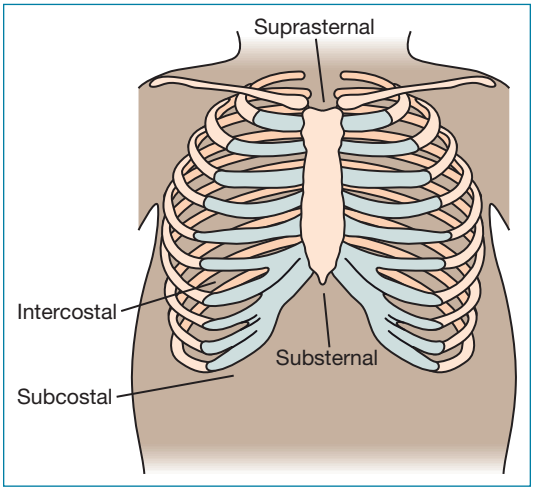


Figure 6.4 Sites of retractions

In 1956, Silverman and Andersen developed a scoring system to objectively measure respiratory distress; this system forms the basis of many of today's scoring systems.[6] This scoring system, shown in Table 6.2, can be used to quantify the extent of respiratory distress experienced by the infant at the time of examination.

Asymmetric chest movement is seen in the presence of conditions such as diaphragmatic hernia, cardiac lesions inducing failure, pneumothorax, or phrenic nerve damage. Sneezing is a common finding because it helps to clear the nasal passages. Coughing in the newborn is considered abnormal.

Pattern

While counting respirations, note the pattern (regularity) of inspiration. Normal newborns have an irregular pattern of respirations, which may vary with environmental temperature, sleep, and following a feeding. The less mature the infant, the more likely the breathing pattern is to be irregular. Periodic breathing (vigorous breaths followed by up to a 20-second pause) is common in preterm infants and may persist for several days after birth in term infants. For most preterm infants, the breathing patterns regularizes by the time they reach term gestation. In extremely preterm infants cardiorespiratory events may not resolve until close to 44 weeks postconceptional age.[7]

TABLE 6.2: ASSESSMENT OF RESPIRATORY DISTRESS

	SCORE		
CRITERION	**0**	**1**	**2**
Chest movement	Chest moves with abdomen	Chest sinks minimally as abdomen rises	Seesaw respirations; chest sinks as abdomen rises
Intercostal retractions	None	Minimal	Marked
Xiphoid retractions	None	Just visible	Marked
Expiratory grunting	None	Heard only with stethoscope	Audible with naked ear
Nasal flaring	None	Minimal	Marked

Note: The distress score is calculated by adding the values (0, 1, 2) assigned to each category. A score of 10 indicates maximum distress.

Source: Adapted from Silverman WA, and Andersen DH. 1956. A controlled clinical trial of water mist on obstructive respiratory signs, death rate, and necropsy findings among premature infants. *Pediatrics* 17(1): 1–10.[6]

A lapse of 20 seconds or more between respiratory cycles (one inspiration and one expiration) accompanied by bradycardia or color changes indicates apnea.[4] This condition, associated with prematurity, is gradually outgrown as the infant approaches 40 weeks postconceptional age. In extremely premature infants, periodic breathing may persist until 43 weeks postconceptional age.[8] Apnea in the term or close-to-term infant is considered abnormal and may indicate underlying illness (e.g., sepsis, hypoglycemia, CNS injury or abnormality, or seizures) or factors such as maternal drug ingestion.

Observe the quantity and quality of the infant's oral and nasal secretions. During transition, the appearance of oral and nasal secretions reflects the lungs' attempt to clear fetal fluid. Normal secretions are usually clear to white frothy mucus. Oral secretions will also reflect the stomach contents swallowed during delivery and therefore may be yellow or green in the presence of meconium, or blood-tinged if maternal blood was swallowed.

Deviations include excessive frothy oral secretions, which may indicate the presence of an esophageal atresia. Nasal stuffiness is associated with maternal drug use.[9] Snuffles (rhinitis) may be found with congenital syphilis.[10] Thick yellow secretions may be seen in the presence of a respiratory infection, and copious white nasal secretions are associated with respiratory syncytial virus (RSV) infection.

Components of the Chest and Respiratory System

Airways

The nasal portion of the airway is supported by bony and cartilaginous structures. The nasal passages are narrowed in the newborn; therefore, resistance to airflow in the nose contributes significantly to total pulmonary resistance. The pharyngeal component of the airway system is shorter in the infant than in adults and very compliant. In the absence of good muscle tone, the tongue can fall back against the soft palate and obstruct the airway, a process accentuated by neck flexion.[11] Look for the presence of nasal flaring or obvious signs of airway obstruction. In particular, infants with poor tone, a large tongue (macroglossia), or a small jaw (micrognathia) can experience upper airway obstruction.

Larynx and Trachea

In the newborn, the tracheal cartilages (hyoid, thyroid, and cricoid) are supported by superficial fascia. In premature infants, this fascia is underdeveloped, which increases the risk for airway obstruction.[12] The trachea and bronchi are supported by cartilaginous rings that are also less well developed than in the adult, increasing the risk for airway collapse and air trapping. Infants have the ability to alter laryngeal airway diameter by active movement of the vocal cords with breathing. Expiratory grunting in an infant with respiratory distress illustrates the infant's attempt to increase laryngeal airway resistance and ultimately to increase functional residual capacity in the lungs. Chemoreceptors in the larynx trigger reflex apnea to prevent entry of foreign substances.

On x-ray, the trachea is normally midline. Deviation suggests pneumothorax, a space-occupying lesion, or significant atelectasis. Rotation of the infant on the x-ray cassette may result in the false appearance of tracheal deviation.

Thorax

Observe the infant's chest and measure its circumference. The average chest circumference in term infants is 30 to 36 cm or 2 cm less than the occipital–frontal head circumference.[13] The difference between head and chest circumference is more exaggerated in premature infants and in those with intrauterine growth restriction, where the chest circumference will be small in comparison to that of the head.

In infants, the thorax is normally rounded rather than dorsoventrally flattened, as in older children. The ribs are oriented more horizontally, limiting the potential for

expansion of the rib cage.[14] The soft cartilaginous structure of the newborn's chest results in a tendency for the chest wall to collapse inward in the presence of decreased lung compliance. To preserve end-expiratory lung volume, the infant compensates by increasing the respiratory rate, shortening the inspiratory time, and grunting.[11]

In neonates, the anterior–posterior (AP) diameter of the thorax is approximately 85% of the transverse diameter.[15] A small thorax is seen in pulmonary hypoplasia, and a bell-shaped thorax may be an indication of neurologic abnormalities or dwarfing syndromes.[16] A barrel chest, characterized by an increased AP diameter is a finding characteristic of the air trapping seen in conditions such as transient tachypnea of the newborn, meconium aspiration, and overzealous mechanical ventilation. Sprengel deformity of the scapula occurs when the scapula fails to descend downward and becomes fixed to the cervical spine.[15]

Sternal deviations include protrusion of the sternum outward (pectus carinatum) or indented sternum (pectus excavatum; Figure 6.5). Pectus excavatum occurs in one to eight per 1,000 newborns and is usually of cosmetic rather than medical significance. Severe cases may cause pain, increase work of breathing, and restrict lung expansion.[17] The incidence of pectus carinatum is half that of pectus excavatum with a male predominance. About 25% of infants with pectus carinatum also have scoliosis. Pectus carinatum is associated with Marfan and Noonan syndromes, among other conditions.[17]

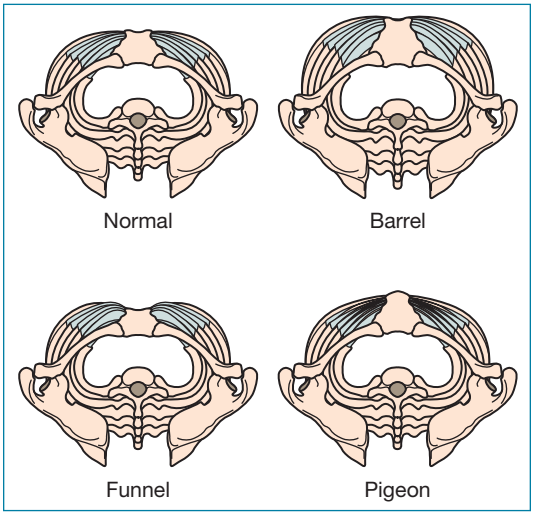


Figure 6.5 Different chest shapes

Rib anomalies are relatively common and usually of no clinical significance.[15] These anomalies may occasionally be noted on inspection or palpation but are more likely to be identified as an incidental finding on chest x-ray. Common anomalies include bifid ribs, rib agenesis, or bridging between the ribs. A cervical rib emanating from the seventh cervical vertebrae may cause compression of the brachial plexus or subclavian artery.[15] Eleven pairs of ribs may be seen in infants with Down syndrome or trisomy 18. Thin ribs are seen in infants with trisomy 13 or 18, and thickened ribs are a feature of thalassemia and mucopolysaccharidosis.[15] Flaring of the lower ribs, known as *Harrison groove*, is another finding that may be normal or associated with rickets. Abdominal distention may accompany hyperinflation as the diaphragm is pushed downward by air trapped in the lungs.

Muscle

Observe the chest for muscle development, symmetry, and bulges. The normal chest wall is symmetric and relatively smooth. Deviations include masses, atrophy, agenesis, and hypertrophy. Unilateral hypoplasia or absence of the pectoralis major muscle is a feature of Poland syndrome; other features of this syndrome include rib defects and upper limb hypoplasia.[15]

Cystic hygromas resulting from dilated lymphatic vessels may be found in the neck, chest wall, or axillae. Hemangiomas may be seen in the neck or chest wall at birth or may develop over the first weeks of life.

Nipples

Observe the number, placement, shape, and pigmentation of the nipples. Also inspect the nipples for fissures and secretions. In the term infant, the areolae normally are raised and stippled, with 0.75 cm to 1 cm of

palpable breast tissue. The distance from the outside of one areola to the outside of the other should be less than one quarter of the chest circumference.[18] The amount of breast tissue present at birth is also a useful indicator in gestational age assessment (Chapter 3).

In some infants, the influence of maternal estrogen results in breasts that are enlarged and engorged with a milky secretion known as *witch's milk*. Secretion of this liquid may last 1 to 2 weeks, and the enlargement may last several months.[19] Rarely, newborn infants may develop mastitis, characterized by redness, tenderness, breast enlargement, and discharge of pus.

Wide-spaced nipples are a feature of Turner syndrome; associated findings include lymphedema and redundant skin at the nape of the neck (see Figure 5.21).[20] Supernumerary (accessory) nipples are most commonly seen as raised or pigmented areas 5 to 6 cm below the normal nipple but can be located anywhere on a vertical line drawn through the true nipple. Accessory nipples are more commonly seen in males and infants of African American descent than those of European descent.[21] Some studies have shown an association between supernumerary nipples and congenital renal anomalies.[21,22] An increased risk of genitourinary malignancies has also been linked to the presence of supernumerary nipples.[21]

AUSCULTATION

Breath sounds are louder and coarser in the neonate than in the adult because the infant has less subcutaneous tissue to muffle transmission. Sounds are very readily referred in the neonate's chest because of its small size; therefore, it is difficult to localize adventitious (abnormal) sounds. Breath sounds may be decreased but are seldom absent, even over areas of atelectasis or pneumothorax. Breath sounds are less readily transmitted if (a) the pleural space contains fluid or air, (b) a bronchus contains secretions or foreign bodies, or (c) the lungs are hyperinflated. Sounds are more readily transmitted in the presence of consolidation, for example, with pneumonia.

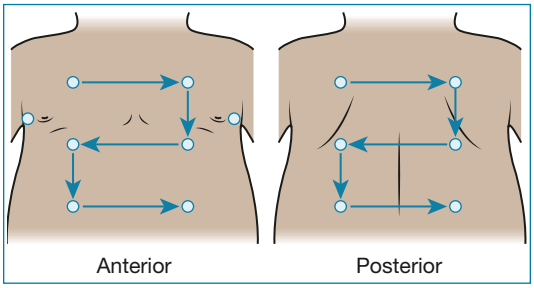


Figure 6.6 Sequence for breath sound auscultation: Anterior and posterior chest

To auscultate breath sounds, use both the bell and diaphragm of a warmed neonatal stethoscope. Begin at the top of the chest, and move systematically from side to side (Figure 6.6). Breath sounds in the lower lobes of the lungs can be assessed adequately only through the infant's back. Therefore, systematic auscultation of both the anterior and the posterior chest should be performed and one side of the chest compared with the other.

Normal Breath Sounds

Assess breath sounds for pitch, intensity, and duration. Normal breath sounds are described in adults and older children according to their location in the chest. As previously noted, localization of breath sounds in the neonate can be difficult. The following terms are sometimes used to describe normal sounds within the lungs. These terms are also listed in Table 6.3.

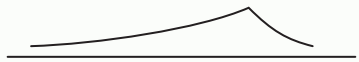
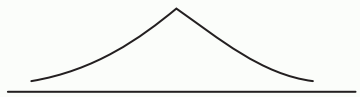
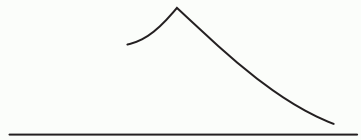
Vesicular Breath Sounds

Vesicular (from the Latin for *sac*) breath sounds are soft, short, and low pitched during expiration and louder, longer, and higher pitched during inspiration. These sounds are normally found over the entire chest, except over the manubrium and trachea.

Bronchial Breath Sounds

The loudest of the breath sounds, bronchial sounds are characterized by a short inspiration and a longer expiration. In adults, these

TABLE 6.3: NORMAL BREATH SOUNDS

SOUND	CHARACTERISTICS	FINDINGS
Vesicular	Heard over most of lung fields; low pitch; soft, short expirations	 Low pitch, soft expirations
Bronchovesicular	Heard over main bronchus area and over upper right posterior lung field; medium pitch; expiration equals inspiration	 Medium pitch, medium expirations
Bronchial	Heard only over trachea; high pitch; loud and long expirations	 High pitch, loud expirations

Source: Adapted from Hagler DA, and Traver GA. 2002. Respiratory system. In *Mosby's Clinical Nursing,* 5th ed., Thompson JM, et al., eds. Philadelphia, PA: Mosby, 156. Reprinted by permission.[23]

sounds are found over the trachea, but may be more widely transmitted in the neonate.[1]

Bronchovesicular Breath Sounds

Bronchovesicular sounds demonstrate an inspiration and an expiration that are equal in quality, intensity, pitch, and duration. Louder than vesicular but less intense than bronchial sounds, bronchovesicular sounds are normally found over the manubrium and intrascapular regions.

Adventitious Sounds

Adventitious breath sounds are usually a sign of disease and may be superimposed on normal breath sounds. Auscultation shortly after birth frequently demonstrates adventitious sounds resulting from the presence of fetal lung fluid and are common immediately following birth. Adventitious sounds heard at the onset of inspiration are more likely to result from secretions in the larger airways; sounds heard at the end of inspiration represent distal disease. Take care to distinguish sounds arising in the lungs from those arising in the upper airway. Placing the stethoscope over the infant's nose and mouth helps to localize upper airway sounds. Suctioning will often clear the upper airway and assist in identifying the presence of referred sounds. Table 6.4 summarizes some adventitious breath sounds.

Crackles

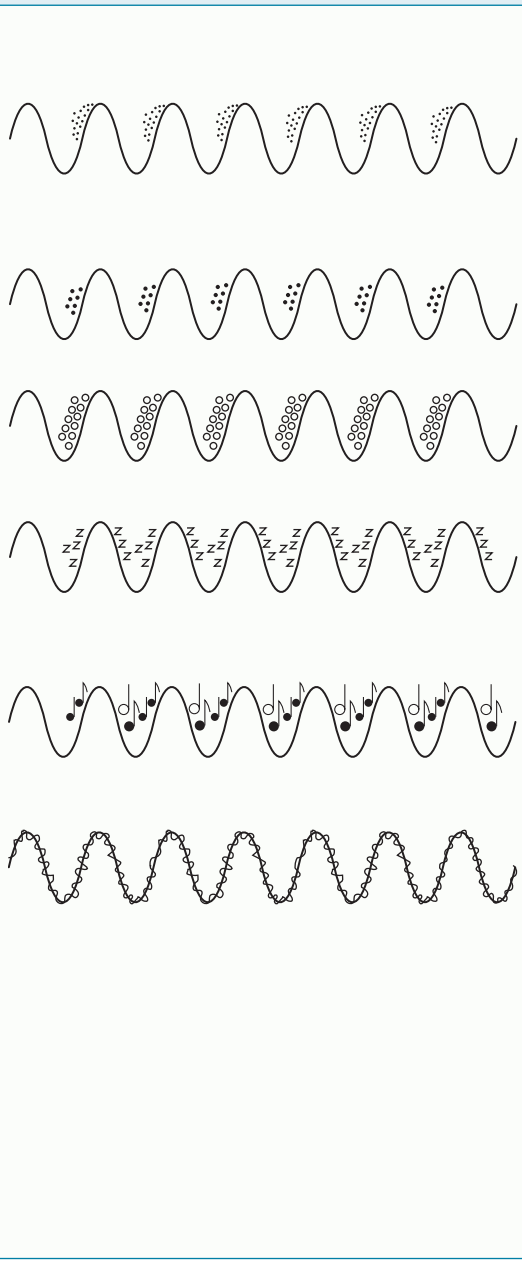
Crackles are defined as a series of brief crackling or bubbling sounds, arising from a sudden release of energy—either an airway popping open or a liquid film breaking. In the past, this type of sound has also been referred to as *rales*.

Fine crackles can be simulated by rubbing together a lock of hair. These sounds commonly originate in the alveoli in the dependent lobes of the lung and are usually heard at the end of inspiration. Frequently heard in the first few hours after birth, fine crackles are also associated with RDS and chronic lung disease (CLD).

Medium crackles sound similar to the fizz of a carbonated drink. Believed to originate in the bronchioles, these sounds are associated with the passage of air through sticky surfaces, such as those found with pneumonia, pulmonary congestion, or transient tachypnea of the newborn.

Coarse crackles are loud and bubbly. These sounds are associated with significant accumulations of mucus or fluid in the larger airways.

TABLE 6.4: ADVENTITIOUS BREATH SOUNDS

SOUND/CHARACTERISTICS	SIMULATION	PICTORIAL FINDINGS
Crackles: Discrete, noncontinuous sounds, sudden energy release		
Fine crackles (rales): High-pitched, discrete, noncontinuous crackling sounds heard during end of inspiration (indicates inflammation or congestion) (RDS)	Lock of hair	
Medium crackles (rales): Lower, moister sound heard during midstage of inspiration; not cleared by a cough (edema, pneumonia)	Fizzy drink	
Coarse crackles (rales): Loud, bubbly noise heard during inspiration; not cleared by a cough		
Rhonchi: Loud, low, coarse sound like a snore heard at any point of inspiration or expiration; coughing may clear sound (usually means mucus or foreign body in trachea or large bronchi)		
Wheeze: Musical noise sounding like a squeak; may be heard during inspiration or expiration; usually louder during expiration (CLD)		
Pleural friction rub: Dry rubbing or grating sound, usually due to inflammation of pleural surfaces; heard during inspiration or expiration; loudest over lower lateral anterior surface (mechanical ventilation in RDS)	Rubbing cupped hand	

CLD, chronic lung disease; RDS, respiratory distress syndrome.

Source: Adapted from: Hagler DA, and Traver GA. 2002. Respiratory system. In *Mosby's Clinical Nursing*, 5th ed., Thompson JM, et al., eds. Philadelphia, PA: Mosby, 156. Reprinted by permission.[23]

Rhonchi

When distinguished from wheezes, rhonchi are described as lower in pitch. They are more musical in quality than crackles. Rhonchi are seldom described in the neonate, but may be heard when either secretions or aspirated foreign matter is present in the large airways.

Wheezes

Also referred to as *high-pitched rhonchi*, wheezes may be heard on inspiration or expiration, but are usually louder on expiration. Seldom heard in the newborn, wheezes may be audible in the infant with CLD because of narrowing of the airways or the presence of bronchospasm.

Rubs

Rubs can be simulated by holding a cupped hand to the ear and rubbing a finger over the cupped hand. Rubs are usually associated with inflammation of the pleura; however, in the neonate, this sound is frequently described during mechanical ventilation.

Stridor

Stridor is a high-pitched, hoarse sound produced at the larynx or upper airways during inspiration or expiration. In a newborn, this sound indicates a partial obstruction of the airway and should be investigated promptly. Stridor may also be heard postextubation in infants with edema of the upper airway.

Bowel sounds are occasionally auscultated over the lung fields. These may be referred sounds transmitted from the abdomen. If these sounds persist over the lung fields, especially on the left side, diaphragmatic hernia should be considered.

PERCUSSION

The small size of the neonatal chest relative to the examiner's hands makes percussion of limited value as a part of the neonatal physical examination. The infant's chest is normally hyperresonant because of the thin chest wall. The experienced examiner may find percussion useful, however, in distinguishing between air and fluid or solid tissue in cases in which the infant is in distress and a pneumothorax, pleural effusion, or diaphragmatic hernia is suspected.

The technique for percussion involves placing one finger firmly against the chest wall and tapping that finger with the index finger of the other hand (see Figure 1.2). A change in resonance indicates a change in the consistency of the underlying tissue.

PALPATION

After a thorough inspection of the chest, auscultation of breath sounds, and percussion, if necessary, the examiner next palpates certain areas of the infant's chest.

Clavicle

The clavicle is the bone most commonly fractured during the birth process, occurring in 0.2% to 3.5% of all live births.[24] Palpate the entire length of the clavicles. Suspect fracture if crepitus, swelling, or tenderness is present. An infant with a fractured clavicle may also demonstrate an incomplete Moro reflex on the affected side (Chapter 11). Absence of one or both clavicles results in hypermobile shoulders and is a feature of cleidocranial dysplasia. Other chest deformities may also be present in this condition.[25]

Breast Tissue

As indicated in the discussion of inspection, the breast buds should be gently palpated to determine the presence of hypertrophy, fissures, secretions, or masses.

Sternum and Ribs

Palpate the sternum and the ribs for crepitus or masses. Crepitus may indicate subcutaneous air from an underlying pulmonary air leak. A lump or mass should be investigated for the presence of an underlying fracture. The tip of the cartilaginous xiphoid process often protrudes anteriorly and may be movable with slight pressure.

Overall Structure

Palpate the costal cartilage between the ribs to assess for hypertrophy. The costal cartilages enlarge in rickets and can be palpated as a series of small lumps down the side of the sternum. This phenomenon is known as the *rachitic rosary*.[26]

TRANSILLUMINATION

Transillumination is a useful adjunct to physical examination when a pneumothorax is suspected. Place a high-intensity fiberoptic light source perpendicular to the neonate's chest. While moving the light source back and forth from side to side, compare the amount of transillumination between the left and right and the upper and lower aspects of the chest. In a darkened room, air pockets will present with a large halo of light.[27]

Subcutaneous edema or air may result in a false-positive reading. Chest-wall edema, dark skin, tape, and equipment may obscure transillumination and result in a false-negative finding. In an early study of transillumination, a false-positive rate of 4% was identified.[28] Diagnoses obtained by transillumination should be confirmed by chest x-ray.

ASSESSMENT DURING MECHANICAL VENTILATION

Conventional Ventilation

The use of mechanical ventilation necessitates increased vigilance in all aspects of physical assessment. Pay particular attention to assessing the adequacy and symmetry of chest expansion and auscultating breath sounds. Eliminate water in the ventilator tubing before assessing chest sounds because gas bubbling through water produces referred sounds.

Breath sounds may be altered by the presence of an endotracheal tube, which effectively narrows the neonate's airway, and by the flow of gases from the ventilator, which may increase turbulence. Wheezes and rubs are more common and harsh or sandpaper breath sounds are often described in infants receiving mechanical ventilation for RDS. These sounds may result from the forced opening of atelectatic alveoli. Harsh breath sounds may also result from air leaking around the endotracheal tube. Listening with the stethoscope over the infant's mouth may be useful in differentiating sounds produced in the larynx from those produced in the chest. Air leaking around the endotracheal tube may produce a high-pitched sound heard during inspiration. Table 6.5 outlines some common concerns in the neonate receiving mechanical ventilation.

Patient-Triggered Ventilation

Ventilators that measure tidal volume allow the clinician to assess the neonate's ability to generate a spontaneous breath and to determine the relative size of that breath compared with the size of the breath generated by the ventilator. As the neonate's lung disease improves, larger tidal volumes will be generated, suggesting a readiness for weaning.

Ventilation in which a preset tidal volume is delivered results in breaths that require varying amounts of pressure to deliver the desired volume of gas. As the infant's lung compliance

TABLE 6.5: COMMON FINDINGS IN INFANTS RECEIVING MECHANICAL VENTILATION

FINDING	POSSIBLE CAUSE
Absence of air entry	Pneumothorax, blocked ETT, accidental extubation, space-occupying lesion
Decreased or unequal air entry	Atelectasis, pneumothorax, intubation of right bronchus
Asymmetric chest movement	Pneumothorax, intubation of right bronchus
Increased chest excursion	Change in compliance resulting in overventilation
Decreased chest excursion	Underventilation, blocked ETT, accidental extubation, air leak

ETT, endotracheal tube.

improves, less inspiratory pressure is required to deliver the same amount of volume. By monitoring the pressure used, the clinician can assess changes in the infant's lungs.

High-Frequency Ventilation

High-frequency ventilation techniques require several alterations in the traditional approach to physical assessment of the chest and lungs. The rapid rates used in high-frequency ventilation cause the infant to shake or vibrate, making it impractical to count respiratory rates. Many infants on high-frequency ventilation will be apneic; however, spontaneous respirations that do occur can be recorded in the usual fashion.

The chest should be both observed and palpated to assess the symmetry and quality of vibrations. Excessive movement of the chest or abdomen may indicate overventilation. Decreased or asymmetric movement may indicate complications such as air leak or airway obstruction.

Breath sounds during high-frequency ventilation will be high pitched, with a jackhammer quality. Auscultation should be performed to assess changes in the pitch or quality of sound. Higher pitched or musical sounds may indicate the presence of secretions. Decreases in pitch may indicate the presence of a pneumothorax. Traditional breath sounds can be assessed during periods of manual ventilation or with sighs given during oscillation.

SUMMARY

The respiratory system undergoes rapid and significant changes during transition to extrauterine life. These changes leave the lungs vulnerable to both transient and more life-threatening maladaptations. Careful scrutiny of the respiratory system is required to identify potential problems requiring treatment. Findings from physical examination of the chest and lungs form the basis for further investigations. Findings should be documented using standard terms and reference points to permit comparison with subsequent assessments.

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Cardiovascular Assessment

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7

Cardiovascular assessment of the newborn requires great skill in the techniques of inspection, palpation, and auscultation. Inspection of the general activity of the neonate, breathing patterns, presence or absence of cyanosis, and activity of the precordium are all important. Palpation of pulses, apical impulse, and thrills is also imperative. Auscultation, however, is the main focus of the cardiovascular examination. Through auscultation, the examiner assesses heart rate, rhythm, regularity, and heart sounds (especially murmurs). A pediatric or neonatal stethoscope with a diaphragm and a bell is very helpful for auscultation. The bell conducts sound without distortion (although it can be difficult to maintain an airtight seal). The bell is useful for low-pitched sounds. If properly sized, the diaphragm maintains its own seal and is useful for high-pitched sounds.

The dynamic properties of the newborn heart make cardiovascular assessment of the neonate challenging. The change from "fetal–placental" to "newborn-lung" circuitry means that the findings of the cardiovascular examination constantly change over the first few hours, days, and weeks of life. Those performing cardiovascular assessments must be aware of these alterations and their timing and incorporate this knowledge into their examinations.

Because changes in ductal flow, decreasing pulmonary vascular resistance, and increasing systemic vascular resistance occur over the first few hours and days of life, cardiovascular assessments ideally should be done shortly after birth, again at 6 to 12 hours of age, then at 1 and 3 days of age, and at regular intervals thereafter. Because this is rarely possible in the normal newborn, it is recommended that, at a minimum, examinations be done shortly after birth, at 1 day of age, and at regular pediatric office visits thereafter.

REVIEWING MATERNAL, FAMILY, AND BIRTH HISTORIES

The first step in a complete cardiovascular assessment is a thorough maternal history. Several maternal conditions can affect the neonate's cardiovascular system. These include maternal diabetes, systemic lupus erythematosus, and a maternal history of congenital heart disease (CHD). Maternal diabetes can increase the risk for CHD in the neonate to 3 to 4 times that of the general population.[1] The prevalence of CHD (for truncus arteriosus, ventricular septal defects [VSDs], atrial septal defects [(ASDs], coarctation of the aorta [coarc], D-transposition of the great arteries, tetralogy of Fallot (TET), Ebstein anomaly, interrupted aortic arch, pulmonary atresia with intact septum, tricuspid atresia, total anomalous pulmonary venous return, double-outlet right ventricle, atrioventricular [AV] canal, and hypoplastic left heart syndrome [HLHS]) in national estimates for the United States between 1998 and 2005 was about 81.4/10,000 live births.[2] VSDs and transposition of the great arteries are common defects seen in infants of diabetic mothers. There is also a specific cardiomyopathy, called *hypertrophic cardiomyopathy*, commonly found in infants of diabetic mothers.[3]

Systemic lupus erythematosus in the mother has been shown to increase the incidence of congenital complete AV block in

TABLE 7.1: ENVIRONMENTAL FACTORS ASSOCIATED WITH CONGENITAL HEART DEFECTS

TERATOGENIC INFLUENCE	RISK OF CARDIAC DEFECT (%)	COMMON TYPES OF DEFECTS
Maternal rubella	35	PDA, PPS, septal defects
Maternal diabetes	3–5	VSD, coarctationof the aorta, complete transposition
Maternal phenylketonuria	25–50	TET
Systemic lupus erythematosus	20–40	Complete heart block
Maternal alcohol abuse	25–30	Septal defects
Hydantoin	2–3	Pulmonary and aortic stenosis, coarc, PDA
Lithium	10–20	Ebstein's malformation, TA, ASD
Retinoic acid	?	Defects of ventricular outflow tracts
Trimethadione (historical)	15–30	TET, complete transposition, HLHS
Thalidomide (historical)	<5	TET, septal defects, truncus arteriosus

ASD, atrial septal defect; HLHS, hypoplastic left heart syndrome; PDA, patent ductus arteriosus; PPS, peripheral pulmonary artery stenosis; TA, tricuspid atresia; TET, tetralogy of Fallot; VSD, ventricular septal defect.

Source: Adapted from Burn J. 2002. The aetiology of congenital heart disease. In *Paediatric Cardiology*, 2nd ed., Anderson RH, et al. eds. New York, NY: Churchill Livingstone, 151. Reprinted by permission.[6]

the neonate.[4] These infants present with low resting heart rates, sometimes while in utero.

As women with CHD are living longer and reaching childbearing age, new information has become available. It has been documented that there is up to a 15% risk for CHD in the offspring of mothers with CHD.[5] Even though only a small percentage of CHD can be related to environmental factors, a history of the pregnancy (especially of the first 2 months, when the heart is forming) is important. Environmental factors known to increase the risk of congenital heart defects are listed in Table 7.1.[6] Drugs known to cause heart defects include amphetamines, alcohol, anticonvulsants (hydantoins, trimethadione, valproic acid, carbamazepine), lithium, retinoic acid, thalidomide, and warfarin (Coumadin).[5,7] In addition, viral infections during the last 2 weeks of pregnancy may cause acute myocarditis in the neonate.

Because of the influence of genetic factors, family history is an important feature of a cardiovascular assessment. The examiner must gather details about other siblings with CHD. If one parent is affected or if an older sibling had a specific defect, there is a 3% to 5% risk for recurrence.[8] Also, several specific disorders that might demonstrate dominant or recessive inheritance patterns are associated with specific congenital heart defects (Table 7.2).[9]

Details of the labor and delivery history are also important to the cardiovascular examination. Knowledge of any causal factors, such as perinatal hypoxia, maternal infection, or drugs given to the mother during labor, will help the examiner determine whether CHD is a likely explanation for abnormal physical findings.

Finally, birth weight, gestational age, and gender must be taken into consideration. There is an increased incidence of CHD in low-birth-weight infants, and premature infants have an increased risk for patent ductus arteriosus (PDA). Several congenital heart defects are more common in one gender than in the other.

OBSERVING APPEARANCE AND BEHAVIOR

Because of the central role the heart plays, a complete cardiovascular assessment encompasses most of the other systems. The cardiovascular

TABLE 7.2: DISORDERS ASSOCIATED WITH CONGENITAL HEART DEFECTS

DISORDER	PATIENTS WITH CARDIAC DISEASE (%)	TYPES OF CARDIAC ABNORMALITIES
de Lange syndrome	30	VSD, ASD, PDA, AS, EFE
Holt–Oram heart–hand syndrome	100	ASD-2, VSD or PDA in 2/3, conduction block, HLHS, TAPVC, truncus arteriosus
Marfan syndrome	Up to 100	Aortic aneurysm, AR, MR, TR, prolapse
Noonan syndrome		PS/dysplasia, hypertrophic cardiomyopathy, PDA, coarctation of the aorta
Carpenter syndrome		PDA, PS, VSD, TET, TGA, ASD, dextrocardia
Ellis van Creveld		ASD
Mucopolysaccharidosis type 1	50–60	Single atrium, primum ASD, coarctation of the aorta, HLHS
Trisomy 21 (Down syndrome)	40–50	AV canal, VSD, PDA, ASD-1 and ASD-2, TET
Trisomy 18 (Edwards syndrome)	90–100	VSD, polyvalvular disease, ASD, PDA
Trisomy 13 (Patau syndrome)	80	PDA, VSD, ASD, coarc, AS, PS
XO (Turner syndrome)	>50	Coarctation of the aorta, bicuspid aortic valve, aortic aneurysm, AS, VSD
Beckwith–Wiedemann syndrome	15 ?	Hypertrophic cardiomegaly, ASD, VSD, PDA, TET
CHARGE association	44	TET, DORV, ASD, VSD, PDA, PS
DiGeorge syndrome	>50	Interrupted aortic arch,TA, TET, right aortic arch
VACTERL association	10	VSD, ASD, TET
Williams–Beuren syndrome	50–80	Supravalvar AS, stenosis of left coronary artery, small aorta, multiple pulmonary arteries, cerebral and renal arteries, mitral valve prolapse, ASD, VSD, MR
Cleft lip and palate	25	VSD, PDA, TGA, TET, SV
Diaphragmatic hernia	25	TET
Omphalocele	20	TET, ASD
Intestinal atresia	10	VSD
Renal agenesis unilateral/bilateral	17/75	VSD

AR, aortic regurgitation; AS, aortic stenosis; ASD, atrial septal defect; ASD-1, primum atrial septal defect; ASD-2, secundum atrial septal defect; AV, atrioventricular; CHARGE, coloboma, heart defect, atresia choanae, growth retardation, genital anomalies, ear abnormalities; DORV, double outlet right ventricle; EFE, endocardial fibroelastosis; HLHS, hypoplastic left heart syndrome; MR, mitral regurgitation; PDA, patent ductus arteriosus; PS, pulmonic stenosis; SV, single ventricle; TA, truncus arteriosis; TAPVC, total anomalous pulmonary venous connection; TET, tetralogy of Fallot; TGA, transposition of the great arteries; TR, tricuspid regurgitation; VSD, ventricular septal defect.

Source: Adapted from Flanagan MF, Yeager SB, and Weindling SN. 2016. Cardiac disease. In *Avery's Neonatology: Pathophysiology and Management of the Newborn*, 6th ed., MacDonald MG, Mullett MD, and Seshia MMK, eds. Philadelphia, PA: Wolters Kluwer, 489–490. Reprinted by permission.[9]

examination cannot be considered in isolation from the neurologic, respiratory, abdominal, and skin examinations.

The initial cardiovascular assessment of the neonate should include general observation of the infant's overall appearance and behavior. A newborn with CHD may exhibit decreased activity and/or appear flaccid. Notation of extracardiac anomalies is important because CHD is associated with such abnormalities in approximately 25% of infants.[10] Increased incidence of CHD is seen with neurologic and gastrointestinal anomalies, tracheoesophageal fistulas, renal and urogenital irregularities, and diaphragmatic hernia.[5]

INSPECTING SKIN AND MUCOUS MEMBRANES

The next step in the neonatal cardiovascular examination is inspection of the infant's color. Accurate assessment of skin color depends on the observer's astuteness and on ambient temperature and lighting conditions.[11] A cyanotic Caucasian infant may look pink under bright lighting, and a centrally pink infant may look cyanotic under dim lighting. In a well-lit room, the infant should be centrally pink—that is, not only should his general color appear pink, but his lips, tongue, earlobes, and (in males) scrotum should also appear pink. The best indicator to use to determine central cyanosis is generally the tongue and oral mucosa due to the rich vascular supply and lack of pigmentation.[5,12]

Polycythemia

Many infants appear pink at rest but become deep red to purplish with crying. This is usually related to polycythemia, a condition found when the infant's central hematocrit is greater than 65%. Although polycythemic/plethoric infants may appear cyanotic, as neonates, they rarely are. The ruddy or reddish color may be mistaken for cyanosis simply because newborns who have increased amounts of hemoglobin usually have a larger percentage of that hemoglobin unsaturated. This unsaturated hemoglobin masks the saturated hemoglobin, and the infants appear purplish in color.

Cyanosis

Central cyanosis refers to the bluish color of the skin, lips, tongue, earlobes, scrotum (in males), and nail beds seen in the neonate with significant arterial oxygen desaturation. Central cyanosis becomes visible when there are at least 5 g of hemoglobin not bound to oxygen/100 mL of blood.[5,12] Central cyanosis must be differentiated from peripheral cyanosis (acrocyanosis), which is blue color in the hands and feet, and circumoral cyanosis, which is blue color around the mouth. Peripheral cyanosis is normal in newborns until about 2 days of age and is thought to be caused by vasomotor instability. No treatment is necessary for peripheral cyanosis. When determining whether cyanosis is central or peripheral, check the infant's arterial saturation via a pulse oximeter.[5] Central cyanosis is difficult to detect unless the arterial desaturation is less than or equal to 80% to 85% in an infant with a normal hemoglobin level.[5,12]

Central cyanosis can be the result of many things: lung disease, sepsis, persistent pulmonary hypertension, or neurologic disease. Cyanosis is also one of the two best indicators of CHD (symptoms of congestive heart failure is the other). It is therefore important to carefully assess cyanosis and the infant's response to oxygen. Cyanosis that does not improve with administration of 100% oxygen is most likely the result of cardiac causes, as is cyanosis that increases with crying.[13]

Pallor/Mottling/Perfusion

Pallor and mottling of the skin should also be considered in assessing cardiac status, as should the infant's general overall perfusion. Infants with a compromised cardiac status may appear pale as a result of

vasoconstriction and shunting of blood away from the skin to more vital organs. Mottling, if present, must be evaluated with any other signs and symptoms associated with it. It may be a sign of cardiogenic shock when associated with hypovolemia or decreased cardiac output. Mottling can also be seen in normal infants under certain circumstances, such as in the stressed or cold neonate, when it is then referred to as *cutis marmorata.* A hypoxic, anemic infant may not appear cyanotic because hemoglobin levels may be too low to produce a bluish color.[5,12]

Capillary filling time can give valuable information about the infant's cardiac perfusion to the skin and should be determined by pressing a finger against the infant's skin in both a central and a peripheral area. When blanching is noted, the examiner counts the seconds required for the color to return to the skin once the pressure is released. Capillary filling time of greater than 3 seconds is considered abnormal.[12]

Edema

In the infant, unlike in older patients, edema is rarely associated with cardiac problems. Isolated peripheral edema is a hallmark sign of Turner syndrome, however. Infants with this syndrome show a high incidence of coarctation of the aorta.

OBSERVING BREATHING PATTERNS

Respiratory activity must be observed in relation to the cardiac examination. Respiratory rate and effort should be documented (as addressed in Chapter 6). Signs of respiratory distress—such as grunting, nasal flaring, retractions, tachypnea, and crackles—may be signs of congestive heart failure. It is also important to keep in mind that a cyanotic infant with nonlabored respiratory effort may be cyanotic as a result of a congenital heart defect that restricts pulmonary blood flow.[14] Nonlabored tachypnea that occurs with central cyanosis is a result of hypoxic respiratory drive.[12]

PALPATING PERIPHERAL PULSES

The character of the peripheral pulses should be assessed next. This is best done when the infant is quiet. Pulse rate, rhythm, volume, and character should all be examined. Pulses represent an approximate determination of cardiac output. The axillary, palmar, brachial, radial, femoral, popliteal, posterior tibial, and dorsalis pedis may be palpated using the index finger (Figures 7.1–7.4). (Note that the dorsalis pedis pulse may not be felt in newborns; this is considered normal.)

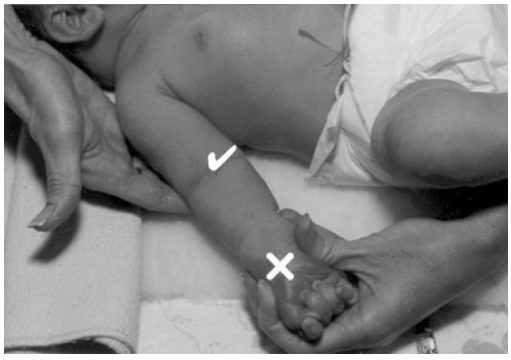

Figure 7.1 Radial and brachial pulses

Demonstrates the location of radial pulse (✖), palpable on the flexor surface of the wrist just medial to the distal end of the radius. Demonstrates the location of the brachial pulse (✔) in and above the groove of the elbow, medial to the biceps muscle and tendon.

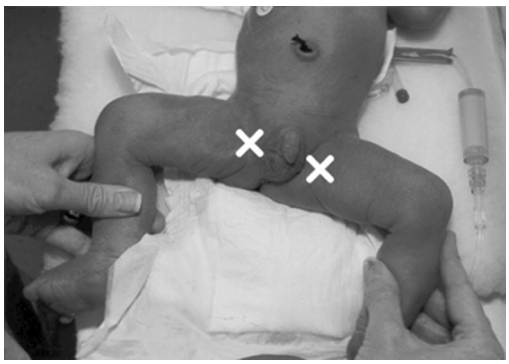

Figure 7.2 Femoral pulses

Demonstrates the location of the femoral pulses (✖), palpable just below the inguinal ligament, an equal distance between the pubic tubercle and the anterior superior iliac spine.

Figure 7.3 Posterior tibial pulse

Demonstrates the location of the posterior tibial pulse (✖), palpable just behind and slightly below the medial malleolus.

Figure 7.4 Dorsalis pedis pulse

Demonstrates the location of the dorsalis pedis pulse (✖), palpable on the dorsum of the foot by following an imaginary line that traces the groove between the first and second toes.

Pulse Rate/Rhythm

On initial palpation, the pulse rate should be noted. The significance of rate is discussed later in this chapter. After determining the rate, document any irregularities in rhythm or any skipped beats. A pulse deficit (the difference between the heart rate counted by auscultation and that counted with a pulse) is frequently seen with ectopic rhythms.

Pulse Volume/Character

Probably the most important determinations in neonatal pulse evaluation are those assessing volume and character. Volume of peripheral pulses is assessed on a scale of 0 to +3, with +3 being the strongest and 0 representing an absent or nonpalpable pulse. Pulses can be classified in the following way[15]:

3+ = Full or bounding

2+ = Normally palpated

1+ = Thready or weak

0 = Absent

At a minimum, the femoral and brachial pulses should be palpated bilaterally, and then one femoral and the right brachial should be palpated simultaneously.[5,12] (The right brachial should be palpated instead of the left because the right subclavian artery is always preductal, but the left subclavian artery may or may not be preductal.) Absent or weak femoral pulses—especially in comparison to the right brachial pulse—are abnormal and may indicate decreased aortic blood flow such as is seen with coarctation of the aorta, aortic stenosis, and HLHS.

Bounding pulses in any extremity should be noted. Bounding to full pulses are usually present with PDA and other aortic-runoff lesions (truncus arteriosus, aortic regurgitation, systemic arteriovenous fistula).[5,12] Normal premature infants have easily felt peripheral pulses, but strong palmar or digital pulses indicate a wide pulse pressure such as that seen with PDA.[16] Weak or absent peripheral pulses occur in the presence of low cardiac output from any cause (e.g., shock and cardiac tamponade), but also occur in lesions with decreased systemic blood flow that rely on the ductus arteriosus for systemic blood flow, such as hypoplastic left ventricle.[17,18]

INSPECTING AND PALPATING THE CHEST

Precordium

In the cardiac examination, assessment of the chest should begin with inspection of the precordium, the area on the anterior chest under which the heart lies (Figure 7.5). Generally, except during the first few hours

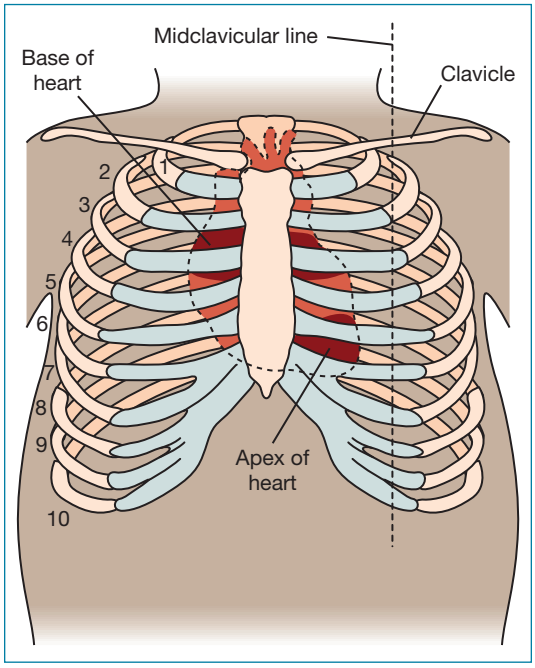

Figure 7.5 Position of the heart within the chest cavity and base and apex of the heart

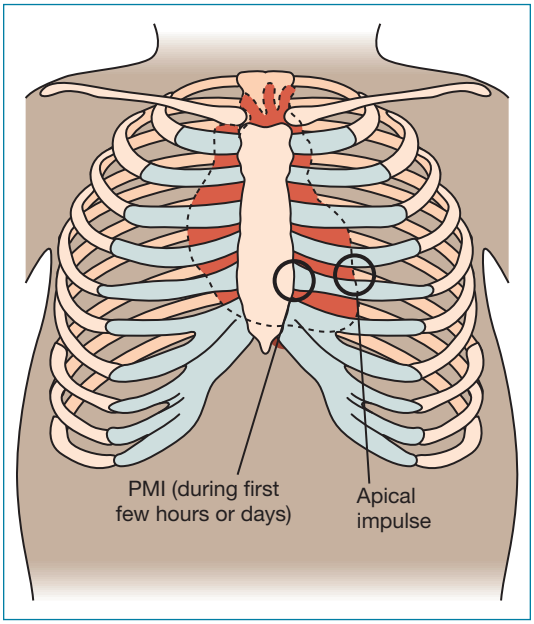

Figure 7.6 Areas of cardiac inspection and palpation

PMI, point of maximum impulse.

of life, the precordium of a term neonate should be quiet. During the first few hours of life, there may be a visible impulse noted along the lower left sternal border in normal newborns because of the right ventricular predominance common to transitional circulation. As the transitional circulatory pattern changes, this sign will generally disappear in the normal newborn. When seen in the term neonate after the first hours of life, a bounding precordium is characteristic of heart disease, typically defects with increased ventricular volume, such as left-to-right shunt lesions (PDA or large VSD), or severe valvular regurgitation such as seen with mitral or aortic insufficiency.[5,12,18] Premature infants may be seen to have an active precordium because they have less subcutaneous tissue than the term newborn.

Apical Impulse

After general assessment of the precordium, the examiner should inspect for position and character of the apical impulse, the forward thrust of the left ventricle during systole. The apical impulse is usually seen in the neonate in the fourth intercostal space, either at or to the left of the midclavicular line (Figure 7.6). It can be further localized and examined with light palpation using the fingertips. An apical impulse placed downward and to the left suggests left ventricular dilation. A very sharp apical impulse is found with high cardiac output or left ventricular hypertrophy.

The point of maximum impulse (PMI) and the apical impulse are usually the same, but this is not always true in neonates during the first few hours to days of life. During that time, an impulse stronger than the apical impulse can be found in the fifth intercostal space at the lower sternal border or even substernally. This then represents the PMI and is normal because of the right ventricular predominance found in the newborn (see Figure 7.6). The PMI or the apical impulse may be displaced in several situations, such as dextrocardia, tension pneumothorax, and diaphragmatic hernia.

Heaves, Taps, and Thrills

Further palpation of the precordium will yield other valuable information. Heaves, taps, and thrills can all be palpated. These findings are usually best felt by palpating with the portion of the palm at the base of the fingers rather than with the fingertips. This is because certain parts of the hand are more discriminatory than others for specific sensations. Vibratory sensations are best felt with the ulnar surface of the hand.

A heave (or lift) is a PMI that is slow rising and diffuse. Heaves are associated with volume overload.[5]

A sharp, well-localized PMI is called a *tap*. Taps are usually associated with pressure overload.[5] Thrills are low-frequency, palpable murmurs that feel similar to touching a purring cat. On the chest they are best felt with the palm of the hand; a thrill is felt best with the fingertips when it is in the suprasternal notch or over a carotid artery. A thrill denotes a loud murmur (at least a grade IV murmur). In the neonate, thrills are not common. When present, they can provide useful information about cardiac problems. A thrill in the upper left sternal border originates from the pulmonary valve or pulmonary artery and may be associated with pulmonary stenosis, tetralogy of Fallot, or rarely, a PDA.[5]

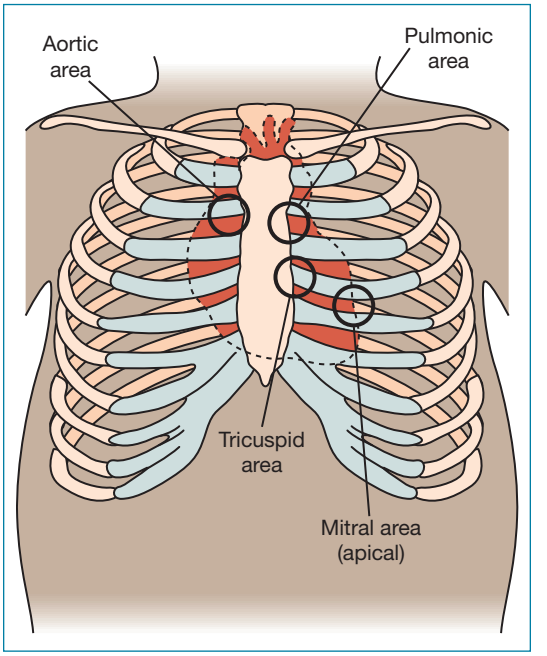

Figure 7.7 Four auscultatory areas of the heart

AUSCULTATING TO ASSESS CARDIOVASCULAR STATUS

Expert auscultation of the neonatal heart requires much practice over time. An experienced mentor can help the fledgling examiner learn to identify and distinguish heart sounds. The neonatal heart should be auscultated with the infant inactive and quiet.

At a minimum, the four traditional auscultatory areas should be examined (Figure 7.7). These are the aortic area (second intercostal space, right sternal angle), pulmonic area (second intercostal space, left sternal angle), tricuspid area (fourth intercostal space, left sternal angle), and mitral area (fourth intercostal space, left midclavicular line). A more thorough examination is recommended, however. It should include the right and left infraclavicular areas, both sides of the back, the right anterior chest, both axillae, the anterior fontanel (examining for cerebral arteriovenous fistulas), and the liver (examining for hepatic arteriovenous fistulas).

Heart Rate

Heart rate should be auscultated first and counted. A term neonate's heart rate at rest should generally be 120 to 140 beats per minute (bpm), although fluctuations above and below these values can be anticipated in a normal newborn. That is, the heart rate may increase to ≥170 bpm with activity or crying, or it may decrease to 70 to 90 bpm in deep sleep.[17] Premature infants have a slightly higher mean heart rate than that seen in term infants.

Sinus bradycardia, or a heart rate less than normal for age (usually <80 bpm), is a common transient finding in both term and premature infants. The reason for this is the predominance of the parasympathetic system. Any stimulus—such as yawning,

stooling, or suctioning—may result in vagal stimulation and subsequent bradycardia. These episodes are usually transient, require no treatment, and are self-correcting. Some episodes, especially in premature infants, may require stimulation or treatment of the underlying cause, such as treatment for apnea of prematurity with methylxanthines.

Sinus tachycardia is defined as a heart rate greater than normal for age (usually >180–200 bpm). It is generally considered the most common form of rapid heart rate in the neonate.[19] It normally occurs with any stimulus—such as crying, feeding, fever, or activity—that causes increased demands on the heart. Normally, with removal of the stimulus, the heart rate slowly returns to baseline. Sinus tachycardia rarely requires treatment.

Variation in the heart rate is normal among neonates and is seen as a positive sign of the infant's ability to react to the environment.[19] Infants who do not respond to stimuli with an increase in heart rate are clearly abnormal and should be observed very closely.

Although sinus tachycardia with rates up to 200 bpm can be tolerated by the neonate, heart rates greater than 200 bpm or supraventricular tachycardia (SVT) cannot. SVT encompasses paroxysmal atrial tachycardia and atrial flutter and fibrillation. Figure 7.8 depicts a rhythm strip of SVT. In the neonate, SVT represents a medical emergency and requires immediate intervention. At such rapid heart rates, cardiac output is extremely compromised because of short diastolic filling time. Without treatment, decreased cardiac output will cause congestive heart failure within 48 hours, and possibly death. Treatment for SVT depends on the cause, but the condition may respond to vagal stimulation (such as applying a cold washcloth or ice to the face), medications, or cardioversion.

Cardiac Rhythm and Regularity

After assessing heart rate, evaluate cardiac rhythm and regularity. Listen carefully to the rhythm of the heart sounds, and determine whether there is any irregularity. Noting patterns and frequencies of the irregularity helps identify the type of arrhythmia. Whenever an arrhythmia is suspected, an ECG and/or continuous heart monitoring is indicated to establish a diagnosis. Arrhythmias are not uncommon in the neonate. Fortunately, they are usually benign and require no treatment. Those most commonly found in the neonate include the following:

- *Sinus arrhythmia*: This is a very common, normal variant in most newborns. It is associated with respirations.[5] Sinus arrhythmia is characterized by irregularity of the R–R interval, with an otherwise normal cardiac cycle. No treatment is required for this rhythm.
- *Premature atrial contractions*: This arrhythmia is marked by an early beat arising from a supraventricular focus.

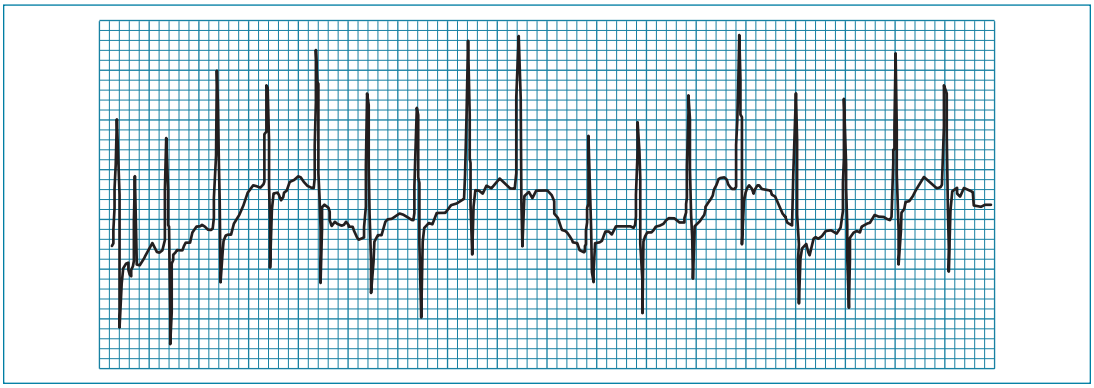

Figure 7.8 ECG rhythm strip of infant with supraventricular tachycardia with a ventricular rate of approximately 300 bpm

Ventricular conduction is usually normal. The arrhythmia is almost always benign in the newborn, but can be abnormal when seen with CHD; sepsis; hypoxia; hyperthyroidism; cardiac tumors; myopathies; electrolyte abnormalities; digoxin toxicity; administration of caffeine, atropine, theophylline, or inotropic agents; irritation from a centrally placed catheter, and severe respiratory distress.[20] If significant, the underlying cause may require treatment (especially if related to digitalis toxicity), but these beats are usually well tolerated, and no treatment is indicated.[5]

- *Premature ventricular contractions*: With this early beat arising from an irritable ventricular focus, ventricular conduction will be abnormal, giving rise to a wide and bizarre QRS complex. The arrhythmia may result from hypoxia, CHD, irritation caused by an invasive catheter, or as the result of a surgical procedure. Treatment is unnecessary if the phenomenon is infrequent.

Heart Sounds

Traditional cardiac physiology teaches that the first heart sound (S1) is produced by closure of the mitral and tricuspid valves, and the second heart sound (S2) is produced by closure of the aortic and pulmonic valves. More recent literature debates this, however, and the true origin of heart sounds has been found to be the sudden slowing of blood flow following closure of the valves.[12] Despite this, the traditional concept is helpful in conceptualizing and learning to recognize heart sounds and therefore continues to be used.

S1

S1, if thought of as representing closure of the mitral and tricuspid valves at the onset of ventricular systole, should be heard most loudly at the apex of the heart. (See Figure 7.5 for location of the apex and base of the heart.) Indeed, S1 is best heard at the mitral or tricuspid area (see Figure 7.7). S1 is usually loud at birth, decreasing in intensity during the first 48 hours of life. Any factor that increases cardiac output also increases the intensity of S1.

Splitting is defined as hearing two distinct components of a heart sound. It is caused by the asynchronous closure of the two valves that create the heart sound. The neonate's rapid heart rate usually makes splitting difficult to distinguish. Although some studies have documented splitting of S1 in the newborn, splitting is not commonly heard; S1 is usually described as being single.

S2

S2, if thought of as representing closure of the aortic and pulmonic valves, should be heard most loudly *at the base of the heart* (see Figure 7.5). Indeed, S2 is best heard *at the aortic or pulmonic area* (see Figure 7.7). S2 is usually single at birth, but by 1 to 2 days of age, splitting of the S2 with the infant's respirations should occur.[12] The trained examiner should be able to recognize, with practice, the splitting of S2 despite the infant's rapid heart rate.

Wide splitting of S2 should be considered abnormal in newborns. It can occur with ASD, pulmonary stenosis, Ebstein's anomaly, partial anomalous pulmonary venous return, mitral regurgitation, or right bundle branch block.[5,17]

S3

In addition to S1 and S2, extra heart sounds can potentially be heard in the neonate. Again, because of the infant's rapid heart rate, they may be difficult to distinguish. A third heart sound (S3) can occasionally be heard in infancy; if present, it is best heard *at the apex of the heart during early diastole* (see Figure 7.5). S3 most often signifies rapid or increased flow across the AV valves (rapid ventricular filling) and is commonly heard in premature infants with PDA. Rarely, it can be heard in the newborn with overt congestive heart failure.[21]

S4

A fourth heart sound (S4) should also rarely be heard in neonates. If present, it is heard *at the apex of the heart and is a low-pitched sound of late diastole* (see Figure 7.5). S4 is always pathologic and is heard in conditions characterized by decreased compliance (especially cardiomyopathy) or congestive heart failure.[5] The term *decreased compliance* refers to myocardium that is relatively stiff and therefore does not expand well as the blood enters the chambers. This affects the volume of blood that is ejected by the heart with contraction.

Ejection Clicks

Ejection clicks (snappy, high-frequency sounds) can, if they are present, best be heard just after S1. Ejection clicks occur at the time of ventricular ejection and resemble in timing, but not in quality, a widely split S1. Ejection clicks are commonly heard during the first 24 hours of life and during that time are usually normal (related to the concurrent pulmonary hypertension). Ejection clicks are always considered abnormal after the first 24 hours of life. The most frequent findings associated with these clicks are aortic or pulmonic stenosis, idiopathic dilation of the pulmonary artery, systemic or pulmonary hypertension, truncus arteriosus, or tetralogy of Fallot.[5]

Murmurs

Murmurs are caused by turbulent blood flow. They are often described as prolonged heart sounds. There are two kinds of murmurs: innocent and pathologic. Pathologic murmurs result from underlying cardiovascular disease, innocent murmurs do not.

Whenever an examiner detects a murmur, the question of its origin arises. The murmur must therefore be fully evaluated as to its timing, location, intensity, radiation, quality, and pitch. The neonate's age in hours and days is also especially significant because of the dynamic properties of the newborn heart. Putting all this information together with other findings of the physical examination should help the examiner determine the significance of the murmur.

The *timing* of a murmur is the first quality the examiner must listen for. To evaluate timing, the examiner must understand what is happening to the heart during systole and diastole. Systole is the period when the heart contracts and the heart chambers eject blood. Systole occurs following closure of the mitral and tricuspid valves, and its onset occurs just after S1. Diastole is the period when the heart is relaxed and the chambers are filling with blood. Diastole occurs following closure of the aortic and pulmonic valves, and its onset occurs just after S2. When establishing the timing of a murmur, the examiner must ask: "Does it occur in systole or diastole?" That is, does the murmur occur after S1 or after S2? Is the murmur early, mid, or late in systole or diastole? Does it occur throughout systole (holosystolic or pansystolic) or during midsystolic ejection? Continuous murmurs are heard through both systole and diastole.

Loudness, or intensity, of the murmur should be determined next. Murmurs are graded from I to VI, as follows:

Grade I: Barely audible, audible only after a period of careful auscultation

Grade II: Soft, but audible immediately

Grade III: Of moderate intensity (but not associated with a thrill)

Grade IV: Louder (may be associated with a thrill)

Grade V: Very loud; can be heard with the stethoscope rim barely on the chest (may be associated with a thrill)

Grade VI: Extremely loud; can be heard with the stethoscope just slightly removed from the chest (may be associated with a thrill)

Sometimes the intensity of a murmur will vary from examination to examination. The reason may be changing pulmonary vascular resistance or anything else that alters the status of cardiac output, such as anemia, activity, or changing ventilatory requirements.

Location of the place on the chest wall where the murmur is heard at maximum intensity is another important feature to evaluate. Location is usually described in terms of the interspace and the midsternal, midclavicular, or anterior axillary lines (see Figure 6.3), because the anatomic site of most murmurs is usually found at the location below where they are best heard. Describing murmurs in terms of aortic, pulmonary, tricuspid, or mitral areas is not recommended in neonates because malposition of valves and vessels may be found in CHD.

Document other locations where a murmur is heard. This is described as radiation or transmission of the murmur. Radiation from the normally positioned pulmonary outflow tract is to the left upper back; radiation from the normally positioned aortic outflow tract is to the carotid arteries.

Quality and *pitch* of the murmur are the final two features that should be assessed. Pitch is described as high, medium, or low. High-pitched murmurs occur when there is turbulence from a high-pressure area to a low-pressure area.[16] This can happen with aortic or mitral insufficiency. Low-pitched murmurs occur when there is a low-pressure difference in the turbulent flow. An example of this is mitral stenosis. Identifying the quality of a murmur (terms include *harsh, rumbling*, or *musical*) also helps to describe it.

Innocent Murmurs

During the first 48 hours of life, many newborns have murmurs, the majority of which are innocent. They are usually associated with the decreasing pulmonary vascular resistance occurring at this time and with the gradual closure of the PDA. Innocent murmurs, sometimes called *flow murmurs*, are most often grade I or II, are associated with normal ECG and chest x-ray findings, are usually systolic murmurs, and are not associated with any other symptoms. Some of the more common innocent murmurs heard during the first 48 hours of life include the following:

- *Systolic ejection murmur*: This is the most common innocent murmur (heard in up to 56% of newborns).[16] Usually grade I to II/VI, it is best heard along the mid and upper left sternal border and described as vibratory. Systolic ejection murmurs present within the first day of life, may last as long as 1 week, and are most likely the result of the significant increase in flow across the pulmonary valve associated with rapidly decreasing pulmonary vascular resistance.[21]
- *Continuous systolic or crescendo systolic murmur*: This murmur occurs in up to 15% of normal infants with a usual intensity of grade I to II/VI; it is best heard in the upper left sternal border. Presenting within the first 8 hours of life, this murmur is caused by the transient left-to-right flow through the ductus arteriosus during the period when pulmonary vascular resistance is falling but ductal closure has not yet been accomplished.[21]
- *Early soft midsystolic ejection murmur* (also called *peripheral pulmonic stenosis, pulmonary flow murmur*, or *pulmonary branch murmur*): Heard often in newborns, especially premature neonates, its intensity is usually grade I to II/VI, and it is medium to high pitched. This murmur is best heard in *the upper left sternal border, with wide radiation to both lung fields, axillae, and back*.[12] It presents within the first or second week of life.[22] Early, soft, midsystolic ejection murmurs last for weeks to months, but generally disappear by 3 to 6 months of age.[5,12] They result from turbulence produced at the relatively acute angle of the bifurcation of the pulmonary artery.[5,23]

Pathologic Murmurs

Pathologic murmurs in the neonate occur at varying times, depending on the anatomic abnormality causing them and on normal changes associated with transitional circulation. For example, pathologic murmurs heard in the delivery room are almost always

caused either by stenosis or by regurgitation; they are almost never the result of shunting because vascular resistances in the lungs and the body are equal at birth.[5] Many specific defects do not present with a murmur until 3 days (when the PDA is generally closing), 1 week, or 4 to 6 weeks of age, when the pulmonary vascular resistance has fallen sufficiently. ASDs sometimes do not present in the infant until 1 to 2 years of age.[5] The absence of a murmur does not rule out the potential for serious CHD. As many as 20% of infants who die from congenital heart defects during the first month of life do not have heart murmurs.[21] In fact, the absence of a murmur may be an ominous sign in both acyanotic and ductal-dependent lesions because it may indicate ductal closure.

Approach all murmurs cautiously. Soft murmurs heard in otherwise asymptomatic infants can be observed carefully by the experienced examiner for the first 48 hours of life. But any murmur that persists beyond this time, is louder than a grade I or II, or occurs in a symptomatic neonate requires further investigation. The cardiovascular workup should include a chest x-ray, an echocardiogram, and a consultation with a cardiologist. Many cardiologists also believe that an ECG is an important component of a complete cardiac evaluation.

Pathologic murmurs are more difficult to categorize than innocent murmurs. The following commonly occur in the immediate neonatal period:[5]

- *Loud systolic ejection murmur*: Usually grade II to V, this murmur appears within hours of birth and is almost always the result of aortic or pulmonary stenosis or coarctation of the aorta.
- *Continuous murmur*: Occurring in one -third of premature infants with a PDA, this murmur may only be systolic and difficult to hear in an infant being ventilated. This type of murmur is also heard in infants with arteriovenous fistulas, regardless of gestational age.

In addition, pathologic systolic murmurs may occasionally be heard with mitral and tricuspid insufficiency of various causes (especially with left ventricular failure in infants with critical left ventricular outlet obstruction and tetralogy of Fallot). Pathologic murmurs associated with VSD and PDA in term neonates do not present until pulmonary vascular resistance has fallen—often not until after discharge from the nursery or at several weeks of age.

PALPATING THE LIVER

Palpation of the liver (described in Chapter 8) is a significant part of the cardiovascular assessment. The liver becomes engorged when central venous pressure increases. A liver located more than 3 cm below the right costal margin is a good indicator of right-sided heart failure in a term infant.

EVALUATING BLOOD PRESSURE

Evaluation of the neonate's blood pressure (BP) should be done with the infant quiet. This assessment should be left until the end of the examination because the pressure of the cuff inflating may make the infant cry. Although the American Academy of Pediatrics does not recommend universal screening of BP, systemic BP should be measured in every neonate with suspected CHD, renal disease, or clinical signs of hypotension.[5,24]

A cuff of the proper size must be used when obtaining a BP measurement. Indeed, the most frequent reason for a hypertensive BP is the use of a BP cuff that is too small for the infant. Using limb length as the sole criterion for establishing cuff size for BP monitoring can be misleading. This method does not take into consideration large-for-gestational-age infants with excessive subcutaneous fat. These infants will show falsely elevated BPs if cuff size is inappropriate. It is therefore recommended that the cuff width (not the cuff length) be 40% to 50% of the circumference of the extremity, equal to 125% or 155% of the diameter of the limb being measured (Figure 7.9).[5] In addition, the cuff should be long enough to entirely encircle the extremity.[6]

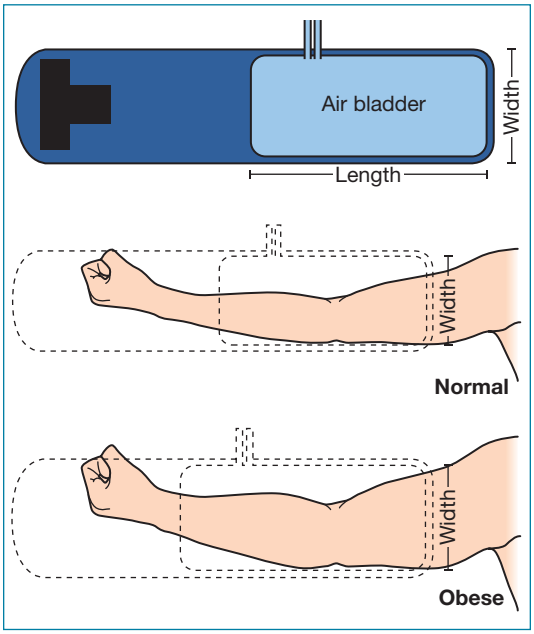

Figure 7.9 Diagram showing how to select accurate blood pressure cuff size

Source: From Park MK. 2014. *Park's Pediatric Cardiology for Practioners*, 6th ed. Philadelphia, PA: Saunders, 19. Reprinted by permssion.[5]

Monitoring Methods

There are several methods for monitoring systemic BP in neonates: flush, palpation, ultrasound Doppler, and oscillometric measurements. BP obtained by auscultation is not appropriate for neonates.

Flush

The flush method, now rarely used, is simple to do and can be useful in certain situations. With this method, the hand or foot is squeezed to blanch it, and the BP cuff is rapidly inflated. The pressure in the cuff is released, and the pressure point at which the extremity suddenly flushes is documented. The pressure obtained by this method represents the mean arterial pressure.

Palpation

BP can also be obtained by palpatory methods in infants. While releasing the pressure in the cuff, the examiner palpates the pulse distal to the cuff. The pressure point at which the pulse reappears upon deflation approximates the systolic BP.

Doppler Ultrasound

A more accurate method, similar to the palpatory method of BP measurement, is done by Doppler ultrasound. Although an ultrasound can transmit images, a Doppler ultrasound can be used to measure blood flow. A transducer is placed over the artery distal to the BP cuff after a conductive gel is applied, and an audible pulse is listened for as the BP cuff is deflated. The pressure point at which the pulse is heard is documented. This method also approximates the systolic BP.

Oscillometric Measurement

Most centers now use oscillometric methods to document BP in neonates. Systolic, diastolic, and mean arterial BP and heart rate are all digitally recorded using special cuffs and oscillometric systems. These systems have been found to be fairly reliable.

Catheter

Finally, indwelling arterial catheters also provide systolic, diastolic, and mean BPs. These catheters provide minute-by-minute readings of BP values. Accuracy of this type of monitoring requires knowledge of the system and the variables that will affect it. Some of these variables include transducer position, air bubbles in the system, and equipment calibration.

Normal Values

Normal BP values in newborns vary, depending on body weight and postnatal age. In the first few hours of life, BP can be significantly affected by type of delivery, birth asphyxia, and placental transfusion. The newborn's initial BP decreases during the first 3 to 4 hours of life, presumably because of fluid shifts into and out of the vascular space.[21] The systolic pressure reaches a minimum at 3 to 4 hours of age and then gradually increases to reach

a plateau at about 4 to 6 days of age to a level closer to the initial postpartum level.[25] BPs in neonates are also affected by activity, temperature, and behavioral state. Figure 7.10 documents confidence limits for newborns (based on birth weight) during the first day of life.[26] Whenever there is any question of CHD, if an examiner has difficulty obtaining BPs, if a murmur is heard, or if there is an absence of femoral pulses on the physical examination, four extremity BPs should be obtained. Pressures obtained in the legs are often slightly higher than those in the arms, but they can be equal or slightly lower in the neonate. A systolic blood pressure in the upper extremities that is significantly higher than that in the lower extremities, especially with the combination of absent or weak femoral pulses, should increase suspicion of coarctation of the aorta.[27] This pressure difference can sometimes be masked in the left arm by a PDA that allows blood to pass around the restricted area; therefore, the right arm will yield the most valuable information because it is always preductal.

In addition to systolic, diastolic, and mean BP readings, the pulse pressure can provide additional valuable information. The *pulse pressure* is defined as the difference between systolic and diastolic BPs. Averages for term neonates are between 25 and 30 mmHg; for premature infants, they are between 15 and 25 mmHg.[21] Wide pulse pressures may be a sign of a large aortic runoff, as seen with PDA. Narrow pulse pressures are documented in neonates with peripheral vasoconstriction, heart failure, or low cardiac output.

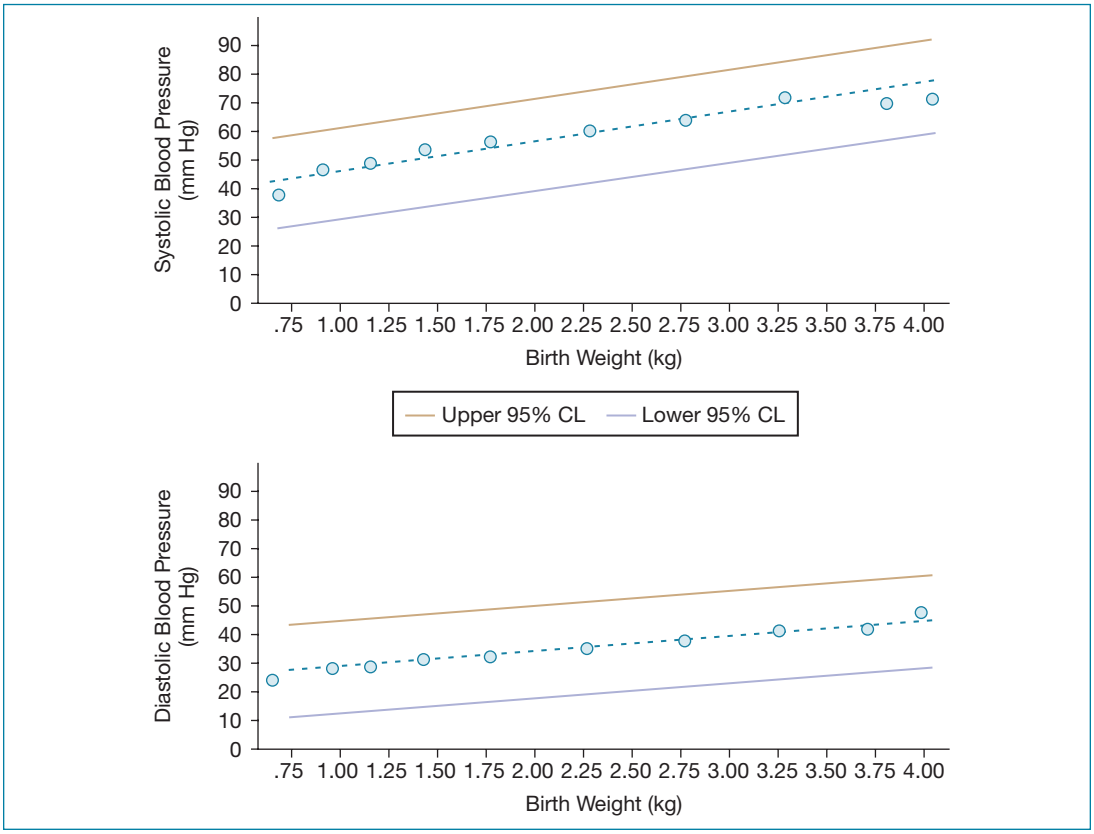

Figure 7.10 Systolic, diastolic, mean, and pulse pressures for newborns (based on birth weight) during the first 12 hours of life

CL, confidence limit.

Source: Redrawn from Zubrow AB, Kushner H, and Falkner B. 1995. Determinants of blood pressure in infants admitted to neonatal intensive care units: A prospective multicenter study. *Journal of Perinatology* 15(6): 472. Reprinted by permssion.[26]

SCREENING FOR CHD WITH PULSE OXIMETRY

Evidence now suggests that screening to detect CHD early, through the use of pulse oximetry during the immediate postnatal period, is an effective method for reducing mortality and morbidity in infants.[28] A work group that included the American Academy of Pediatrics and the American Heart Association has made recommendations for a standardized approach to screening and diagnostic follow-up of all infants in the first 48 hours of life (Figure 7.11).[28] In these recommendations, oxygen saturation is measured using a pulse oximeter probe on the right hand and one foot either in direct sequence or in parallel in infants between 24 and 48 hours of age. The screen is considered positive if any of the following occur:

- Any oxygen saturation reading is less than 90%.
- The oxygen saturation is less than 95% in both extremities on three measures (each measure should be separated by 1 hour).

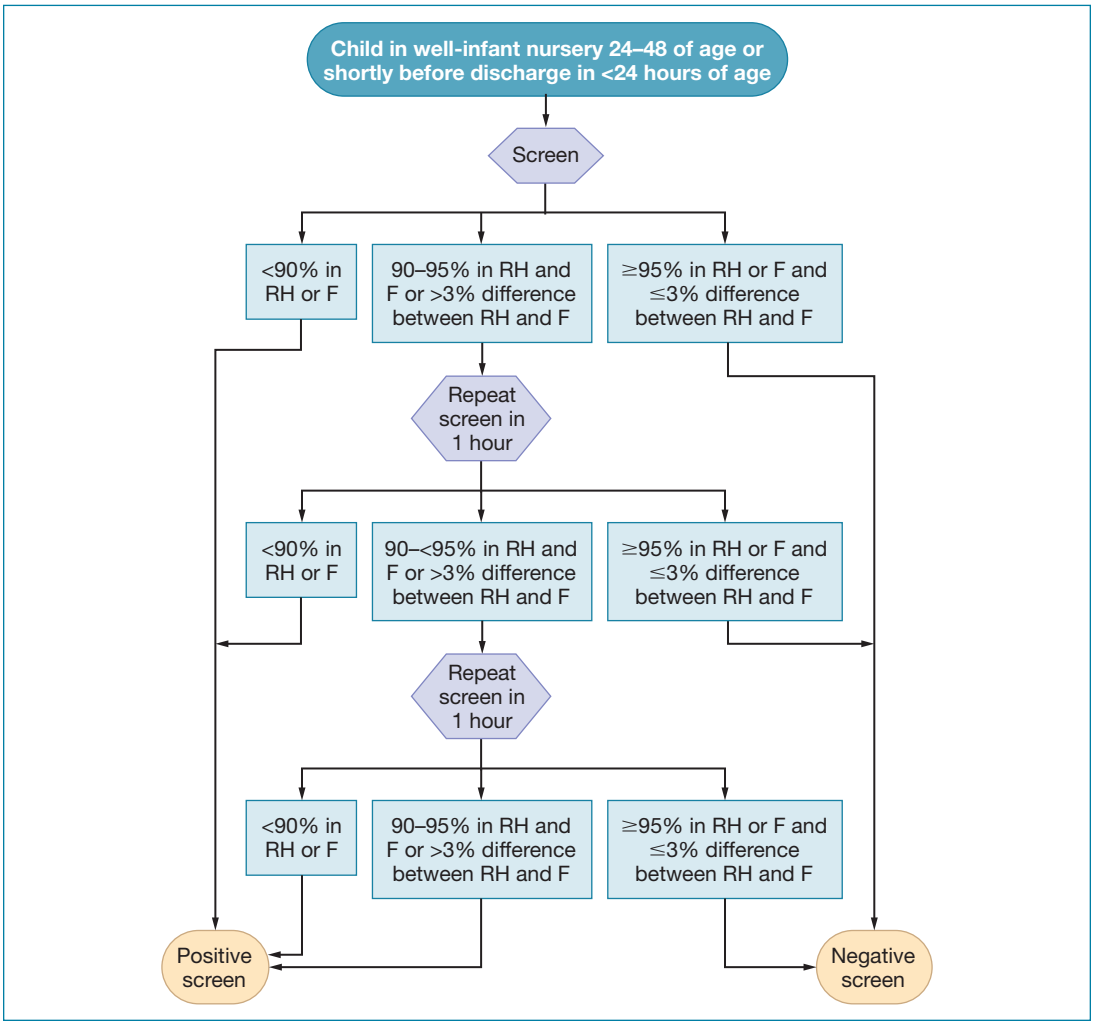

Figure 7.11 The proposed pulse-oximetry monitoring protocol based on results from the right hand and either foot

F, foot; RH, right hand.

Source: Adapted from Kemper AR, et al. 2011. Strategies for implementing screening for critical congenital heart disease. *Pediatrics* 128(5): e1259. Reprinted by permission.[28]

- There is a greater than 3% difference in saturation levels between the foot and right hand on three different measures separated by 1 hour.

Any newborn with a positive screen should have a comprehensive evaluation for the cause of the hypoxemia, and CHD should be excluded based on the results of an echocardiogram.[28]

SUMMARY

Properly executed, the cardiovascular assessment provides a great deal of information about the overall health of the newborn, as well as valuable information about congenital heart defects that might be present. Time, patience, and experience with inspection, palpation, and auscultation are all necessary to develop the skills essential to a thorough cardiac examination.

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Abdominal Assessment

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8

Assessment of the newborn abdomen is best performed during the first few hours of life, when the bowel is not yet filled with gas. The ideal time to perform the examination is when the infant is in a quiet state and the abdominal muscles are relaxed. Techniques used include observation, auscultation, and palpation. Although percussion is used to examine the adult abdomen, it is of limited usefulness when examining the newborn. Common variations and abnormal findings in the newborn abdominal assessment are addressed as the examination techniques are discussed.

REVIEW OF PREGNANCY AND DELIVERY HISTORY

Evaluation of the maternal history is discussed in detail in Chapter 2; however, specific findings are of interest when examining the abdomen and gastrointestinal (GI) tract. Any antenatal ultrasound finding of abnormalities, such as enlarged kidneys, dilated bowel, or unusual masses, will alert the examiner to evaluate thoroughly these areas on the initial examination. Ultrasound techniques, including Doppler flow studies and three-dimensional exams, are able to identify abnormaliaties that would have remained undetected even a decade ago. Bowel obstructions, abdominal wall defects, and abnormalities of the genitourinary (GU) tract are now readily identifiable in fetal life.[1,2] *Polyhydramnios*, defined as an amniotic fluid volume of greater than 2,000 mL occurs in 1% to 3% of pregnancies. The finding of polyhydramnios must be evaluated as the risk for anomaly increases with greater amounts of amniotic fluid. The prevalence of severe congenital abnormalities is as high as 75% in cases of severe polyhydramnios. Associated fetal abnormalities include duodenal or esophageal atresia, tracheoesophageal fistula, gastroschisis, omphalocele, and diaphragmatic hernia.[2,3] The history of the events surrounding delivery should be reviewed as well. The finding of an extremely large amount of amniotic fluid in the stomach at delivery may indicate the presence of duodenal atresia. The presence of copious oral secretions coupled with the inability to pass a soft catheter to the stomach may indicate an esophageal atresia, usually associated with a tracheoesophageal fistula. The presence of bilious gastric secretions at delivery is also abnormal and should alert the examiner to the possibility of intestinal obstruction.

INSPECTION

Skin

The skin of the abdomen should be pink. Newborn rash may be present, and there may be bruising from the process of delivery, although this is rare. A few large veins may be visible, especially on a light-skinned infant, but marked venous distention should not be present. The normal preterm infant will have more visible vasculature because of decreased subcutaneous tissue. The postmature baby may have superficial cracking and peeling of the skin over the abdomen.

Shape and Movement

The abdomen of the term newborn is soft and rounded, with easy movements associated with respirations. The abdominal and diaphragmatic muscles work together during respiration, causing the chest and abdomen to move parallel with each other. Asynchronous movements (the chest and abdomen moving in opposition to each other) can indicate respiratory distress.

The abdomen can vary from nearly flat to slightly distended, depending on how recently the infant has been fed and whether air has been swallowed while crying. The abdominal circumference measured at the greatest diameter (just above the umbilicus) is normally less than the head circumference up until 30 to 32 weeks gestational age. From 32 to 36 weeks gestational age, the abdominal and head circumferences will be equal. After 36 weeks gestational age, the abdominal circumference will be greater than the head circumference.[4]

A sunken, or scaphoid, abdomen can indicate a diaphragmatic hernia due to displacement of abdominal contents up into the chest. The abdomen of a normal preterm infant may appear distended because of lack of muscle tone; a term infant may have decreased muscle tone related to maternal medications received during labor. This is easily distinguished from the flaccid, lumpy abdomen of the infant with congenital absence of the abdominal musculature, also called *Eagle–Barrett syndrome* (Figure 8.1). This very rare syndrome, discussed further in Chapter 9, occurs mostly in males and is associated with severe renal and urinary tract abnormalities.[5] A markedly distended abdomen in any infant warrants further investigation. Bowel obstruction presents with distention and vomiting, the timing and character of which vary, depending on the location of the obstruction. Isolated distention of the lower abdomen may be associated with bladder distention, GU abnormalities, anomalies of the female reproductive tract, or teratomas.

Obstructions of the upper GI tract, such as esophageal and duodenal atresia, tend to present with excessive salivation and nonbilious vomiting. As the level of obstruction progresses down the intestinal tract, symptoms shift to abdominal distention and bilious vomiting. Bilious vomiting is the result of obstruction beyond the level of the ampulla of Vater, where bile from the gallbladder enters the small intestine.[6] Meconium ileus is the only bowel obstruction that can present with abdominal distention at birth. It is the result of abnormal pancreatic enzyme function seen with cystic fibrosis.[6] Abdominal distention otherwise presents only after the infant has swallowed air to fill the bowel.

The presence of bilious emesis in any infant, with or without abdominal distention, requires immediate investigation to rule out a malrotation with midgut volvulus. A malrotation results from abnormal fixation of the intestine in the abdomen; midgut volvulus is the abnormal rotation of the bowel around the mesentery and subsequent obstruction of

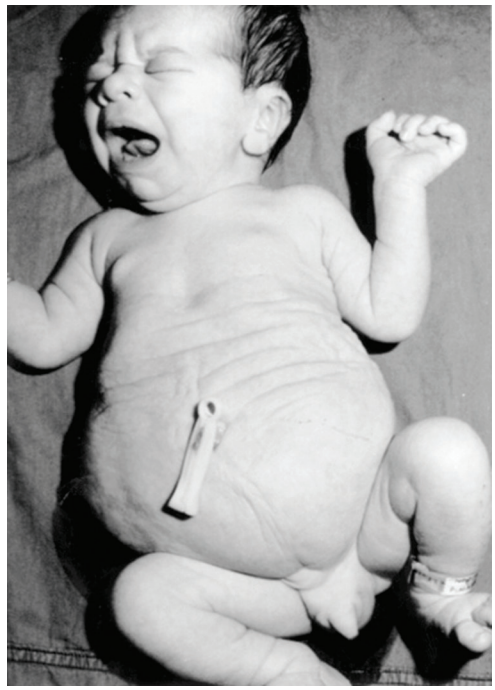

Figure 8.1 Eagle–Barrett syndrome—congenital deficiency of the abdominal musculature, showing typical "prune" (wrinkled) belly appearance

Courtesy of Dr. David A. Clark, Albany Medical Center, Albany, NY.

blood flow to the bowel. This is one of the most urgent emergencies in neonatal surgery. *Immediate intervention is required to avoid irreversible infarction of the intestine*.[3]

The shape of the abdomen should be symmetric, without obvious swellings or depressions. Occasionally, a loop of bowel may be visible beneath the abdominal skin. This is more common in the preterm infant and is of concern only if it remains fixed in one spot or is associated with generalized distention and symptoms of bowel obstruction. Diastasis recti is a midline separation of the rectus abdominis muscles and can be seen as a midline, elevated ridge extending from below the sternum to the umbilicus when the infant is crying. This is a normal finding and will resolve without intervention.

An umbilical hernia may also be seen on examination. It is a common finding in 30% of term African American infants and is also seen in low-birth-weight males. An umbilical hernia is often seen in hypothyroidism, a relatively rare condition. There is protrusion of abdominal contents into the hernia, which is soft and easily reducible (Figure 8.2). Erythema is not associated with umbilical hernia. Umbilical hernias usually close spontaneously by 2 years of age.[7]

An epigastric hernia is a small, firm, palpable nodule seen between the umbilicus and the xiphoid process. It results from fat protruding through a small opening in the muscle. Surgical intervention is necessary for this uncommon hernia.

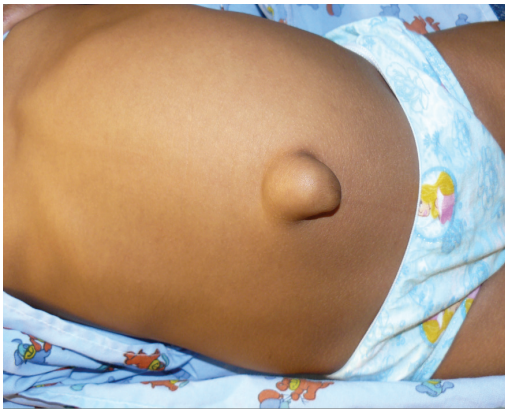

Figure 8.2 Umbilical hernia

Source: From Children's Mercy Hospital, Department of Surgery, Kansas City, MO.

Umbilical Cord

The umbilical cord of the healthy newborn is shiny, pearly white, and gelatinous. It contains two arteries and one vein. The average size is 1.5 to 2 cm at the base. The umbilical cord contains Wharton's jelly, which protects the vessels; it also serves as an indicator of the infant's nutritional status. A thick cord is often seen in large-for-gestational-age infants, whereas a small, thin cord is often seen in infants who are small for gestational age, postmature, or affected by placental insufficiency. Any unusual bulging or herniation in the cord requires further investigation and could indicate the presence of a small omphalocele.

If abnormalities are noted, the cord should be clamped and cut distal to the areas to avoid damage to any structures, such as bowel or bladder, that might be present within the cord. When the cord is cut, the vessels should be visible. The paired arteries are small, thick walled, and constricted. The vein is large, thin walled, and open. The absence of one of the arteries is seen in 1% of infants. It is associated with fetal abnormalities of the cardiovascular, GI, or GU systems or may be an isolated finding in an otherwise normal baby.[8]

The cord is normally white. A yellow or green cord may be seen when meconium staining has occurred 6 to 12 hours prior to delivery. Green color may rarely be an indicator of infection. Redness encircling the cord and extending onto the abdomen can be a sign of omphalitis (infection of the umbilical cord). Although the incidence is low, less than 1% in developed countries, it may be as much as 6 times greater in home births. Omphalitis must be treated promptly and properly because it can spread rapidly to underlying structures, causing severe systemic disease and even death.[9]

The cut surface of the cord may initially ooze a small amount of clear, sticky fluid, but

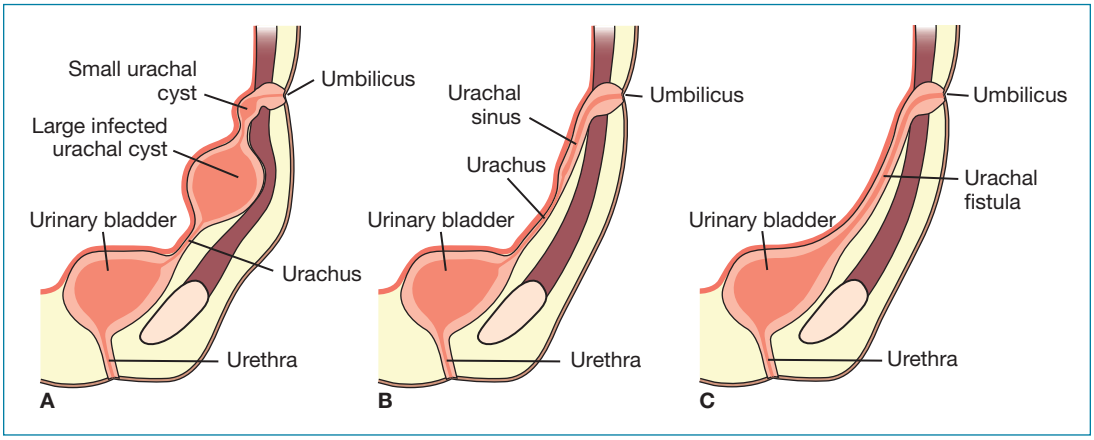

Figure 8.3 Malformations of the urachus

Source: From Moore KL, Persaud TVN, and Torchia MG. 2013. *The Developing Human: Clinically Oriented Embryology*, 9th ed. Philadelphia, PA: Saunders, 261. Reprinted by permission.[10]

it will dry quickly. In normal situations, the cord will begin to dry soon after birth and will shrivel and fall off within 10 to 14 days.

Purulent drainage from the umbilical cord may indicate the presence of an abscess. Excessive amounts of clear drainage may indicate the presence of a patent urachus (persistence of an embryologic connection from the bladder to the umbilicus; Figure 8.3). Persistence of the embryologic tract connecting the ileum to the umbilicus is called an *omphalomesenteric duct*. An infant with this defect will have leakage of ileal contents through the umbilical cord. A small, red, raw-appearing granuloma will occasionally form at the site of the separation of the umbilical cord; this is sometimes referred to as an *umbilical polyp*.

Abdominal-Wall Defects

Defects in formation of the abdominal wall may occur during embryonic life, resulting in abnormal externalization of the abdominal contents. Three such defects are omphalocele, gastroschisis, and exstrophy of the bladder.

Omphalocele

An omphalocele is the herniation of abdominal contents into the umbilical cord (Figure 8.4).

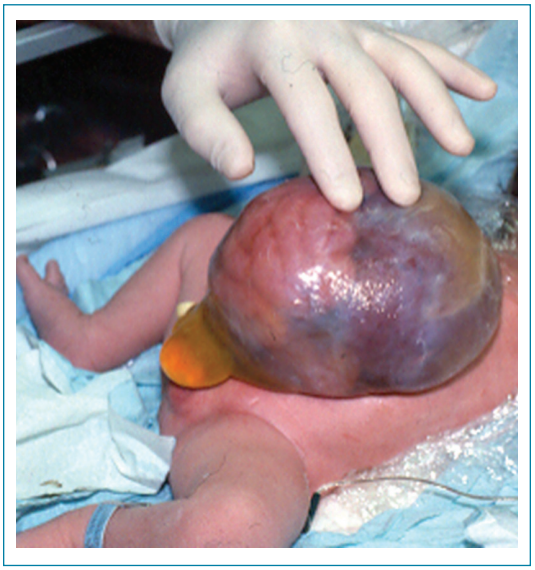

Figure 8.4 Omphalocele

Source: From Children's Mercy Hospital, Department of Surgery, Kansas, City, MO.

The hernia is contained in a translucent sac that is contiguous with the umbilical cord. The sac may rupture at or before delivery, so careful examination is necessary (Figure 8.5). The incidence of omphalocele is 2 to 2.5 per 10,000 live births. Omphalocele is thought to result from failure of the bowel to reenter the abdominal cavity after its normal extrusion into the cord before the 10th week of gestation. When this defect occurs above the

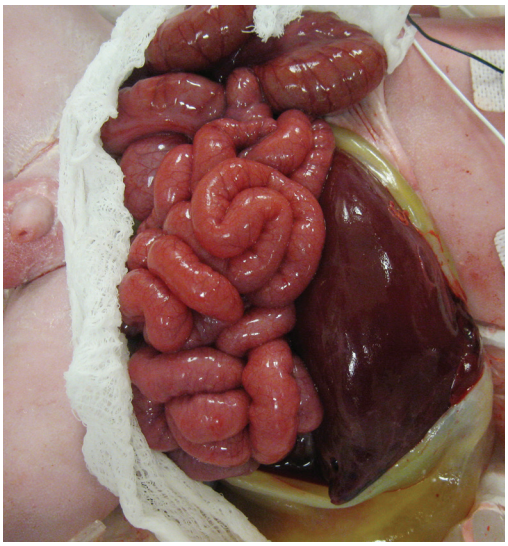

Figure 8.5 Ruptured omphalocele

Source: From Children's Mercy Hospital, Department of Surgery, Kansas City, MO.

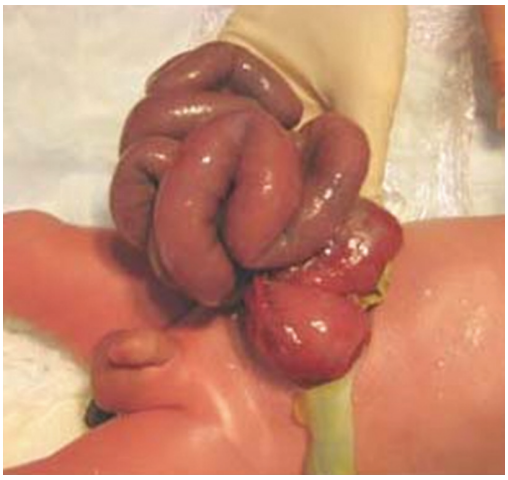

Figure 8.6 Gastroschisis

Source: From Children's Mercy Hospital, Department of Surgery, Kansas City, MO.

umbilical area, it can produce the pentalogy of Cantrell, which involves sternal, diaphragmatic, and pericardial defects, including ectopia cordis (externalization of the heart). This failure results from faulty migration and fusion of embryonic tissues and is associated with cardiac, neurologic, GU, skeletal, and chromosomal abnormalities in 50% to 75% of infants.[3] Omphalocele is frequently associated with Beckwith–Wiedemann syndrome and with trisomies 13, 18, and 21.[3,11]

Gastroschisis

Gastroschisis is a defect in the abdominal wall through which the viscera protrude. There is no sac covering this defect, and the umbilical cord is discrete from it. The gastroschisis is usually to the right of midline (Figure 8.6). The etiology of gastroschisis is suggested to be a failure in the normal attachment between the umbilical cord and umbilical ring in embryonic life. Other theories include vascular accident interfering with normal formation of abdominal musculature or failure of mesenchymal migration.[3] The incidence of gastroschisis is approximately 1.5 per 10,000 live births. There is a 5% to 20% incidence of associated anomalies with gastroschisis, far less than with omphalocele, although atresias of the bowel and ischemic enteritis can result from inadequate mesenteric blood flow. Both gastroschisis and omphalocele vary greatly in size, from smaller than a golf ball to so large that all of the abdominal contents (including the liver) are externalized.

Exstrophy of the Bladder

Exstrophy of the bladder (Figure 8.7) is a very rare defect. It is a malformation sequence resulting from lack of normal formation of the lower abdominal wall early in gestation. The posterior wall of the bladder is exposed, and urine drains onto the abdomen. Exstrophy of the bladder is more common in males, and there may be associated urogenital abnormalities in both males and females.[11] This condition is discussed further in Chapter 9.

Perianal Area

The perianal area should be inspected for presence and placement of an anus, for anal sphincter tone, and for abnormalities such as fistulas. Sphincter tone can be assessed by

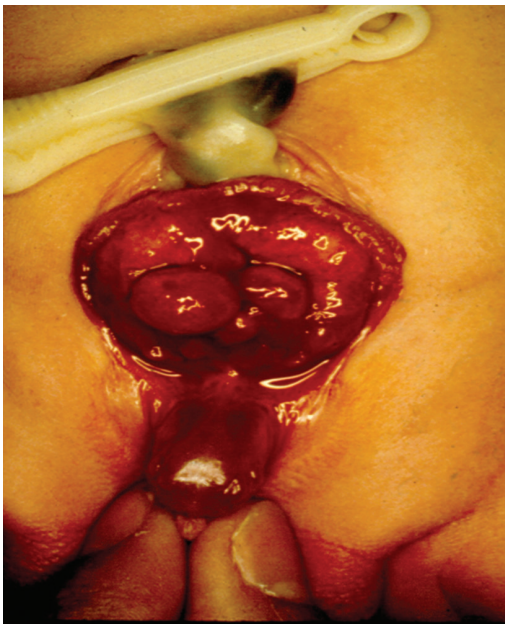

Figure 8.7 Exstrophy of the urinary bladder

Source: From Children's Mercy Hospital, Department of Surgery, Kansas City, MO.

gently stroking the anal area. An anal wink will occur. Absence of an anal wink suggests an abnormality of the central nervous system.

If the anus is completely absent, or imperforate, this may be evident immediately (Figure 8.8). However, atresia and stenosis can occur at any level of the anorectal canal, so patency of the anus cannot be established until the passage of meconium. Digital examination of the rectum or the insertion of instruments is not recommended because of the risk of damage to the anal canal. Passage of stool usually occurs within 24 hours of birth, often within 12 hours, but a normal infant may not stool until 48 hours of life. In otherwise asymptomatic infants, investigation is not warranted until this point. Continued absence of stool suggests anal atresia. Passage of very-small-caliber stools suggests stenosis.

A fistula is an anomalous connection between the intestinal tract and the GU tract. Presence of meconium in the vagina suggests a rectovaginal fistula (Figure 8.9). Meconium in the urethral orifice indicates a rectourethral fistula.

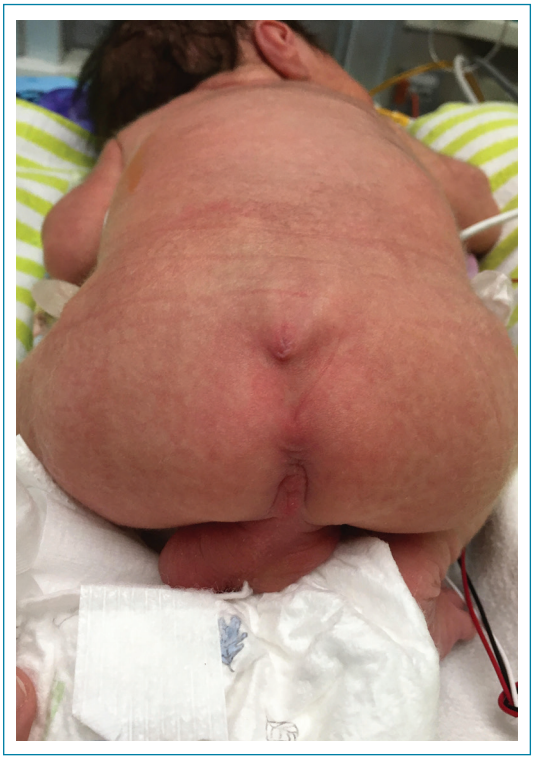

Figure 8.8 Imperforate anus

Source: From Children's Mercy Hospital, Department of Surgery, Kansas City, MO.

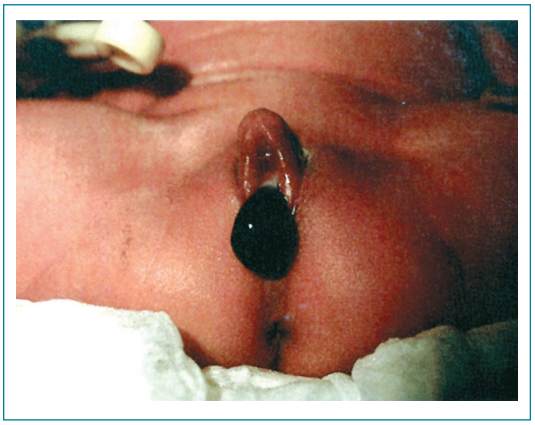

Figure 8.9 Rectovaginal fistula

Source: From Clark DA. 2000. *Atlas of Neonatology.* Philadelphia, PA: Saunders, 173. Reprinted by permission.[12]

AUSCULTATION

Auscultation should be performed before palpation because palpation can interfere with normal bowel sounds as well as cause

the infant to become agitated. Auscultation should be performed with a warmed stethoscope while the infant is quiet. The examiner should patiently listen to all four quadrants of the abdomen.

Bowel sounds will be audible beginning about 15 minutes after birth, although they are relatively quiet until feedings have begun. Their presence, quality, and intensity should be noted. Normal bowel sounds have a metallic, tinkling quality and are heard every 15 to 20 seconds. If bowel sounds are infrequent, the examiner should listen for a full 5 minutes before diagnosing them as absent. This part of the examination must be correlated with other clinical findings. Hyperactive bowel sounds in a healthy-appearing neonate who has just fed may be normal. The same finding in an ill-appearing infant with a distended abdomen is not normal and could indicate obstruction. Maternal sedation can result in hypoactive bowel sounds.[4]

Breath sounds may be audible when the upper abdomen is auscultated. This is a normal finding because sounds transmit easily throughout an infant's body. Vascular sounds are not normally auscultated over the abdomen of the term infant. Rarely, a bruit will be heard on abdominal exam. A *bruit* is the sound of blood flow through a restricted or tortuous vessel. It is heard as a swooshing sound similar to a cardiac murmur. Bruits may be heard anywhere over the abdomen and can indicate malformations of hepatic and renal vessels or hemangiomas.

PALPATION

Palpation can be performed with the infant in any state, but it is easiest when the infant is quiet. Palpation is the last step in the abdominal examination because it often results in agitation. If the infant is fussy, flexing the hips to relax the abdominal muscles during the examination may help.

The skin of the abdomen should be warm and pink, with brisk capillary refill. Muscle tone should be assessed at this point in the examination. Hypertonicity of the abdominal musculature may indicate pain or peritoneal irritation. Hypotonicity may be present in neuromuscular disease, perinatal depression, or if the mother has taken medications that cause neonatal depression. It can also be a normal finding in the extremely premature infant.

Organs

Palpation should begin with the superficial structures and then progress to the deeper organs (Figure 8.10). The shape of the thorax in the newborn is such that the upper abdominal organs are not thoroughly covered by the anterior rib cage and can be easily palpated. The liver is a superficial organ and should be identified first. The normal newborn liver occupies a wide area of the upper abdomen, extending well into the left upper quadrant. It extends 1 to 2 cm below the right costal margin. To palpate the liver, the examiner should begin just above the iliac crest on the right. Using the palmar surface of the fingers parallel to the costal margin,

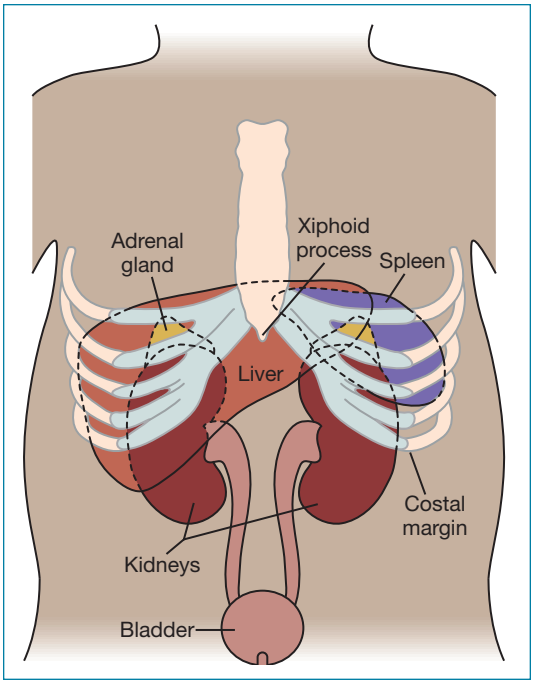

Figure 8.10 Abdominal organs of the infant

the examiner gently palpates in a progressively caudal fashion. The abdomen should be depressed 1 to 2 cm. Care should be taken not to lift the hands completely off the abdomen because this may result in missing the edge of an enlarged liver. The liver edge is normally smooth, firm, and sharp. A boggy liver is a sign of congestion and may indicate congestive heart failure. A hard or nodular liver edge is also abnormal.[4]

Using the technique just described, the left side of the abdomen is examined for the presence of the spleen. The tip of the spleen may be felt, but in many cases, it is not palpable at all. A spleen that is palpable more than 1 cm below the costal margin may be enlarged and warrants further investigation. No spleen will be found in those infants with certain cardiac conditions involving asplenia.

Rarely, the liver will be found on the left and the spleen on the right. This condition is known as *situs inversus*. Complete situs inversus is reversal of position of all the thoracic and abdominal organs, a "mirror image" of normal positioning. Partial situs inversus is the reversal of the positions of some but not all of the abdominal organs. Both types of situs inversus may be associated with cardiac defects, but such defects are more common in partial situs inversus.[13]

Palpation of the kidneys can be difficult, but it is easiest when done early, before the infant has eaten or begun prolonged crying. The kidneys are found using deep palpation, at a 45° angle caudal and lateral to the umbilicus. This can be most easily accomplished with one hand under the infant's back and the other pressing downward on the abdomen (see Figure 8.10). The normal newborn kidney is 4.5 to 5 cm from the upper to lower pole. On examination, the kidneys have the size and consistency of a large ripe olive. The right kidney is normally situated slightly lower than the left. A very easily palpated kidney is usually enlarged and may indicate hydronephrosis or a cystic kidney. Any texture other than smooth and firm is abnormal and should be investigated further as well.

The bladder is situated from 1 to 4 cm above the symphysis pubis. Palpation of the bladder should begin at the level of the umbilicus and progress downward until the smooth upper aspect of the bladder is felt. The bladder may also be percussed. A dull sound indicates the presence of urine in the bladder. Continuous bladder distention is abnormal and may indicate a urinary tract obstruction or central nervous system abnormality.

Masses

Once the abdominal organs have been examined, the entire abdomen should be palpated for the presence of masses. In a systematic fashion, the examiner should explore each quadrant using first light and then deeper palpation. Normal findings include stool in the colon, felt as a sausage shape in the right and left lower quadrants. Gaseous distention may also be palpated in an infant who has swallowed air. Serial examinations will differentiate between normal stool in the colon that progresses and an abnormal mass that is fixed. A firm, oval-shaped mass may be felt in the upper midabdomen with pyloric stenosis, although this does not usually present in the newborn period. Most abdominal

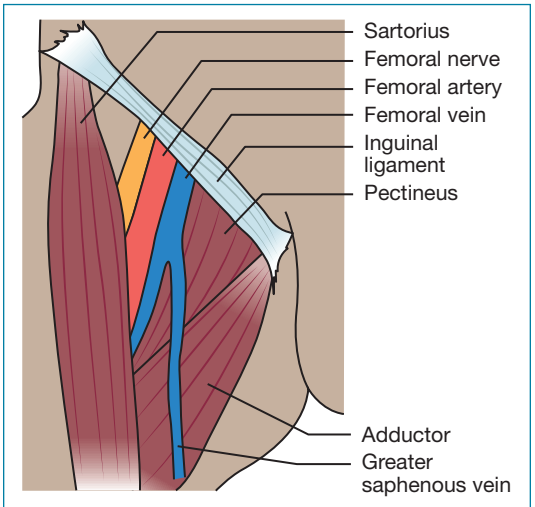

Figure 8.11 The femoral triangle

Source: Adapted from Plaxico DT, and Bucciarelli RL. 1978. Greater saphenous vein venipuncture in the neonate. *Journal of Pediatrics* 93(6): 1025. Reprinted by permission from Elsevier.[14]

masses in the newborn involve the urinary tract and are cystic or solid on examination. Neoplasia is very rare in the newborn.

Groin and Femoral Area

As the final step in the examination, the groin and femoral regions should be inspected and palpated (Figure 8.11). Ideally, this should be done both while the infant is quiet and while he or she is crying. The groins are normally flat. A visible femoral pulse may be present in a preterm infant or a thin, term infant. The femoral artery pulses must be palpated and their character evaluated. The pulses should be compared bilaterally and also compared with the brachial pulse. Any difference in quality should be noted. An absent or weak femoral pulse may be noted with coarctation of the aorta or an interrupted aortic arch. Full and bounding femoral pulses may indicate a patent ductus arteriosus. When a pulse discrepancy is discovered, blood pressure readings should be obtained in the upper and lower extremities. A difference of 20 mmHg or greater should be investigated further. Complete evaluation of pulses is discussed in detail in Chapter 7. Pulse oximetry screening of upper and lower extremities for detection of congenital cardiac disease is now recommended for all infants prior to hospital discharge.[15]

The groin should also be observed for bulges in the inguinal and femoral areas. An inguinal hernia is a defect in the muscle wall that allows the intestine to slip into the scrotum in males and into the soft tissue in females (Figure 8.12). A bulge in the labia majora may be a hernia or an abnormal gonad. The presence of a hernia is most easily evaluated when the infant is crying because intra-abdominal pressure increases. Inguinal hernias are more common in males than in females, and they are very common in the extremely preterm male. The hernia should be soft and reducible into the body cavity without difficulty. Surgical repair is required for inguinal hernias, in most cases surgery is deferred to allow for growth of the infant. It is done emergently if any evidence of strangulation is noted.

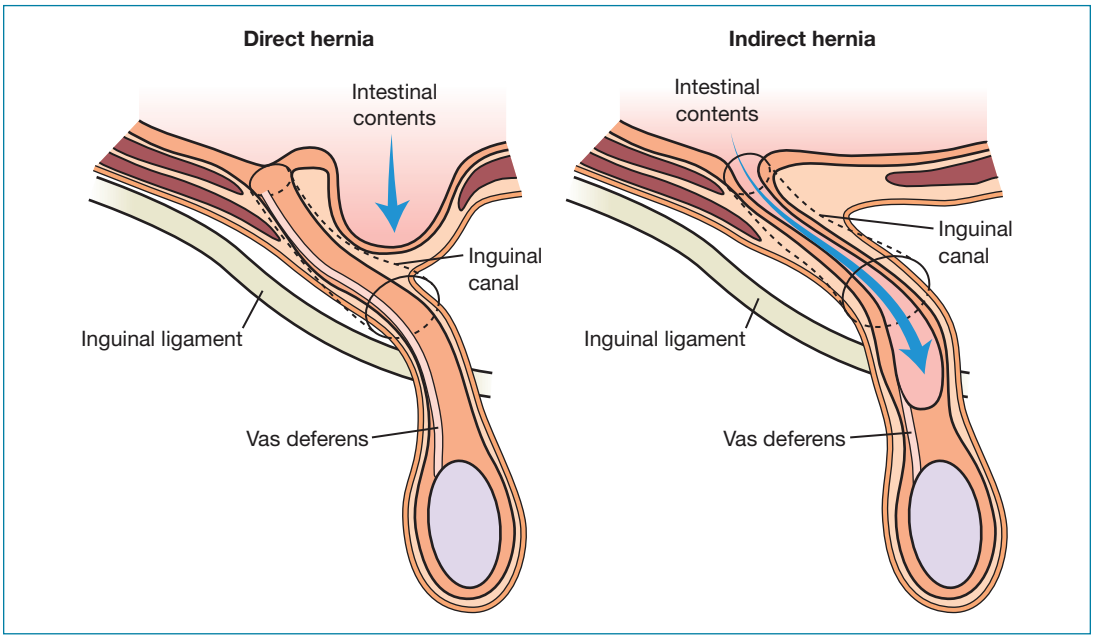

Figure 8.12 Inguinal hernia

Source: From Alexander MM, and Brown MS. 1979. *Pediatric History Taking and Physical Diagnosis for Nurses*, 2nd ed. New York, NY: McGraw-Hill, 223. Reprinted by permission of the authors.[16]

A small bulge adjacent and medial to the femoral artery may be a femoral hernia. Although it is an uncommon finding in an infant, it is seen more often in females than in males.

SUMMARY

The examination of the abdomen involves observation, auscultation, and palpation. Following a systematic routine and taking measures to ensure a quiet and cooperative infant will facilitate the exam. With practice, the examiner will learn to differentiate normal variations from abnormal findings, permitting timely intervention when abnormalities are found.

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Genitourinary Assessment

Terri A. Cavaliere, DNP, RN, NNP-BC

9

A comprehensive physical assessment of the newborn includes evaluation of the genitourinary (GU) system, which consists of kidneys, urinary tract, and reproductive tract. These organs are closely related both anatomically and embryologically. This chapter focuses on examination of the GU system in the neonate using techniques of inspection, palpation, and occasionally, percussion. Normal newborn characteristics and simple variations from normal are presented first, followed by a discussion of abnormalities and malformations.

HISTORY REVIEW

A review of the prenatal history is part of any newborn physical examination. A history of polyhydramnios or oligohydramnios during pregnancy should alert the examiner to the possibility of abnormalities of the GU tract or renal impairment.[1,2] Antenatal sonography often assists in diagnosis of disorders before delivery or onset of signs and symptoms in the newborn. For example, variations in the amount of amniotic fluid and structural abnormalities may have been detected by in utero ultrasound. Family history is an important consideration as certain GU anomalies have a genetic predisposition.[2,3] Parents of newborns with GU disorders should be questioned as to the occurrence of anomalies in family members. Genetic counseling may be appropriate in some instances.

Related Findings

Even without a documented history of oligohydramnios, physical signs of intrauterine compression in the newborn, such as flattened facies, malformed ears, and contraction deformities of the limbs (also known as *oligohydramnios sequence*; Figure 9.1), suggest urogenital defects. Pulmonary hypoplasia, presenting as respiratory distress, may occur secondary to oligohydramnios or limited movement of the diaphragm caused by an intra-abdominal mass.[1,2]

There is a well-established association between GU anomalies and those of other systems. Careful evaluation of the GU system is warranted in newborns with other congenital anomalies such as myelomeningocele, complex congenital heart disease, and VACTERL (**v**ertebral anomalies, **a**nal atresia, **c**ardiac abnormalities, **t**racheo**e**sophageal abnormalities, **r**enal abnormalities, and **l**imb anomalies) association.

The presence of oliguria and anuria may indicate underlying urologic disease. Both full-term and premature newborns may void at delivery. As this event often goes unnoticed or undocumented the exact percentage is difficult to estimate, however 98% of babies void in the first 30 hours of life. Lack of urine output in the first 48 hours of life in an *otherwise healthy* newborn should not be cause for alarm. However, diagnostic evaluation and/or intervention are necessary if there are any signs of illness or abnormalities (palpable bladder, abdominal mass, renal disease) or if more than 48 hours pass without urination.[1] Anuria or oliguria that develops after the first 48 hours of life can also be a sign of malformation or obstruction of the urinary tract.

Urine output is normally low during the first 2 days of life: full-term newborns may

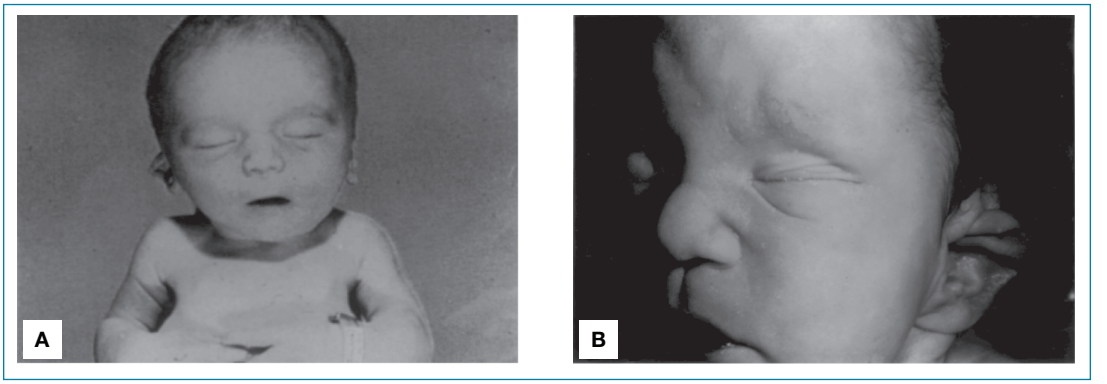

Figure 9.1 Compression effects of oligohydramnios, which may signal possible GU or renal abnormalities. (A) Joint contractures,narrow thorax, malformed ear. (B) Typical facies; flattened nose, epicanthal fold, furrowed brow.

Courtesy of J. Hernandez, MD, The Children's Hospital, Denver, CO.

void one to four times, whereas a preterm newborn may void more frequently. The volume of urine production rises as intake increases.[4] Documentation of adequate urinary output is important, especially with early hospital discharge of newborns. Parent counseling and adequate follow-up care are essential.

NORMAL PHYSICAL EXAMINATION

Abdomen

A comprehensive abdominal examination is described in Chapter 8. Those details pertinent to the GU system are presented here also. On inspection, the abdomen of a term newborn is rounded and symmetric, with smooth, opaque skin. A preterm newborn has a more protuberant abdomen because of immature muscle development, and the skin covering the abdomen may be thinner, with more prominent blood vessels. The umbilical cord should be evaluated for appearance, length, diameter, number of vessels, and insertion site. At birth, the umbilical cord is gelatinous and bluish white in color and contains three vessels: two arteries and one vein. There should be no exudate or discharge from the umbilicus. At term, the average cord length is 55 cm, with an average diameter of 2 cm.[5]

Cord length and appearance can provide clues to abnormalities. For example, abnormally short cords can be accompanied by conditions, such as "fetal hypotonia, oligohydramnios, and uterine constraint, and with increased risk for complications of labor and delivery for both mother and infant" (p. 890).[6] Long cords (>70 cm) have higher risks for forming knots, entangling and wrapping about fetal body parts (neck, arm), and/or prolapse. Straight untwisted cords are associated with fetal distress, anomalies, and intrauterine fetal demise.[6]

Palpation of the abdomen is easiest in the first 24 to 48 hours of life, before air fills the entire gastrointestinal tract and before abdominal tone increases. All four quadrants should be examined to detect any masses. Palpation is best performed on a quiet newborn lying in a supine position with knees and hips maintained in flexion by the examiner's hand. Providing a gloved finger to evoke the sucking reflex may help to relax the abdominal musculature. The presence of a fussy, crying newborn, or stool-filled intestines may make this portion of the examination somewhat difficult. If the examiner is unable to palpate the kidneys, documentation of voiding ensures the presence of some renal tissue.

The newborn's kidneys are in a lower position in the abdomen than they will be in later life. Normally, the inferior poles of both kidneys can be felt; the right kidney is usually lower than the left.[2] Normal-sized kidneys, 4.5 to 5 cm in length in a term newborn,

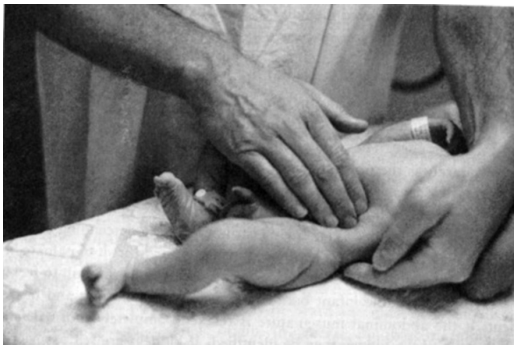

Figure 9.2 One technique for palpating the kidneys

Source: From Coen RW, and Koffler H. 1987. *Primary Care of the Newborn.* Boston, MA: Little, Brown, 30. Reprinted by permission.[7]

can be palpated in the flank areas above the level of the umbilicus.[8] They should be approximately equal in size and smooth to the touch. Ureters are not palpable under normal conditions. Enlarged kidneys are easy to detect; this finding should prompt further investigation.

Figure 9.2 illustrates one technique for palpating kidneys. The upper hand palpates the upper and lower quadrants, while the other hand supports the flank. The process is repeated on the opposite side. An alternate method is depicted in Figure 9.3. Place the fingers of one hand under the infant, with the thumb on the abdomen. To palpate the kidneys, compress the fingers against the thumb, the fingers support the flank while the thumb explores the area.[8]

The bladder can usually be palpated between the umbilicus and the symphysis pubis. Because the bladder wall is thin, this organ may be difficult to palpate. Percussion may then be helpful to detect a full bladder over the symphysis pubis, or in differentiating between a urine-filled organ and a solid mass. A bladder containing urine will yield a tympanic sound, whereas a solid mass will invoke a dull sound.[2]

Male Genitalia

Inspect the urogenital area with the male newborn in the supine position. Figure 9.4

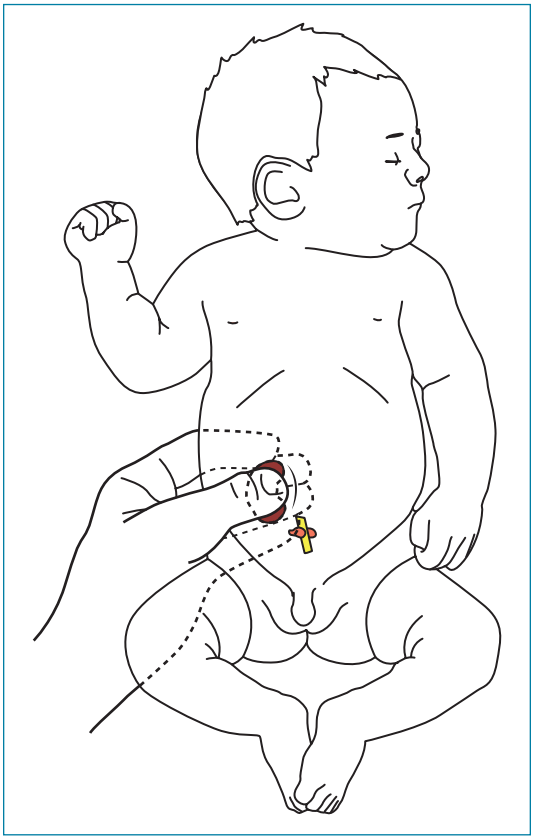

Figure 9.3 Alternate method for palpating the kidneys

is a diagram of normal male genitalia. Gestational age has a great impact on the appearance of external genitalia. Figure 9.5 illustrates changes in external genitalia with advancing gestation. Rugae (wrinkles or creases) begin to form on the ventral surface of the scrotum at approximately 36 weeks gestation. At term, the scrotum is fully rugated and more deeply pigmented than surrounding skin.

Palpate the scrotal sac and inguinal canal to locate the testes and to detect any masses. Prior to 28 weeks gestation, the testes are abdominal organs; at 28 to 30 weeks, they begin to descend into the inguinal canal. By term, the testes should be well situated in the scrotum.[1,9] When palpated, normal testes are firm and smooth and comparatively equal in size. They are ovoid in shape, usually mobile, and measure, on average, 1.4 to 1.6 cm in the term newborn.[10,11] Schulman

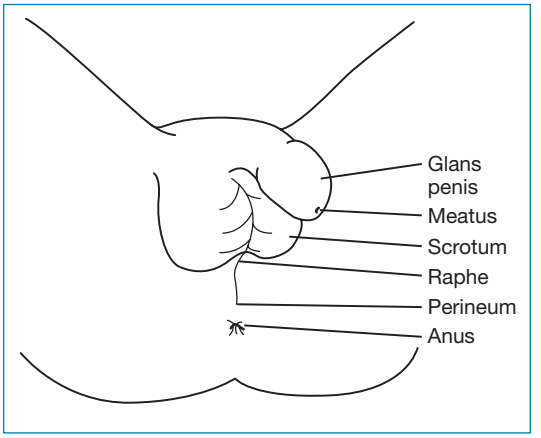

Figure 9.4 Normal newborn genitalia (male)

Source: Adapted from Grumbach MM, and Conte FA. 1992. Disorders of sexual differentiation. In *William's Textbook of Endocrinology*, 8th ed., Wilson JD, and Foster DW, eds. Philadelphia, PA: Saunders, 331. Reprinted by permission.[12]

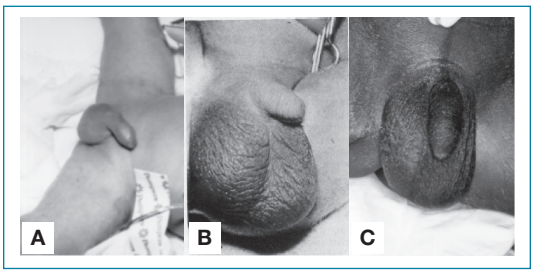

Figure 9.5 Appearance of genitalia in (A) preterm, (B) term, and (C) postterm male neonates

Source: Term infant is from: Lepley CJ, Gardner SL, and Lubchenco LO. 1989. Initial nursery care. In *Handbook of Neonatal Intensive Care*, 2nd ed., Merenstein GB, and Gardner SL, eds. Philadelphia, PA: Mosby, 88. Reprinted by permission.[13]

and colleagues suggest the following technique for detecting testes in the inguinal area: (a) Place the infant supine. (b) Apply soap or oil to the skin and examiner's fingers. (c) Using gentle to firm pressure, slide two or three fingers along the inguinal canal toward the scrotum. It may be necessary to repeat this maneuver numerous times before testes can be felt.[14]

Newborn males may have edema of the genitalia due to effects of transplacentally acquired maternal hormones. Trauma may occur during breech delivery (Figure 9.6) or in very large babies. Ecchymosis, edema, or even hematomas may be seen. These findings

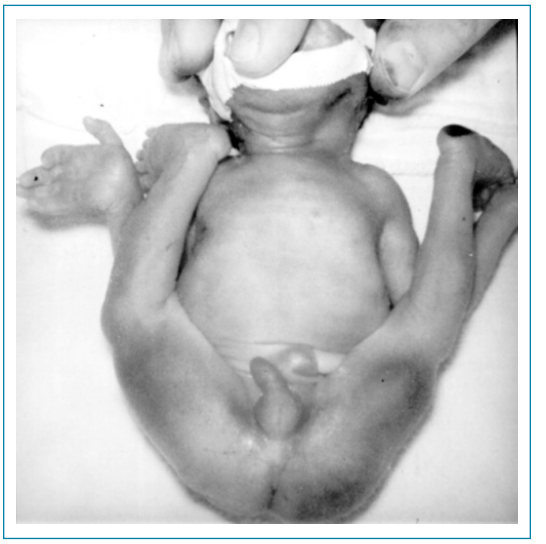

Figure 9.6 Genital bruising with breech presentation

Courtesy of Dr. David A. Clark, Albany, NY, Medical Center and Wyeth-Ayerst Laboratories, Philadelphia, PA.

are transient and should begin to dissipate after a few days.

The prepuce, or foreskin, covers the entire head of the penis in an uncircumcised male newborn. The function of the foreskin is to protect the urethral meatus from minor trauma.[11,15] Normally, the prepuce is tight, with a tiny orifice, but the opening is usually adequate to allow urination.[1] In newborns, the prepuce is adherent to the glans and cannot be retracted without disrupting its natural adherence to the surface of the glans. Therefore, forceful retraction should be avoided.[1,16] Physiologic phimosis, the inability to retract the foreskin, is normal in newborns and generally becomes retractable during the first few years of life.[11,15,16]

Gentle traction is applied on the foreskin to visualize the urethral meatus at the central tip of the penis. The penis should be straight; erections are commonly seen in newborns. Observation and documentation of the force and direction of the urine stream while voiding are important. The urine stream should be forceful, straight, and continuous.

Penile length and width should be assessed. The average stretched length of the penis in a term newborn is 3.5 cm,

measured from the pubic bone to the tip of the glans (omitting excess foreskin).[10] Some newborns have a large deposit of adipose tissue overlying the pubic bone, giving the illusion of a small penis. In such cases, it is important to depress the fat pad while stretching the shaft for assessment of length. Comparative nomograms are available to enable the examiner to evaluate an abnormally sized penis.[14] Shulman and associates describe findings that are consistent with a normal genital examination in a newborn male.[14] There may be gestational and racial differences in the size of genitalia.[14,17,18] Male newborns with measurements that lie outside of the normal range should be compared to those of same gestational age and racial background if such information is available. Figure 9.7 demonstrates a measurement technique for penile length. Penile width is measured midshaft on a stretched penis and should be 0.9 to 1.3 cm in a term newborn.[11]

If catheterization is necessary, a #5 French catheter or feeding tube should pass easily through the external meatus and urethra of either a preterm or a term male newborn. By 1 year of age, the urethra should be able to accommodate a #8 French tube.[11,20]

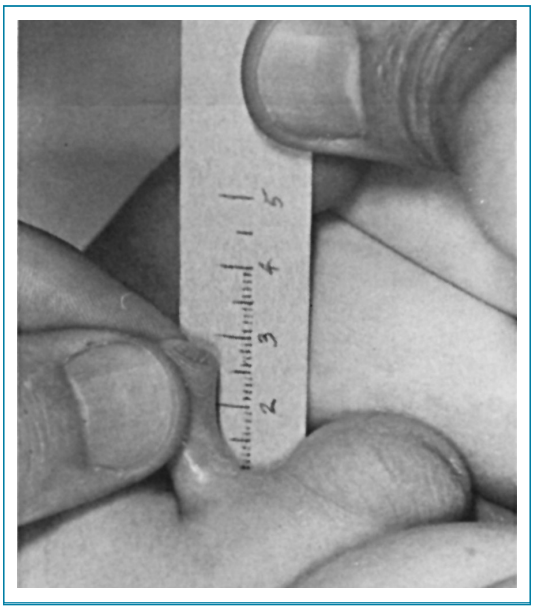

Figure 9.7 Technique for measuring penile length

Source: From Shulman RM, Palmert MR, and Wherrett DK. 2011. Metabolic and endocrine disorders, Part 4: Disorders of sex development. In *Fanaroff and Martin's Neonatal–Perinatal Medicine: Diseases of the Fetus and Infant*, 9th ed., Martin RJ, Fanaroff AA, and Walsh MC, eds. Philadelphia, PA: Mosby, 1592. Reprinted by permission.[19]

Female Genitalia

The female genitalia are inspected with the newborn in the supine position. An illustration of the normal external female anatomy is provided in Figure 9.8. The labia majora are the outermost structures, extending from the mons pubis to the labial commissure. Medial to the labia majora are the labia minora, which join anteriorly to form the prepuce of the clitoris.[10]

For the first 8 weeks of life, the term female newborn may have prominent labia, a large clitoris, and a urethral meatus that is difficult to visualize because of the influence of maternal estrogen. Maternal hormone exposure can stimulate a white, mucoid vaginal discharge and/or bleeding (pseudomenses). These findings may persist for up to 10 days. The genitalia of breech-positioned and large newborns may be edematous and ecchymotic for several days after delivery.[9] Gestational age influences the appearance

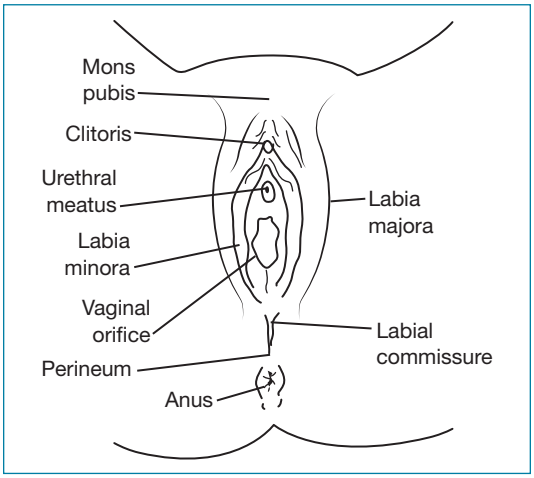

Figure 9.8 Normal newborn genitalia (female)

Source: Adapted from Grumbach MM, and Conte FA. 1992. Disorders of sexual differentiation. In *William's Textbook of Endocrinology*, 8th ed., Wilson JD, and Foster DW, eds. Philadelphia, PA: Saunders, 331. Reprinted by permission.[12]

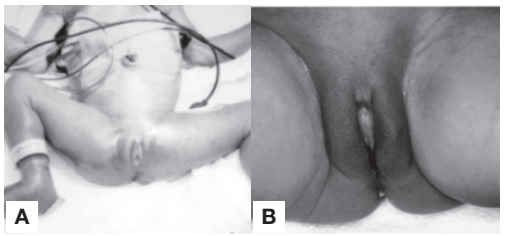

Figure 9.9 Appearance of genitalia in (A) preterm and (B) term female neonates

of the female genitalia (Figure 9.9). In preterm females, the labia minora and clitoris are very prominent, and the labia majora are small because of a lack of adipose tissue. The labia majora are larger in more mature newborns; in term females, they usually cover the clitoris and labia minora. The urethral meatus should be just ventral to the vaginal opening. Deviation from this position may indicate a urogenital sinus or ambiguous genitalia.[14]

The labia and the inguinal and suprapubic areas are inspected and palpated to detect any masses, bulges, or swelling. The labia are then separated with gentle lateral and downward traction of the examiner's fingers. The clitoris is the uppermost structure, located at the junction of the labia minora. Normal values for clitoral measurements in the newborn have been published.[10,17]

Directly below the clitoris and above the vaginal opening is the urethral meatus. The perineum is the area between the vaginal opening and the anus; it should be smooth, without dimpling or fistulas. Normally, in the term female, the perineum is as wide as a fingertip. Abnormal length of the perineum with abnormal spacing between the vaginal, urethral, and anal orifices is occasionally associated with GU anomalies.[14] The vaginal orifice measures 1.5 cm in the average term female.[21] The hymen is a thickened avascular membrane with a central orifice. A hymenal tag (Figure 9.10) is a common neonatal variation that usually disappears in a few weeks.[9]

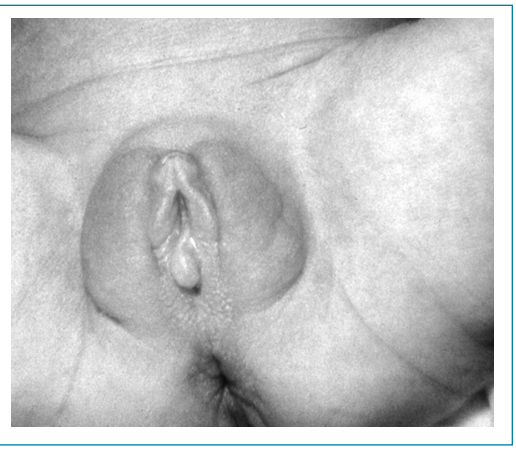

Figure 9.10 Hymenal tag in a neonate

ABNORMAL FINDINGS ON ABDOMINAL EXAMINATION

Abdominal Distention

Abdominal distention is a frequent finding in the newborn because of poorly developed abdominal musculature. Masses or, less common, ascites can be a cause of abdominal distention. Most palpable abdominal masses in newborns are of renal origin, and the most common causes are hydronephrosis and multicystic, dysplastic kidneys.[22] The urinary bladder is higher in the abdomen in the newborn than in the older infant, so bladder distention is a frequent cause of abdominal distention and the most common midline abdominal mass. Persistent bladder distention can signify structural urethral defects, bladder obstruction, or neuromuscular disease. Dilated, enlarged ureters caused by urinary tract obstruction can also present as abdominal masses. An abdominal mass is a cause for concern and warrants further investigation.

Ascites refers to an intra-abdominal collection of fluid. Percussion reveals its presence. Lower urinary tract obstruction, particularly posterior urethral valves in the male, can be a cause of ascites. In these cases, ascitic fluid is composed of urine that has either escaped through a frank rupture in the collecting system or leaked through a renal calyx into the peritoneal cavity.[1]

Abdominal-Wall Defects

Eagle–Barrett syndrome (EBS), or *triad syndrome*, formerly called *prune belly syndrome*, is a congenital deficiency of the abdominal musculature that is readily apparent at birth (see Figure 8.1). The incidence of EBS is approximately one out of 35,000 to 50,000 births.[1] Characteristics of this syndrome are a large, flaccid, wrinkled abdominal wall; undescended testes; and various GU malformations, such as hydroureter, hydronephrosis, and renal dysplasia. Occasionally, imperforate anus, intestinal malrotation, rib cage anomalies, cardiovascular abnormalities, and lower limb defects are also present. This syndrome is seen almost exclusively in males; reports of affected females are rare.[23,24]

The spectrum of malformations called *exstrophy–epispadias complex (EEC)* involves defects of the urinary and genital tracts, musculoskeletal system, and sometimes the intestinal tract. *Exstrophy of the bladder* is a part of this spectrum. EEC is a rare condition, occurring in approximately one in 10,000 births, with a male predominance.[1,24,25] There is an absence of muscle and connective tissue in the anterior abdominal wall over the bladder caused by an embryologic defect, the incomplete closure of the inferior part of the abdominal wall. This presents as eversion and protrusion of the bladder through an abdominal-wall defect (Figure 9.11). The ureteral orifices can be identified on the exposed bladder surface. Complete exstrophy of the bladder is associated with epispadias in some male infants (Figure 9.12). In bladder exstrophy and epispadias, the upper urinary tract is usually normal, but renal agenesis, horseshoe kidneys, hydronephrosis, and other renal anomalies have been reported.

Cloacal exstrophy, the most severe defect, is more extensive and may include

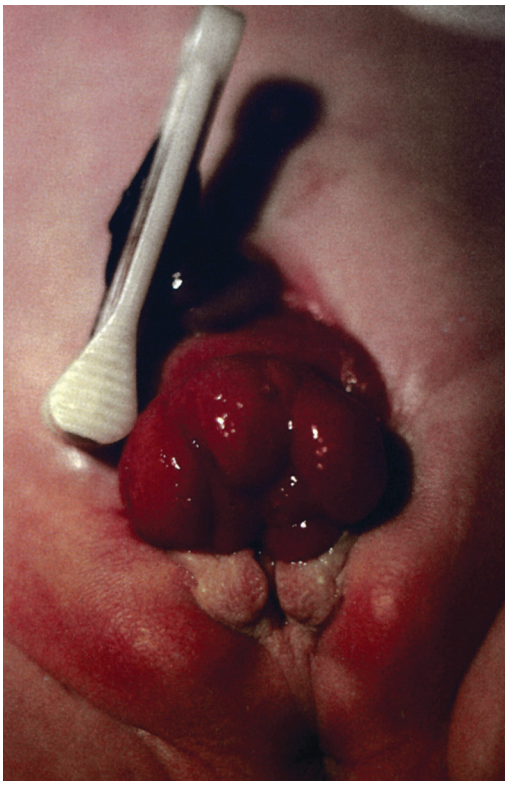

Figure 9.11 Exstrophy of the bladder

Source: Clark DA. 2000. *Atlas of Neonatology*. Philadelphia: Saunders, 201. Reprinted with permission.[26]

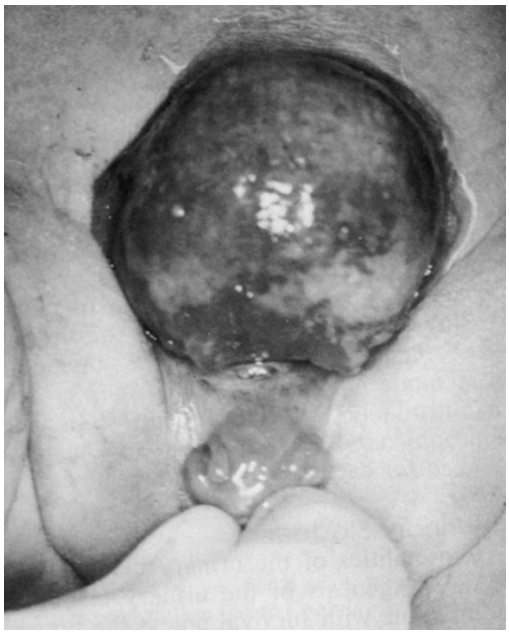

Figure 9.12 Exstrophy of the bladder with epispadias

Source: Kaplan GW, and McAleer IM. 2005. Structural abnormalities of the genitourinaty tract. In Neonatology: Pathophysiology and Management of the Newborn, 6th ed., MacDonald MG, Mullett MD, and Seshia MMK, eds. Philadelphia: Lippincott Williams & Wilkins, 1077. Reprinted by permission.[27]

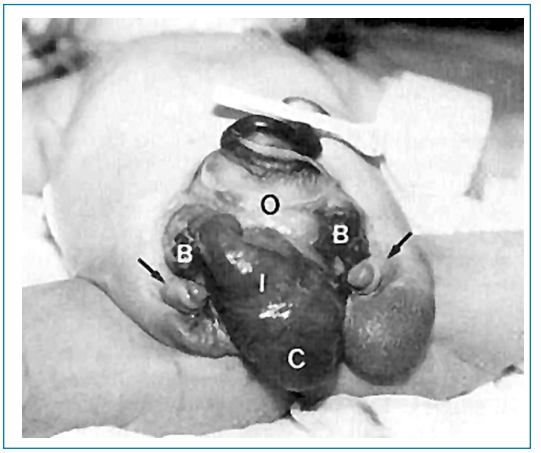

Figure 9.13 Newborn male with cloacal exstrophy

Note: Separate bladder components (**B**), associated omphalocele (**O**), terminal ileum (**I**), cecum (**C**), and bifid penis (*arrows*).

Source: From Zaontz MR, and Packer MG. 1997. Abnormalities of the external genitalia. *Pediatric Clinics of North America* 44(5): 1282. Reprinted by permission.[28]

an omphalocele containing intestines, liver, and spleen (Figure 9.13).[24,29] Loops of cecum and terminal ileum may be seen dividing the exposed bladder into two sections. In males, the penis is small and divided in half, whereas females may present with a bifid clitoris and uterus and a duplicate or exstrophic vagina.[1,25,29]

A wide array of abnormalities involving the cardiac, gastrointestinal, renal, musculoskeletal, and neurologic systems is associated with cloacal exstrophy.[1,25,29]

Umbilical Cord Anomalies

A single umbilical artery (SUA) is found in 0.3% of newborns. When occurring as an isolated finding in an otherwise normal infant, further diagnostic workup is no longer recommended because SUA is associated with renal abnormalities in only 7% of cases.[30,31] The urachus (see Figure 8.3) is an embryologic structure that connects the fetal bladder with the umbilicus. Postnatal patency of the urachal remnant can result in a clear discharge (urine) from an otherwise normal appearing umbilical cord. The discharge of urine may be intermittent or minimal. Obtaining the specific gravity of the discharge can confirm it to be urine. A large, edematous umbilical cord that does not separate in the normal amount of time may be the only sign of a patent urachus or urachal cyst.[5] Urinary drainage from a urachal remnant can lead to umbilical granuloma, redness, or swelling below the umbilicus. A sign of a persistent urachus is retraction of the umbilical cord during urination. Evaluation for lower urinary tract obstruction should be considered in a newborn with a patent urachus.[10]

ABNORMALITIES OF THE MALE GENITALIA

The Penis

Aphallia (absence of a penis) and *diphallia* (duplicated penis) are rare conditions that require extensive reconstructive surgery.[16,32] There are two types of duplications: bifid penis and true duplication. Bifid penis may occur in EEC and is characterized by separation of the corporal body of the penis into two halves, with a single urethral meatus at the base of the shafts.[25,29] In true diphallia, there may be total duplication, with voiding and erectile function in individual shafts.[11]

Hypospadias is the abnormal location of the urethral meatus on the ventral surface of the penis Hypospadias is a common abnormality, occurring in approximately 8.2/1,000 births, and results from the incomplete development of the anterior urethra.[14] Failure of the urethra to develop inhibits proper development of the prepuce. Most newborns with hypospadias also have a hooded or malformed prepuce (Figure 9.14).[14,24] It is important to locate the urethral meatus in a newborn with a malformed or hypoplastic prepuce and not simply to dismiss the malformation as a "natural circumcision."

Hypospadias can be classified into three categories based on meatal position: (a) Balanic (glanular) hypospadias exists when the urethral opening is ventrally situated at the base of the glans (Figure 9.15). (b) Penile hypospadias occurs when the meatus is found between the glans and the

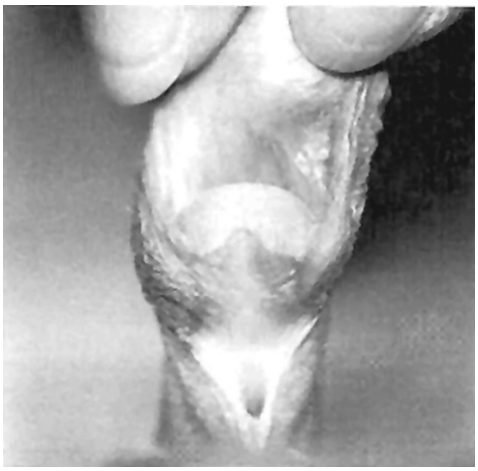

Figure 9.14 Distal hypospadias

Note: Dorsal hooded foreskin, flattened glans, and distal shaft meatus.

Source: From Zaontz MR, and Packer MG. 1997. Abnormalities of the external genitalia. *Pediatric Clinics of North America* 44(5): 1268. Reprinted by permission.[28]

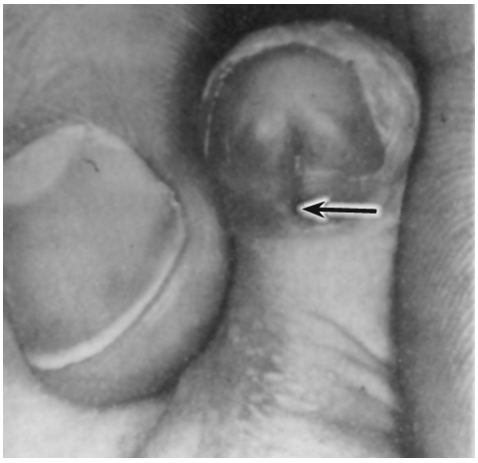

Figure 9.15 Balanic (or glanular) hypospadias

This is the most common form of hypospadias. The external urethral orifice is indicated by the arrow. There is a shallow pit at the usual site of the orifice. Note the moderate degree of chordee, causing the penis to curve ventrally.

Source: From Jolly H. 1968. *Diseases of Children*, 2nd ed. Oxford, UK: Blackwell Scientific Publications. Reprinted by permission via Copyright Clearance Center. Reprinted with permission.[33]

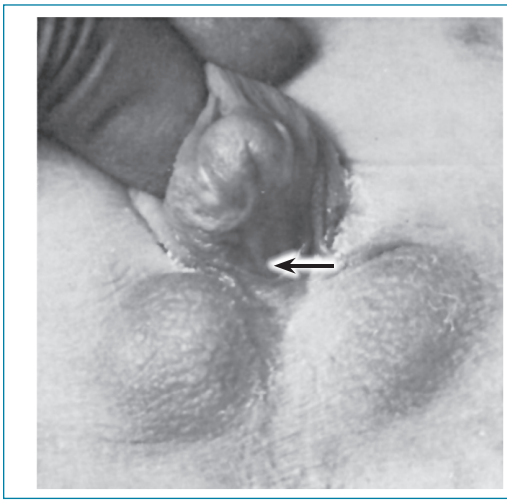

Figure 9.16 Penile hypospadias

The penis is short and curved (chordee). The external urethral orifice (*arrow*) is near the penoscrotal junction.

Source: From Moore KL, and Persaud TVN. 1998. *Before We Are Born: Essentials of Embryology and Birth Defects*, 5th ed. Philadelphia, PA: Saunders, 320. Reprinted by permission.[34]

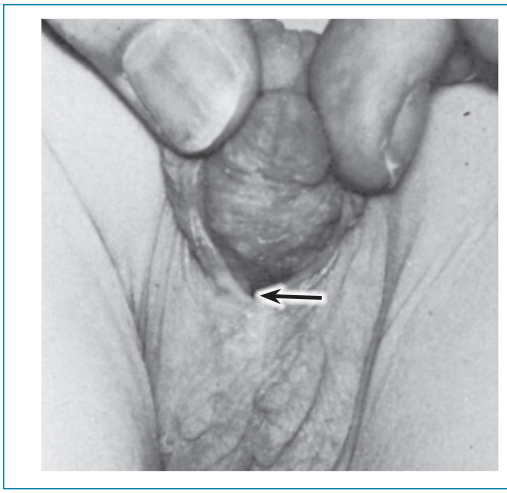

Figure 9.17 Penoscrotal hypospadias

The extenal urethral orifice (*arrow*) is located at the penoscrotal junction.

Source: From Moore KL, and Persaud TVN. 1998. *Before We Are Born: Essentials of Embryology and Birth Defects*, 5th ed. Philadelphia, PA: Saunders, 320. Reprinted by permission.[34]

scrotum (Figure 9.16). (c) *Penoscrotal hypospadias* (Figure 9.17) and *perineal hypospadias* are defined as the urethral opening at the penoscrotal junction and on the perineum, respectively.[32,35] Isolated balanic (glanular) and penile hypospadias without other genital abnormalities or dysmorphic features are rarely associated with chromosomal or endocrine disorders or with problems of sexual

differentiation. Newborns with penoscrotal or perineal hypospadias have a higher risk for these problems. In newborns with hypospadias and additional genital anomalies, such as cryptorchidism or micropenis (both discussed later in this chapter), the risk for endocrine imbalance or problems with sexual differentiation rises.[24,36] Further investigation is warranted in these cases.

Chordee is a bend in the shaft of the penis, an aspect of penile development that occurs between the 16th and 20th week of gestation and may be seen in some normal premature newborns. In these newborns, chordee may resolve spontaneously within the first few months of life.[20] However, it is more common for chordee to be caused by fibrous tissue growth in an area of failed urethral development or by skin traction from skin deficiency as seen in hypospadias or epispadias.[1,32] Chordee may not be evident without the presence of an erection.[11] Ventral chordee frequently, but not always, accompanies hypospadias (see Figure 9.15).

A variant of hypospadias that can be identified only after circumcision is called *mega-meatus with intact prepuce.* In this type, there is a well-formed, complete foreskin; the hypospadias is hidden by the prepuce, and there is no chordee.[16]

Epispadias, the location of the urethral meatus on the dorsal aspect of the penis, varies in severity from a glanular defect (Figure 9.18) to the complete version seen in exstrophy of the bladder (see Figure 9.12). All forms of epispadias are associated with differing degrees of dorsal chordee.[24,32]

Circumcision should be delayed in newborns with hypospadias or epispadias until after a consultation with a pediatric urologist; the foreskin may be used in the repair of these defects, if a surgical repair is deemed necessary. Parents should be notified of the possibility of reconstructive surgery by a pediatric urologist. Repair is generally performed after 6 months of age.[32]

Phimosis, which is generally physiologic in infants (see earlier in "Male Genitalia"), can become pathologic as a result of inflammation and scarring at the tip of the foreskin. Phimosis can occur as a result of forcefully retracting the foreskin.[16] Paraphimosis is a condition in which the foreskin has been retracted proximal to the glans penis and cannot be restored to its proper position because of swelling due to venous congestion.[16]

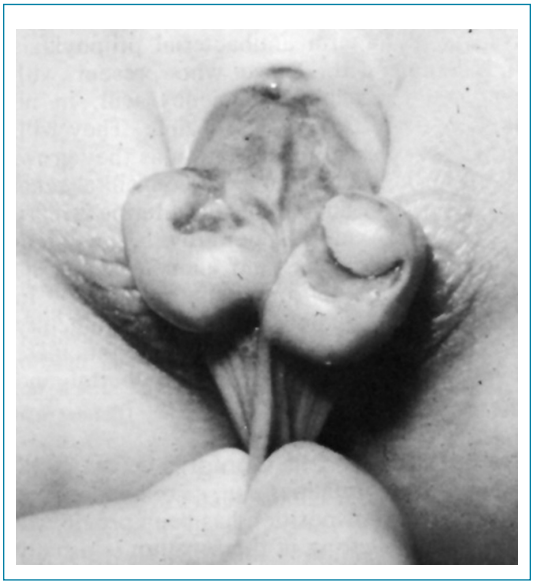

Figure 9.18 Epispadias

Source: Kaplan GW, and McAleer IM. 2005. Structural abnormalities of the genitourinaty tract. In Neonatology: Pathophysiology and Management of the Newborn, 6th ed., MacDonald MG, Mullett MD, and Seshia MMK, eds. Philadelphia: Lippincott Williams & Wilkins, 1087. Reprinted by permission.[27]

Hypospadias or epispadias can cause abnormalities in voiding.[16,37] In the male newborn, a weak urine stream, especially in the presence of a distended bladder, suggests the possibility of lower urinary tract obstruction (commonly, posterior urethral valves). The newborn with a spinal cord abnormality, such as a sacral tumor, spinal cord tethering, caudal regression syndrome, or myelomeningocele can also have abnormalities in voiding as the result of a neurogenic bladder. In these newborns, lesions of the nervous system interrupt the conduction of impulses from the brain to the bladder, preventing normal micturition.

A *micropenis* is an abnormally short or thin penis that is more than 2.5 standard deviations below the mean of length and width for age using standard charts.

Occasionally the penis appears small but, in reality the measurement is within acceptable limits when stretched, termed an *inconspicuous penis* (see Figure 9.7).[32] Evaluation and management of a newborn with a true micropenis frequently requires an endocrinologist and a geneticist.[1,14,24] (See "Disorders of Sexual Development.")

Priapism is a persistent, seemingly painless, penile erection that lasts less than 4 hours and usually resolves spontaneously. A common cause is sickle cell hemoglobin. Other factors that may contribute to its development are polycythemia and birth trauma. Many cases of priapism are idiopathic.[38] The neonate with a *webbed penis* (Figure 9.19) has a normal urethra and scrotum. However, the scrotal skin extends onto the ventral surface of the shaft, obscuring the penoscrotal angle and making the penis appear short.[6,16,38]

Penile torsion is a rotational deformity of the shaft, most often counterclockwise (to the left). This deformity is not obvious until after a circumcision or foreskin retraction and may be an isolated finding or may accompany other penile abnormalities.[32]

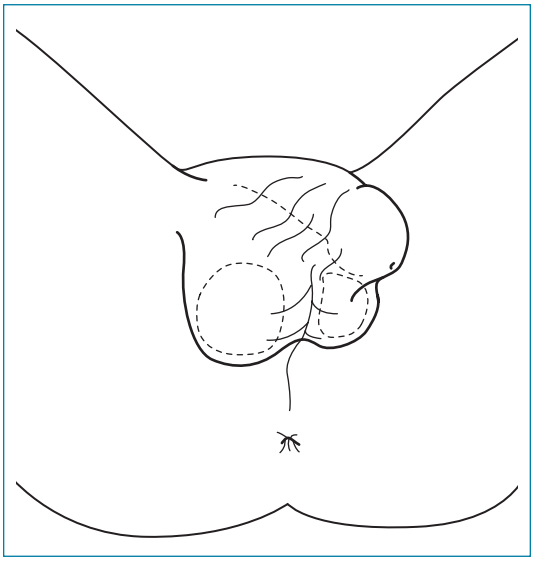

Figure 9.19 Webbed penis

Source: Adapted from Rozanski TA, and Bloom DA. 1997. Male genital tract. In *Surgery of Infants and Children: Scientific Principles and Practice,* Oldham KT, Colombani PM, and Foglia RP, eds. Philadelphia, PA: Lippincott-Raven, 1547. Reprinted by permission.[11]

The Scrotum

Cryptorchidism—literally, hidden testis—refers to a testis or testes that assume an extrascrotal location.[14,32] The condition occurs when one or both testes fail to descend completely into the scrotum.[1,39] Cryptorchidism is detected by the inability to palpate one or both testes in the scrotal sac. Cryptorchidism is seen in approximately 3.4% of term males and in 30% of all preterm newborns, with an incidence of nearly 100% in extremely preterm neonates.[14,40] A unilateral, undescended testis is more common than bilateral cryptorchidism. Bilateral cryptorchidism presents as an empty, hypoplastic scrotal sac.

A truly undescended testis is one that is interrupted in its usual path of descent, whereas an ectopic testis pursues an abnormal course of descent and may be found in a superficial inguinal pouch.[14,39,40] *Anorchia* is the absence of testicular tissue. A retractile testis is a normally descended organ that recedes into the inguinal canal because of activity of the cremasteric muscle. Because this muscle is inactive in newborns, retractile testes do not occur in this age group.[40] Thus, an empty or a hypoplastic scrotal sac in a newborn, when detected by observation and palpation, indicates truly undescended testes, ectopic testes (outside the external inguinal ring), or anorchia.[40]

Most undescended testes will descend by 3 months of age. Spontaneous descent rarely occurs after 9 months of age.[14,38] Diagnosis of true cryptorchidism and subsequent surgical intervention, called *orchiopexy*, is important because of an associated risk for infertility and malignancy in the cryptorchid testis.[14,39,40] Retractile testes usually grow and develop normally.[14]

Cryptorchidism may be an isolated defect, or it may exist with other GU anomalies such as hypospadias or micropenis. Newborns with undescended testes and other abnormal features, such as micropenis, bifid scrotum, and hypospadias, should be evaluated for endocrine problems and gender ambiguity.[14,24]

Hydroceles and hernias commonly present as bulges or swellings in the groin

or scrotum.[31] Both are the result of a patent processus vaginalis; however, they differ in several ways.[41] A *hydrocele* (Figure 9.20) is a nontender, fluid-filled, scrotal mass overlying the testis and spermatic cord presenting as a scrotal swelling caused by the passage of peritoneal fluid through the patent processus vaginalis into the scrotum or by the persistence of peritoneal fluid that has not been resorbed.[40,43] Newborns commonly (57.9% of all newborn males) present with hydroceles.[10] Upon examination, the entire circumference of the testis can be palpated.[31,43]

An actual or a potential indirect *inguinal hernia* may be associated with a hydrocele. Loops of intestine can herniate through the persistent processus vaginalis into the scrotum (Figure 9.21). The incidence of hernias is inversely proportional to gestational age and birth weight. Both a positive family history and prematurity increase the incidence of inguinal hernia in both males and females, although they present more often in males.[44] Other conditions associated with the development of inguinal hernias are hydrops fetalis, meconium peritonitis, urinary and

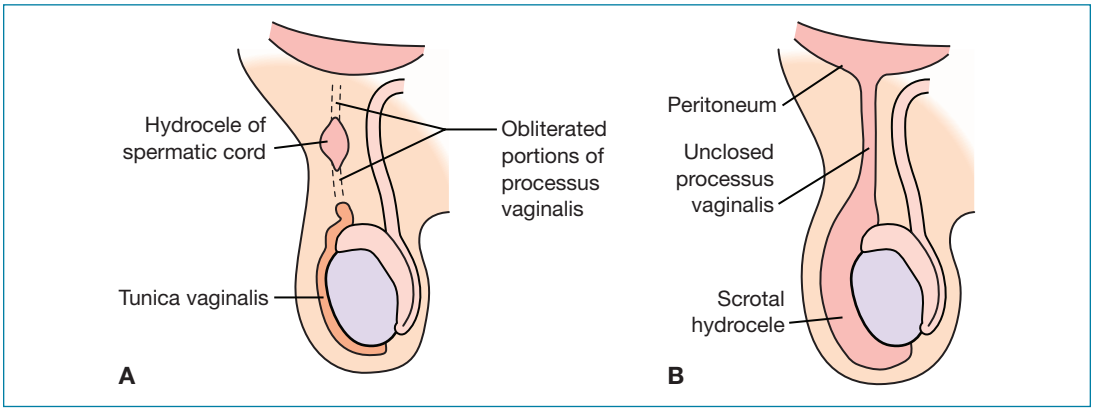

Figure 9.20 Hydrocele (A) Large hydrocele that resulted from an unobliterated portion of the processus vaginalis. (B) Hydrocele of the testis and spermatic cord resulting from peritoneal fluid passing into an unclosed processus vaginalis.

Source: From Moore KL, Persaud TVN, and Torchia MG. 2013. *The Developing Human: Clinically Oriented Embryology*, 9th ed. Philadelphia, PA: Saunders, 286. Reprinted by permission.[42]

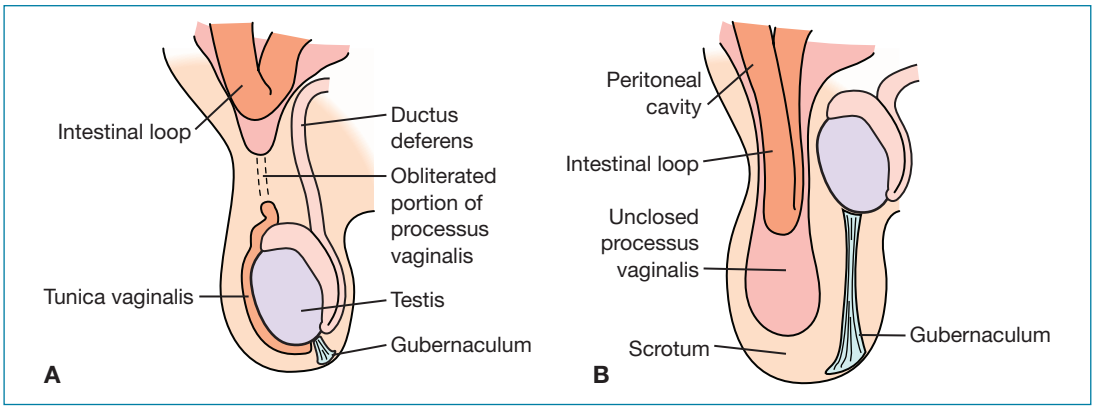

Figure 9.21 Inguinal hernia (A) Incomplete congenital inguinal hernia resulting from persistence of the proximal part of the processus vaginalis. (B) Complete congenital inguinal hernia into the scrotum resulting from persistence of the processus vaginalis. Cryptorchidism, a commonly associated anomaly, is also illustrated.

Source: From Moore KL, Persaud TVN, and Torchia MG. 2013. *The Developing Human: Clinically Oriented Embryology*, 9th ed. Philadelphia, PA: Saunders, 286. Reprinted by permission.[42]

chylous ascites, ambiguous genitalia, hypospadias and epispadias, cryptorchidism, cystic fibrosis, ventriculoperitoneal shunts, congenital hypothyroidism, and Beckwith–Wiedemann syndrome.[44]

According to Palmer and Palmer,[32] the examiner should be able to palpate a testis located in a soft hydrocele but may not be able to do so in the presence of a tense hydrocele. The presence of intestine in the scrotal sac renders the entire circumference of the testis impalpable.[9]

A scrotal mass or swelling from a hydrocele may be further distinguished from swelling caused by a hernia because a hydrocele appears translucent on transillumination (Figure 9.22). This may not be a universal finding, however. Hernias may also transilluminate because the bowel contains air.[43] Hernias are reducible, and hydroceles are not. To reduce a hernia, the examiner should grasp the most distal portion of the scrotum on the affected side with the fingers of one hand and apply firm, steady, upward pressure in the direction of the internal ring. The scrotal sac will decrease in size as the intestines return to the abdomen through the internal ring.[45]

Bowel incarceration, strangulation, and ischemic injury to the testes are potential complications of inguinal hernias. Incarceration is the inability to easily reduce the hernia; in strangulation, the loop of bowel becomes trapped, leading to ischemia and ultimate necrosis. Redness, pain, symptoms of intestinal obstruction, and difficulty in reduction are evidence of incarceration.[32]

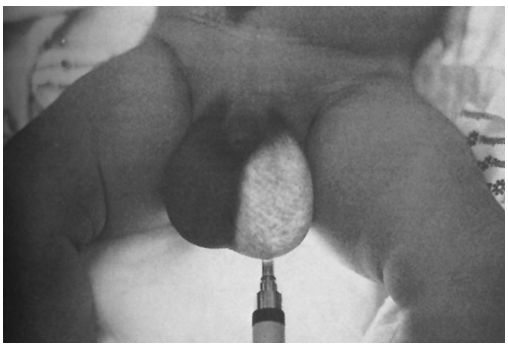

Figure 9.22 Transillumination of a scrotal hydrocele

Source: From Coen RW, and Koffler H. 1987. *Primary Care of the Newborn.* Boston, MA: Little, Brown, 33. Reprinted by permission.[7]

Hernia repair can be deferred if the hernia is easily reducible and there are no signs of incarceration, but surgery should be scheduled as soon as possible. Most hydroceles resolve in the first year of life.[32,43] Communicating hydroceles are, in reality, indirect inguinal hernias; therefore, repair is recommended if they persist after 2 years of age.[44]

Testicular torsion, or twisting of the testis on its spermatic cord, may occur prenatally and is usually unilateral. The neonate presents with a hard, swollen scrotum that is red to bluish red in color and does not transilluminate. This condition compromises blood supply to the testis; therefore, it requires urgent evaluation and possibly emergency management.[32] Ischemia of more than 4 to 6 hours duration usually results in irreversible damage and loss of the gonad.[32] With prenatal torsion, the duration of the torsion is unknown; by birth, irreversible ischemic damage to the testis may have occurred. Testicular torsion is painful in older children, but pain is not a universal finding in newborns.[14,32] Therefore, one should not be misled into discounting the possibility of testicular torsion in the case of a newborn male with a red, swollen scrotum that is not tender.

ABNORMALITIES OF THE FEMALE GENITALIA

There are several types of perineal masses found in newborn females. *Periurethral cysts* are the most common, appearing as a whitish epithelial covering adjacent to the unaffected urethral meatus. An *imperforate hymen* is frequently identified as a white midline bulge symmetrically located between the labia. *Hydrocolpos* (distention of the vagina) and *hydrometrocolpos* (distention of the vagina and the uterus; Figure 9.23) are the result of either incomplete canalization of the vagina during gestation or an imperforate hymen.[46,47] These conditions present in the newborn as lower abdominal masses and frequently as urinary tract obstructions. Urine retention may occur

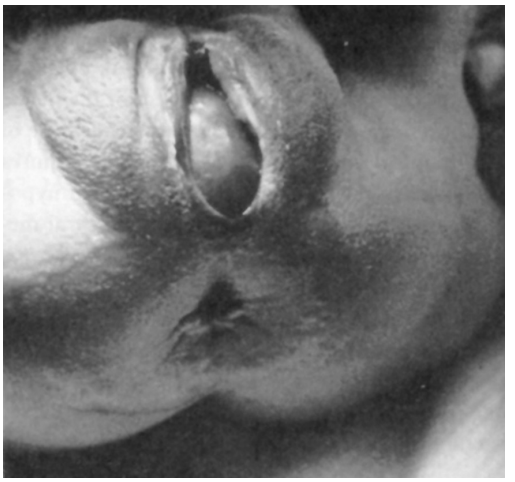

Figure 9.23 Hydrometrocolpos

Source: From Coen RW, and Koffler H. 1987. *Primary Care of the Newborn.* Boston, MA: Little, Brown, 90. Reprinted by permission.[7]

secondary to mass effect.[47] When the hymen is imperforate, it may bulge secondary to accumulation of vaginal secretions, creating the appearance of a shiny, cystic mass between the labia.[46,47] Prolapse of an ectopic ureterocele may be similar in appearance, but often is accompanied by edematous, congested or necrotic tissue, protruding from the posterior urethra.[46] Inguinal hernias occur less frequently in females than in males. When they do occur, they may present as a reducible swelling of the labia (Figure 9.24). On occasion, a gonad may be palpated in the suprapubic area. The question may then arise as to whether the infant is a female with a prolapsed ovary or a genotypic male with ambiguous genitalia. This is due to the fact that ovaries usually do not descend.[46] According to Karfer[46] all phenotypic females with inguinal hernias should be evaluated to determine chromosomal gender.

Clitoromegaly has been defined as an eight- to 10-fold increase in clitoral index (defined as width times length). Causes of clitoromegaly in the newborn include endocrine abnormalities (congenital adrenal hyperplasia), maternal factors (increased androgen production, drugs), and syndromes (Beckwith–Wiedemann, true hermaphroditism).[10]

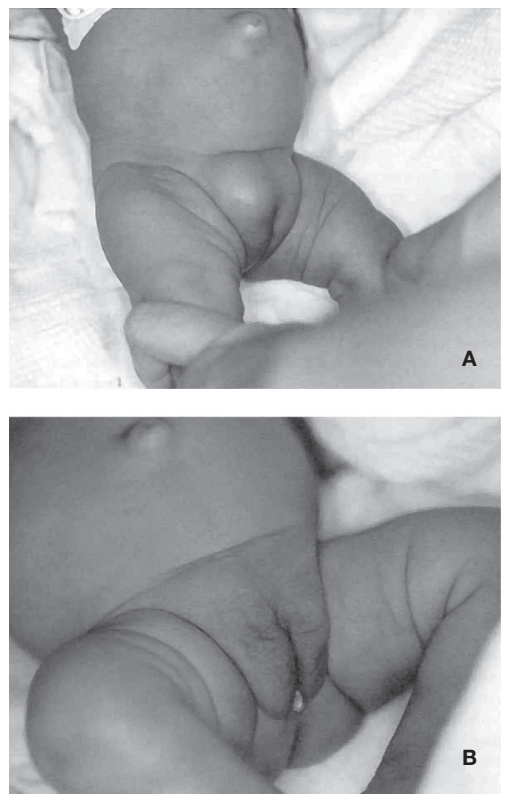

Figure 9.24 Inguinal hernias (A) before and (B) after reduction

Inguinal hernias occur more frequently in extremely premature females than in those born closer to term. The uterus, fallopian tubes, and ovaries, as well as the bowel, may herniate. (A) Hymenal tag is apparent after reduction in (B).

Source: From Fletcher M. 1998. *Physical Diagnosis in Neonatology.* Philadelphia: Lippincott-Raven, 388. Reprinted by permission.[11]

Disorders of Sexual Development

Ambiguous genitalia (Figures 9.25 and 9.26) may be defined as the presence of a phallic structure that is not discretely male or female, an abnormally located urethral meatus, and inability to palpate one or both gonads in males.[14] One should suspect problems of sexual differentiation in phenotypic males with bilateral impalpable testes, perineal hypospadias, or a unilateral undescended testis with hypospadias. Similarly, phenotypic females with clitoral hypertrophy, a palpable gonad, inseparably fused labia, or abnormal openings or dimpling on the perineum should be evaluated.[1,14,36,49] The association

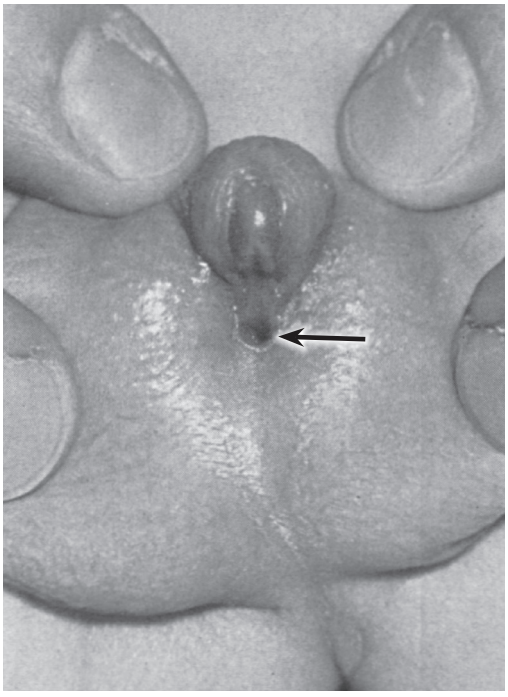

Figure 9.25 Newborn male infant (46, XY) with ambiguous genitalia

Note: Penoscrotal hypospadias (*arrow*). Testes are palpable in the scrotum.

Source: From Palmert MR, and Dahms WT. 2006. Abnormalities of sexual differentiation. In *Fanaroff and Martin's Neonatal–Perinatal Medicine: Diseases of the Fetus and Infant*, 8th ed., Martin RJ, Fanaroff AA, and Walsh MC, eds. Philadelphia, PA: Mosby, 1565. Reprinted by permission.[48]

of ambiguous genitalia with serious underlying endocrine disorders and the understandable distress of the parents mandate rapid identification and evaluation of these newborns. It is imperative that an endocrinologist, a genetic specialist, a urologist, and a psychologist/social worker be included on the evaluation team. The newborn should be referred to simply as "baby" until the appropriate sex of rearing is determined.

SUMMARY

Evaluation of the GU tract in the newborn is important to ensure rapid detection and treatment of abnormalities. Too frequently, this examination is cursory, and abnormalities, both major and minor, go unnoticed.

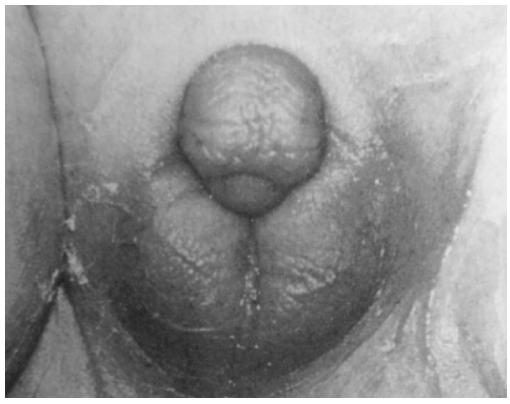

Figure 9.26 Newborn female with ambiguous genitalia: clitoral enlargement and fusion of labia majora

Source: From Moore KL, and Persaud TVN. 1998. *Before We Are Born: Essentials of Embryology and Birth Defects*, 5th ed. Philadelphia, PA: Saunders, 307. Reprinted by permission.[34]

Identification of problems in the neonatal period may preserve organ function and prevent mortality and morbidity in the future.

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Musculoskeletal System Assessment

Ellen P. Tappero, DNP, RN, NNP-BC

10

The musculoskeletal system of the body provides stability and mobility for all physical activity. It consists of the body's bones (Figure 10.1), joints, and their supporting and connecting tissues. In addition to allowing movement and providing structure, the musculoskeletal system protects vital organs (brain, spinal cord), stores minerals (calcium, phosphorus), and produces red blood cells and white blood cells. Careful scrutiny of the musculoskeletal system during the newborn's physical examination is imperative because (a) it is vital to the confirmation of musculoskeletal abnormalities detected on prenatal ultrasound such as spinal malformations, limb length discrepancies, and clubfoot deformities and (b) the information recorded during the first examination forms the database for all future examinations.

The bones of the newborn are soft because they are composed mostly of cartilage, which contains only a small amount of calcium. The process of ossification occurs rapidly during the first year of life. Compared with the skeleton of an adult or a child, the newborn's skeleton is flexible, and the joints are elastic. This elasticity is necessary to enable the infant to pass through the birth canal.

Unlike the bones of the skeletal system, the muscular system is almost completely formed at birth. Growth in the size of the muscle is caused by hypertrophy, rather than by hyperplasia of the cells.[1,2]

A thorough evaluation of the musculoskeletal system involves the techniques of inspection, palpation, and, on some occasions, listening. It includes an appraisal of the following:

1. Posture, position, and gross anomalies
2. Discomfort from bone or joint movement
3. Range of joint motion
4. Muscle size, symmetry, and strength
5. Configuration and motility of the back

Normal variations in shape, size, contour, or movement may be the result of position in utero or genetic factors. These normal variations should be distinguished from congenital anomalies and birth trauma. Early diagnosis of musculoskeletal disorders and early intervention often preempt the need for complex medical treatment and lead to more favorable outcomes.[3] Disorders that affect the musculoskeletal system may also originate from the neurologic system. An asymmetric Moro reflex, for example, may be caused by pain from a broken bone or a muscle injury, or it may be the result of a neurologic defect. Because there is some overlap between the musculoskeletal and neurologic examinations, assessment of muscle strength and motor activity are discussed with other neurologic assessments in Chapter 11.

PRENATAL HISTORY

Obtaining a comprehensive prenatal history is vital to the musculoskeletal assessment because a normal uterine environment is essential to the development of the fetus.

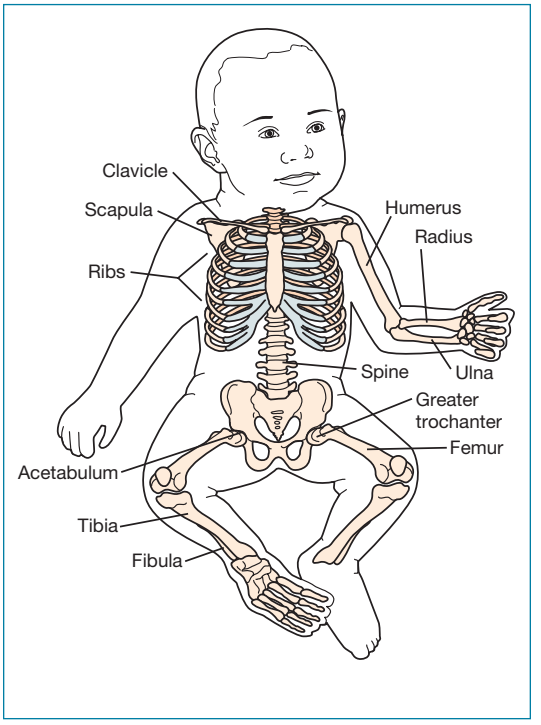

Figure 10.1 Infant's skeletal structure

Any event or condition that changes the intrauterine environment can alter fetal growth, movement, or position. Such prenatal factors as oligohydramnios, maternal uterine malformations, abnormal growth patterns, exposure to teratogenic agents, and breech presentation may adversely affect the development and maturation of the musculoskeletal system in utero. The perinatal history should be reviewed for possible birth trauma or neurologic insult. The examiner must note such factors as duration of labor, signs of fetal distress, and the type of delivery (vaginal or cesarean). These factors may have a bearing on conditions such as cerebral palsy, brachial plexus palsy (BPP), facial asymmetry, and torticollis. The birth order is also worth noting because there is a higher incidence of developmental dysplasia of the hip (DDH) in firstborn children.[4,5] An accurate gestational age assessment or estimated date of confinement from the obstetric record is necessary for accurate assessment of the infant's posture and muscle tone.

GENERAL SURVEY

Ideally, a thorough physical examination should be done within the first 24 hours after delivery. Because it is difficult to find a totally motionless, cooperative newborn, much of the musculoskeletal examination must be done while watching the newborn or while examining other systems. Nondisturbing maneuvers should be performed early in the examination and potentially distressing maneuvers near the end. For example, the head and neck should be palpated early and the hips examined near the end. The examiner must compile all the information produced by the examination and record all findings concerning the musculoskeletal system in a systematic manner. See Table 10.1 for pertinent terms used for the musculoskeletal examination.

For examination, the newborn should be completely undressed and positioned initially on the back. The examination area should be well lit, warm, and free of drafts. A radiant warmer or other heat source is necessary to prevent loss of body heat. The practitioner should develop a routine for examining the newborn so that no part of the musculoskeletal system is overlooked. The examination routine varies from practitioner to practitioner, but should be performed in a systematic head-to-toe manner. During assessment, the practitioner should consider general appearance and then proceed to specific areas.

Examination of the bony structures is important in a newborn examination because it is one of the first opportunities to assess intrauterine development. Deviations from normal may be the first indicator of a genetic abnormality or disease.[6]

The best instrument for measuring an infant's length, head circumference, and chest circumference is a narrow steel measuring tape or flexible plastic tape. However, most nurseries provide paper tapes, which may be less accurate. Folding the paper tape in half lengthwise may add strength as well as decrease slippage when measuring rounded contours such as the head.

TABLE 10. 1 MUSCULOSKELETAL TERMINOLOGY

Use the following terms to accurately and consistently describe skeletal positions and muscle movements observed during the examination:	
Abduction	Moving a limb away from the midline of the body
Adduction	Moving a limb toward or past the midline of the body
Dorsiflexion	Flexing a limb toward the back, as in flexing the foot so that the forefoot is higher than the ankle
Everted	Turning a limb out and away from the midline of the body
Extension	Straightening a limb at a joint
Flexion	Bending a limb at a joint
Inverted	Turning a limb inward toward the midline of the body
Plantar flexion	Extending the foot so that the forefoot is lower than the ankle
Pronation	Turning the face down
Rotation (neck)	Turning the face to the side
Supination	Turning the face up
Valgus	Bent outward or twisted away from the midline of the body
Varus	Turned inward

Measurements

Measurement of growth as reflected in increasing body weight and length along expected pathways and within certain limits is one of the most important indicators of health in an infant.[6] Measurements taken soon after birth demand careful attention to detail because they will act as a baseline for subsequent assessments of growth and development. All measurements are plotted on a growth chart and correlated with gestational age (Chapter 3).

Weight

Infants should be weighed without clothing or diaper and at approximately the same time each day. Using an infant scale, put a protective cloth or paper liner in place, and then place the infant on the scale.[7,8] Weigh newborns in both pounds and ounces, and in grams. Newborn weight varies with gestational age, gender, ethnicity, and socioeconomic status. Average weight for a term newborn is 2,500 to 4,000 g (5 lb. 8 oz.–8 lb. 13 oz.).[1,7–9] All newborns initially lose weight, with a loss of 10% to 20% of birth weight considered acceptable. Variations in weight are a result of body water changes. Preterm infants lose more weight and regain birth weight slower than term infants. Weight gain usually begins within the first 2 weeks, with an average daily weight gain of 10 to 20 g/kg/day.[7,10] In general, infants double their birth weight by 4 to 5 months of age and triple their birth weight by 12 months of age.[7]

Length

The infant's length is a better indicator of body mass and long-term growth because it is not influenced by changes in body water content. The infant's recumbent length is measured from the heel to the crown (top of the head). The infant should be placed supine, with legs extended and the head flat. Make a mark on the bed to indicate the crown, and a mark at the heel of the infant. Measure the distance between those two marks. Direct measurement of the infant is difficult because of head molding

and incomplete extension of the knees. An alternate method for measuring length, although less accurate, is to hold the zero point of the tape at the heel and to run the tape along the surface of the bed to the top of the head. Mark the spot on the tape with a finger, remove the tape, and read the measurement.[11] Specialized devices to measure length may be available in some facilities.

The length of term newborns at birth is 46 to 56 cm (18–22 inches).[1,8,9] Average linear growth in preterm infants is 0.8 to 1 cm/week; in term infants, linear growth is 0.69 to 0.75 cm/week.[10] The average infant grows 10 cm (3 inches) in the first 3 months of life and length increases by 50% in the first year.[7,8]

Head Circumference

The head should be measured with a disposable tape measure around the occipital and parietal areas and over the frontal prominence, avoiding the ears. Intrauterine head growth is 0.5 to 0.8 cm/week and is an indicator of brain growth. The average findings in a term newborn are 33 to 35 cm (13–14 inches), with normal variations of 32.5 to 37.5 cm (12.5–14.5 inches).[1,7,8] The head circumference measurement can change markedly in the first few days of life because of molding of the head during delivery. Head circumference in preterm infants should be obtained weekly and plotted on a gestation-specific growth chart to be used as a reference for the future. Head circumference is correlated with long-term neurodevelopment.[10] As a general rule, the head circumference in centimeters is equal to one fourth of the body length in centimeters.[8]

Chest Circumference

Measurement of chest circumference is no longer a routine part of newborn physical examination in many hospitals. It can, however, be a useful measurement when compared with the head circumference if the examiner suspects a problem in the head or chest size. If it is measured, the chest circumference should be assessed at the nipple line during expiration. In term infants, the average chest circumference is approximately 2 cm smaller than the head circumference, with the average being 30.5 to 33 cm (12–13 inches).[1,7,11] The head circumference may exceed the chest circumference for the first 5 months of life. From the fifth month of life to 2 years of age, head and chest circumferences can be about the same.[7]

OBSERVATION

Observation, or inspection, should proceed from the general to the specific. General inspection includes observation for symmetry of movement, as well as size, shape, general alignment, position, and symmetry of different parts of the body. Soft tissues and muscles should be observed for swelling, muscle wasting, and symmetry.

In the extremities, no asymmetry of length or circumference, constrictive bands, or length deformities should be noted. Unequal length or circumference has been associated with skeletal anomalies, tumors, and intra-abdominal neoplasms.[7,12]

The ratio of extremity length to body length is also observed. If a discrepancy is seen, measurements of thoracic length and extremities should be recorded. The gestational age of the newborn determines the normal values for these measurements. In the term newborn, the ratio of upper body length to the length of the lower body segment should not exceed 1.7:1.[13,14] If this ratio is too high, then the extremities are short; if the ratio is too small, then either the limbs are long or the trunk is too short. This ratio is most useful in determining whether a small newborn is proportionate or has congenitally shortened lower extremities, as in achondroplasia (Figures 10.39 and 10.40).[14,15]

Term newborns lie in a symmetric position with the limbs flexed and the legs partially abducted at the hips so that the soles of the feet may nearly touch each other (Figure 10.2). The head is slightly flexed and positioned in the midline or turned to one side. The resting position of the newborn is often that of the tonic neck reflex

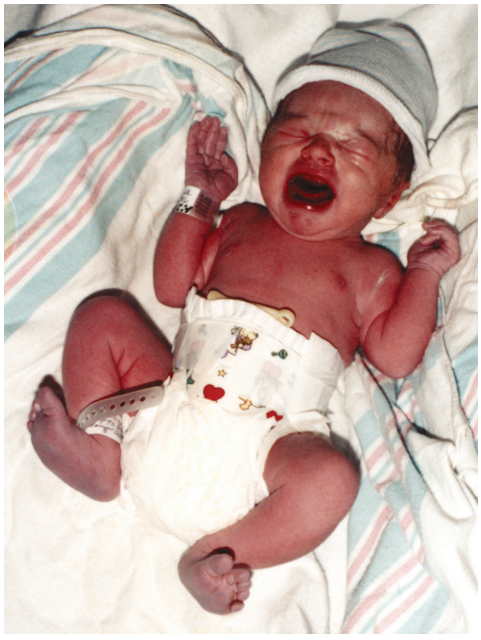

Figure 10.2 Posture of a term newborn

(see Figure 11.5). Spontaneous motor activity of flexion and extension, alternating between the arms and legs, is random and uncoordinated. The fingers are usually flexed in a fist, with the thumb under the fingers. Slight tremors may be seen in the arms and legs with vigorous crying during the first 48 hours of life. Any tremors noted after 4 days of age while the infant is at rest are considered abnormal and signal a neurologic problem.[6] The resting posture of the preterm infant is one of extension (Figure 10.3) and is discussed in Chapter 11.

The position and appearance of the extremities at birth can reflect intrauterine position. Because the lower extremities of the fetus have been folded on the abdomen, the newborn's lower extremities often appear externally rotated and bowed, with everted feet. The infant delivered in a breech presentation often has flexed, abducted hips and extended knees (Figures 10.4 and 10.5). These positional deformities can usually be corrected by passive joint manipulation and should not be confused with congenital malformations.

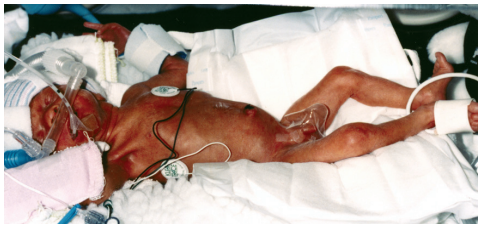

Figure 10.3 Posture of the preterm infant

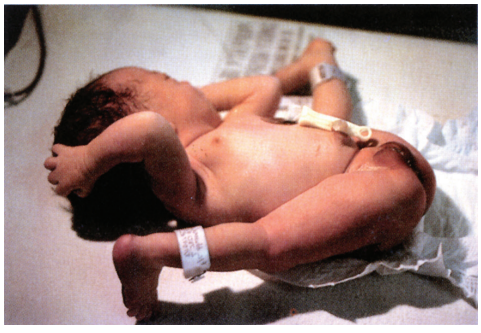

Figure 10.4 Breech presentation showing flexed, abducted hips and extended knees

Source: From Clark DA. 2000. Atlas of Neonatology. Philadelphia: Saunders, 9. Reprinted by permission.[16]

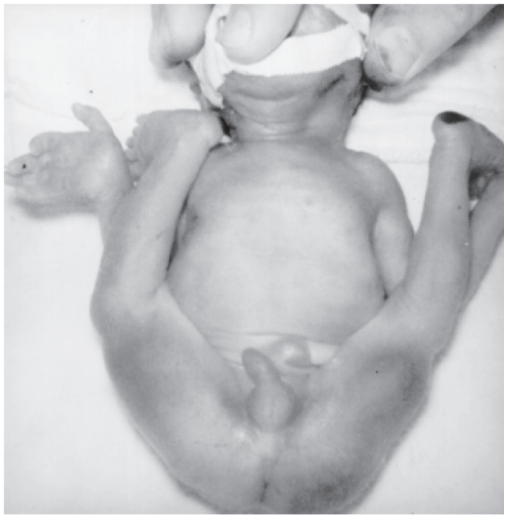

Figure 10.5 Abducted hips and perineal bruising with breech presentation

Courtesy of Dr. David A. Clark, Albany Medical Center, Albany, NY, and Wyeth-Ayerst Laboratories, Philadelphia, PA.

PALPATION

Palpation is the next important technique used in the examination of the newborn's musculoskeletal system. This technique, along with observation, is used on each extremity to identify component parts (e.g., the two bones in the forearm), function, and normal range of motion. Some aspects of this assessment are shared with the gestational age assessment. Muscular contour in the term infant is smooth, and despite lack of strength, the infant's muscles should feel firm and slightly resist pressure. If an infant feels limp, the condition should never be mistaken for a mere characteristic of immaturity. Further assessment is necessary to rule out a neurologic defect. When testing range of motion, note any asymmetry, tightness, or contractures. Range of motion of all joints is greatest in infancy, gradually lessening as the infant matures. As with posture and muscle tone, the range of joint motion varies with gestational age. It is not necessary to assess the exact number of degrees of range of motion, but only whether the range of movement is less than normal or significantly beyond normal findings. Never use excessive force to assess range of motion.

Neck

The neck is passively examined for rotation and for anterior and lateral flexion and extension. Rotation of 80° and lateral flexion of 40° to both the right and left sides are considered normal. In anterior flexion, the chin should touch or almost touch the chest, and on extension, the occipital part of the head should touch or almost touch the back of the neck. When there is asymmetric rotation or lateral flexion or when range of motion is limited, x-rays of the neck should be taken.

Upper Extremities

Examination of the upper extremities includes the bones of the shoulder girdle (the clavicle and scapula) as well as the humerus, elbow, forearm, and hand. Normal ranges for joint movements of the upper extremities are listed in Table 10.2. Asymmetry in range of motion may indicate weakness, paralysis, fractures, or infection. Failure to spontaneously move an extremity can indicate spinal cord injury or BPP.

Clavicles

The clavicles are inspected and palpated for size, contour, and crepitus (grating that can be felt or heard on movement of ends of a broken bone). A fractured clavicle should be suspected when there is a history of difficult delivery, irregularity in contour, shortening, tenderness, or crepitus on palpation. An asymmetric Moro reflex (Chapter 11) may also be seen. A broken clavicle is one of the most common birth injuries in newborns.[17,18]

Humerus

Length and contour of each humerus should be noted. A fractured humerus should be suspected if there is a history of difficult delivery, one feels a mass caused by hematoma formation, or there are signs of pain during palpation. After the clavicle, the humerus is the bone most often fractured during the birth process.[17,18]

Elbow, Forearm, and Wrist

The elbow, forearm, and wrist are examined for size, shape, and number of bones, as well as for range of joint motion. It is sometimes difficult to evaluate the elbow in infants because the normal neonate has a mild flexion contracture that does not disappear until a few weeks after birth. Wrist flexion varies with the infant's gestational age, with greater wrist flexion seen in the term than in the preterm infant.

Hand

The hand should be examined for shape, size, and posture while the fingers are examined for number, shape, and length. Inspection

TABLE 10.2 NORMAL NEONATAL RANGE OF MOTION IN THE UPPER EXTREMITIES

JOINT OR BONY UNIT	FLEXION	EXTENSION	EXTERNAL ABDUCTION	INTERNAL ROTATION	ROTATION
Shoulder	Close to 180°	≥25°	Close to 180°	≥45°	≥80°
Elbow	145°	165°–170°			
Forearm				Supination[a] ≥80°	Pronation[a] ≥80°
Wrist	75°–80°	65°–75°			
Digits	Able to clench	Full extension			
Metacarpal–phalangeal		0°			
Interphalangeal		0°→ 5°–15°			

[a]These maneuvers are done while the humerus is held immobile and the elbow is at 90°.

Source: From Scanlon JW, et al. 1979. *A System of Newborn Physical Examination.* Baltimore: University Park Press, 40. Reprinted by permission of the author.[13]

of palm creases should also be included. Although a single crease across the palm (Figure 10.6) is usually associated with Down syndrome, it is often found in normal infants. It can be found in 4% of the population and is seen bilaterally in 1%.[19] However, a combination of short fingers, an incurved little finger, a low-set thumb, and a single palmar (simian) crease should lead the examiner to investigate the possibility of Down syndrome. In term infants, the distance from the tip of the middle finger to the base of the palm is 6.75 ± 1.25 cm. Middle finger length to total hand length is usually 0.38 to 0.48:1 (Figure 10.7).[13,14] Macrodactyly, an abnormal enlargement of the finger or toe due to bone, blood vessel, nerve, and other soft-tissue enlargement, may be normal, or it may be a sign of neurofibromatosis, lymphedema, hemangioma, arterial vascular fistulas, fibrous dysplasia, or lipomas (Figure 10.8).[3] Flexed fingers with the index finger overlapping the third finger (Figure 10.9) should lead one to investigate the possibility of trisomy 18.[14] Puffy hands and feet in a newborn may appear as a result of lymphedema and are a characteristic finding of Turner and Noonan syndromes (Figure 10.10).[14,21]

Examine the nails for size and shape. The nails are usually smooth and soft and extend to the fingertips. They may be long in postterm infants or may be absent or spoon-shaped in the presence of some syndromes. Nails may appear hypoplastic if the infant's hands are edematous. A detailed discussion of nail examination is found in Chapter 4.

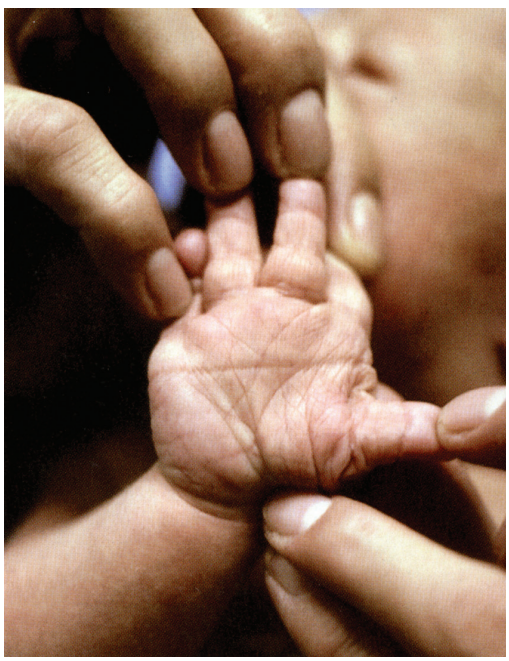

Figure 10.6 Single palmar crease (simian crease)

Source: From Clark DA. 2000. *Atlas of Neonatology.* Philadelphia: Saunders, 31. Reprinted by permission.[16]

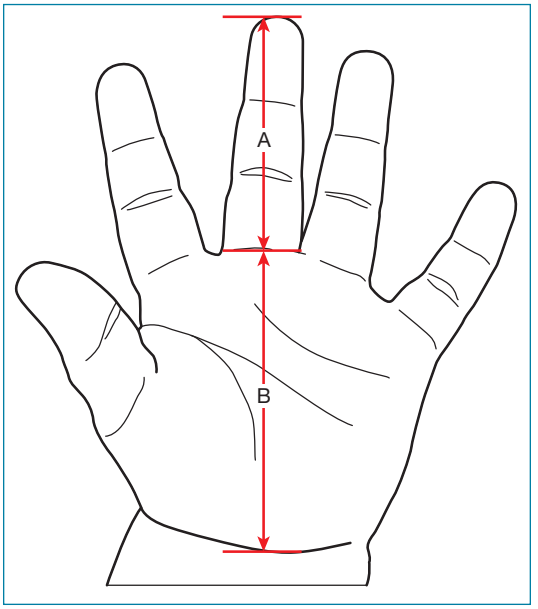

Figure 10.7 Landmarks used in measuring palm and finger length

Source: Adapted from Feingold M. 1994. Congenital malformations. In *Neonatology: Pathophysiology and Management of the Newborn,* 4th ed. Avery GB, Fletcher MA, and MacDonald MG, eds. Philadelphia, PA: Lippincott Williams & Wilkins, 761. Reprinted by permission.[20]

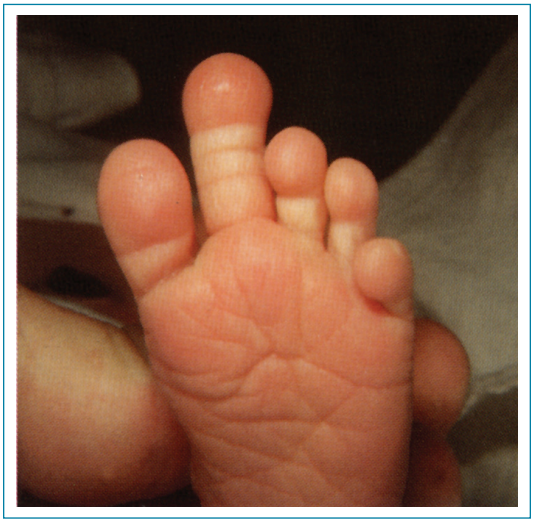

Figure 10.8 Macrodactyly

Source: From Clark DA. 2000. *Atlas of Neonatology.* Philadelphia: Saunders, 225. Reprinted by permission.[16]

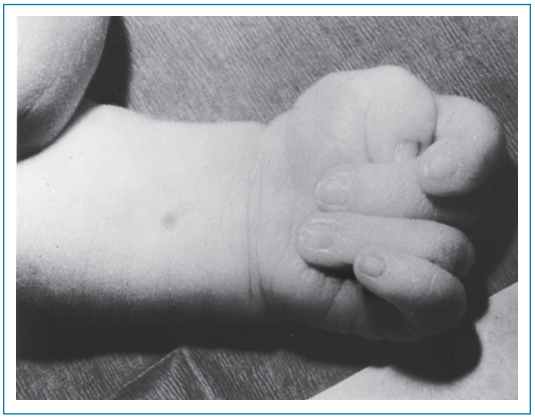

Figure 10.9 Finger position in an infant with trisomy 18

Courtesy of Eva Sujansky, MD, Associate Professor of Pediatrics, University of Colorado Health Sciences Center, Denver, CO.

Figure 10.10 Turner syndrome

Spine

The back is examined with the newborn lying prone or held suspended with the examiner's hand under the chest. First inspect the spine from the base of the skull to the coccyx, noting any skin disruption, tufts of hair, soft or cystic masses, hemangiomas, a pilonidal dimple (Figure 10.11), cysts, or sinus tracts. Such pathologic conditions may be signs of a congenital spinal or neurologic anomaly. The position of the scapulae should also be noted while the infant

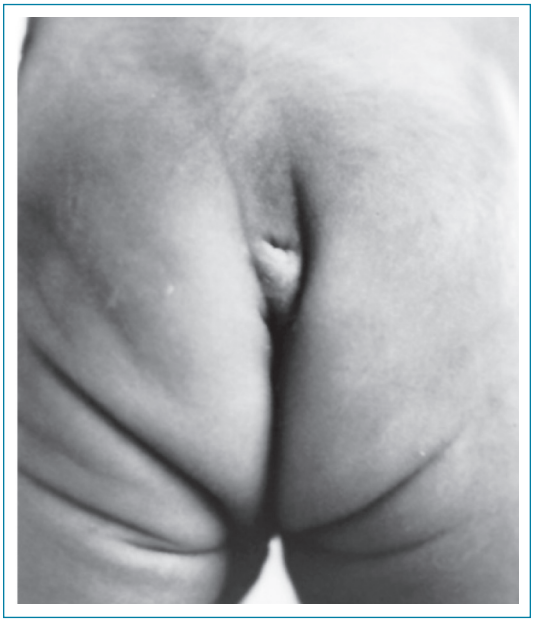

Figure 10.11 Pilonidal dimple

Courtesy of Dr. David A. Clark, Albany Medical Center, Albany, NY.

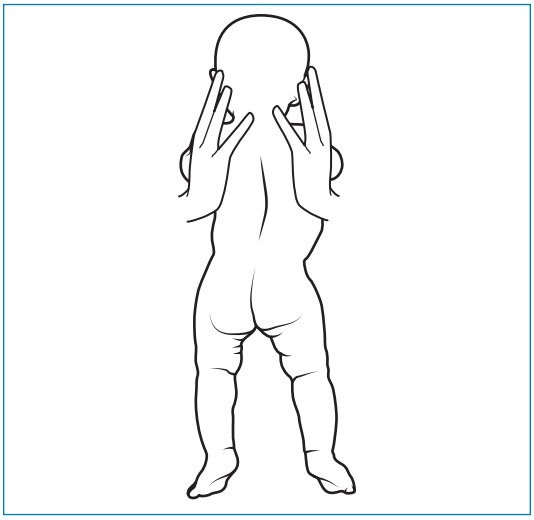

Figure 10.12 Scoliosis - denotes spinal curvature convex to the right or left. When the baby is lifted by the axilla, the scoliosis is usually obvious.

Source: Adapted from Milner RDG, and Herber SM. 1984. Color Atlas of the Newborn. Oradell, New Jersey: Medical Economics, 88. Reprinted by permission of Blackwell Science.[22]

Figure 10.13 Convex curvature of the thoracic and lumbar spine

Source: From Jarvis C. 2012. *Physical Examination & Health Assessment*, 6th ed. Philadelphia, PA: Saunders, 600. Reprinted by permission.[23]

is in the prone position to rule out Sprengel deformity, a winged or elevated scapula.

The entire length of the spine should be palpated to determine the presence of dorsal spinal processes and any abnormal curvatures. Gross abnormalities, such as scoliosis (Figure 10.12), lordosis, and kyphosis, are easily observed. A lateral curvature, however, is usually secondary to in utero position. A convex curvature of the thoracic and lumbar spine will be apparent when the infant is in a sitting position (Figure 10.13). The lumbar and sacral curves that are seen in adults develop later, when the infant sits up and begins to stand (Figure 10.14).[11] Extension and lateral bending of the spine can be noted by passive flexion. Flexion and extension should be smooth and rhythmic, without muscle spasm. A neurologic evaluation is necessary to complete the examination of the spine. Techniques are explained in Chapter 11.

Lower Extremities

Examination of the lower extremities includes the hips, femurs, tibias, fibulas, knees, ankles, and feet. Normal ranges for joint movement in the lower extremities are listed in Table 10.3.

Hips

The hips of a newborn generally have a flexion contracture. When the pelvis is stabilized and the lumbar spine is flattened out by extending one leg flat and flexing the

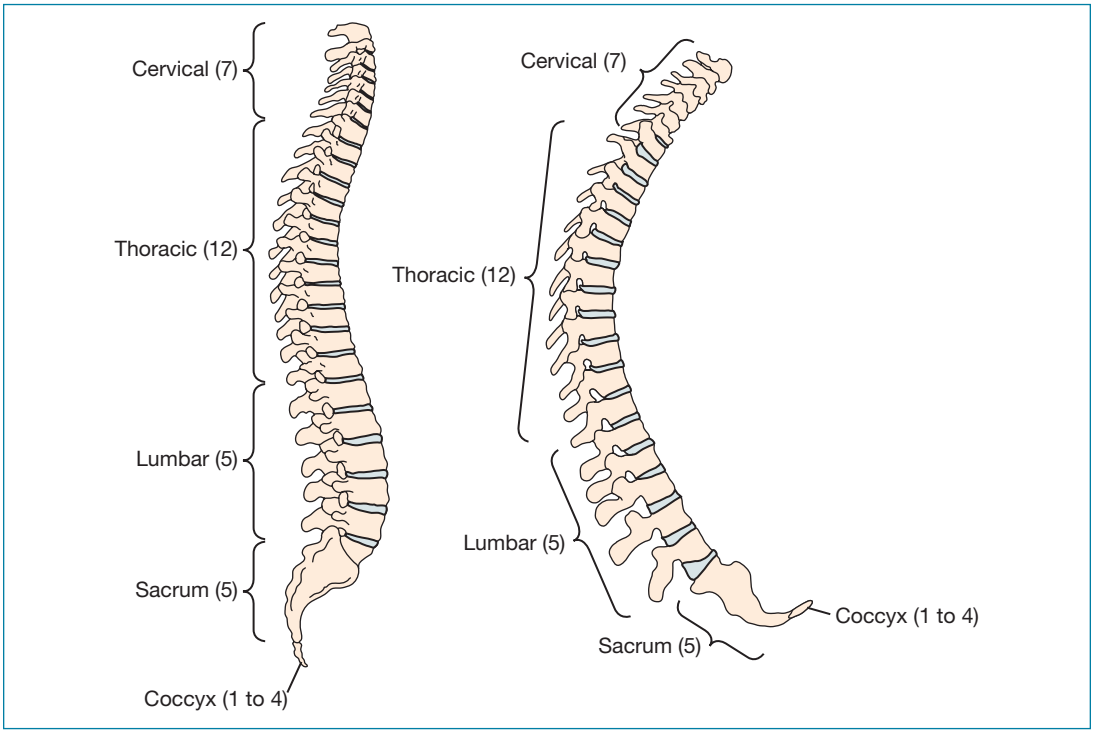

Figure 10.14 Spinal curves of the adult *(left)* and infant (*right*).

Source: From Alexander MM, and Brown MS. 1979. *Pediatric History Taking and Physical Diagnosis for Nurses*, 2nd ed. Philadelphia, PA: Mosby, 292. Reprinted by permission of the authors.[24]

TABLE 10.3 NORMAL NEONATAL RANGE OF MOTION IN THE LOWER EXTREMITIES

JOINT OR EXTREMITY	FLEXION	EXTENSION	ABDUCTION	ADDUCTION	INTERNAL ROTATION	ROTATION
Hip	145°		90°	10°–20°	40°	80°
Knee	120°–145°	90°				
Ankle	Dorsiflexion: Above resting position Plantar flexion: >10° from resting position					
Forefoot			≥10°–15°	≥10°–15°		
Hindfoot			Valgus ≥10°	Varus ≥5°		

Source: From Scanlon JW, et al. 1979. *A System of Newborn Physical Examination.* Baltimore, MD: University Park Press, 42. Reprinted by permission of the author.[13]

other leg with the knee to the chest, a flexion contracture of the hip can be detected in the extended leg. The degree of flexion contracture on the extended leg is the angle that is measured between the thigh and the horizontal plane of the bed or examining table.[7] Normal newborns have an approximately 20° to 30° flexion contracture that

usually resolves by 4 to 6 months of age. The newborn hip is externally rotated in extension to 80 to 90°, and internal rotation has a limitation of 0° to 10°.[3] The stability of the hip must also be evaluated to rule out DDH. Asymmetry of skin folds in the gluteal and femoral regions is a sensitive but nonspecific indicator that suggests that the hip is abnormal (Figure 10.15).[7,25]

The Ortolani and Barlow maneuvers are the most reliable screening tests for evaluating hip stability (Figure 10.16). Although they are described as separate tests, in clinical practice, both maneuvers are done in a sequence, not as separate examinations. The infant must be in a supine position and on a relatively firm surface. These are not forceful examinations; a forceful examination only makes the infant cry and yields unreliable results. A crying, kicking infant can generate enough muscle strength by tightening the adductors and hamstrings to create a false result.[25] The cooperation of the infant and the patience of the examiner affect the accuracy of the examination.

The Ortolani maneuver is a test of hip reduction. It produces the reduction of the dislocated femoral head into the acetabulum by abduction. A helpful tip is to remember that the *O* in Ortolani means that the hip is "out." The maneuver is used to place the hip back into a normal position. With the infant positioned supine, the practitioner flexes the infant's knee and hip, and then grasps the thigh with the thumb positioned along the inner thigh and the third or fourth finger placed over the greater trochanter laterally. While the examiner abducts the hip, the finger on the greater trochanter presses up against the head of the femur, and the hand presses the shaft of the femur toward the mattress.

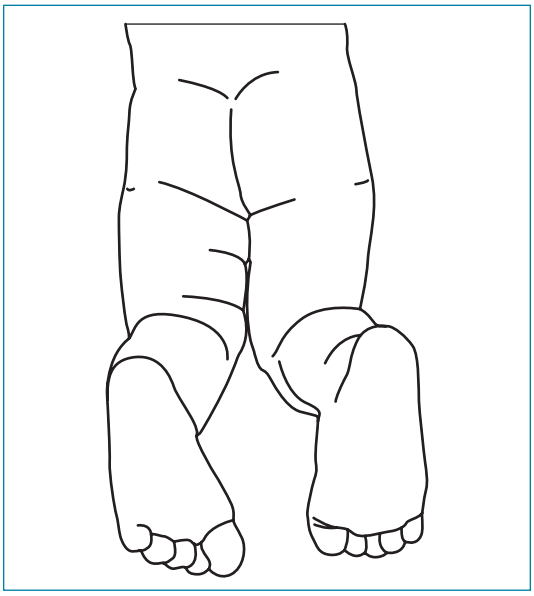

Figure 10.15 Asymmetric gluteal folds

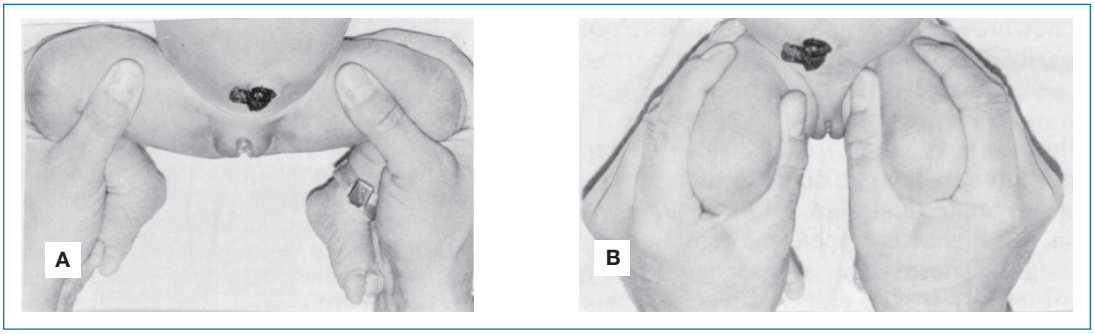

Figure 10.16 Ortolani and Barlow maneuvers (A) Ortolani: The fingers are on the trochanter and thumb grips the femur as shown. The femur is lifted forward as the thighs are abducted. If the femur head was dislocated, it can be felt to reduce. (B) Barlow: The thighs are adducted, and if the femur head dislocates, it will be both felt and seen as it suddenly jerks over the acetabulum.

Source: From Robertson WW. 2005. Orthopedics. In Neonatology: Pathophysiology and Management of the Newborn, 6th ed. MacDonald MG, Seshia MMK, and Mullett MD, eds., Philadelphia: Lippincott Williams & Wilkins, 1429. Reprinted by permission.[26]

A positive Ortolani maneuver is produced when a palpable "clunk" is noted, indicating that the femoral head has slipped from a dislocated position into the acetabulum. Higher pitched clicks and snaps can be heard and felt but are not associated with hip pathology; they usually result from movement of tendons, ligaments, or fluid in the hip joint.[18,25]

The Barlow maneuver determines whether the femoral head can be dislocated from the acetabulum and is the opposite of the Ortolani maneuver. The practitioner's hand position is the same as for the Ortolani maneuver, with the infant's hip and leg flexed. As the knee is brought to the center (adducted) from the abducted position, the practitioner's thumb pushes laterally on the upper inner thigh. A "clunk" indicates that the femoral head has slipped over the lateral edge of the acetabulum and demonstrates an unstable hip joint that is dislocatable. The amount of force needed to push the femoral head out of or into the acetabulum is minimal.

A variation of the Barlow maneuver is to stabilize the pelvis with one hand while using the other hand to try to move the thigh anteriorly and posteriorly (upward and downward) without flexing the hip. This maneuver enables the practitioner to determine whether the femoral head can be displaced posteriorly out of the acetabulum.

Legs

Palpate the legs to confirm the presence of the femur, tibia, and fibula. Fractures should be suspected when there is a history of difficult delivery or when palpation reveals irregularities in contour, crepitation, or masses resulting from hematoma formation.

Although birth trauma rarely causes a fracture of the femur (0.13/1,000 births), dislocation or avulsion of the femoral epiphysis can occur and should be suspected if there is pain on passive movement or little spontaneous movement of the leg.[17,18] Femoral length can be observed by testing for the Galeazzi (or Allis) sign. Keeping the feet flat on the bed and the femurs aligned, flex both of the infant's knees. With the tips of the big toes in the same horizontal plane, face the feet and observe the height of the knees.[7,11,25] It will be apparent if one knee is higher than the other, a positive sign. A discrepancy in knee height should also lead one to investigate for DDH (Figure 10.17).[5,7,27] Twenty-four percent of patients with congenital constricting bands have leg length discrepancy.[12]

The lower extremities are examined for length and shape. Congenital limb deficiencies are easy to identify, and an x-ray will confirm the deficiency (Figures 10.18 and 10.19). The lower extremities of newborns frequently have mild bowing and internal rotation of the lower leg because of intrauterine environmental conditions and fetal positioning. The bowed appearance is usually a combined rotational deformity caused by external rotation of the hip and internal tibial torsion from in utero positioning.[3,18] With the infant supine, draw an imaginary line connecting the anterior superior iliac spine with the midpatella, and continue down to the foot. If the line falls medial to the big toe, external tibial torsion is present. If the line falls lateral to the second toe, internal tibial torsion (a slight varus curvature) is present (Figure 10.20). Internal tibial torsion is a common

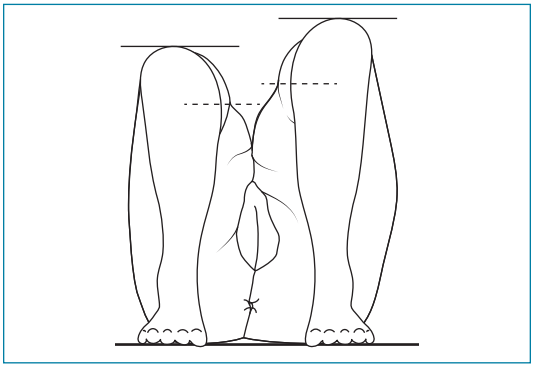

Figure 10.17 Allis sign

Source: Adapted from Alexander MM, and Brown MS. 1979. *Pediatric History Taking and Physical Diagnosis for Nurses*, 2nd ed. St. Louis, MO: Mosby-Year Book, 301. Reprinted by permission of the authors.[24]

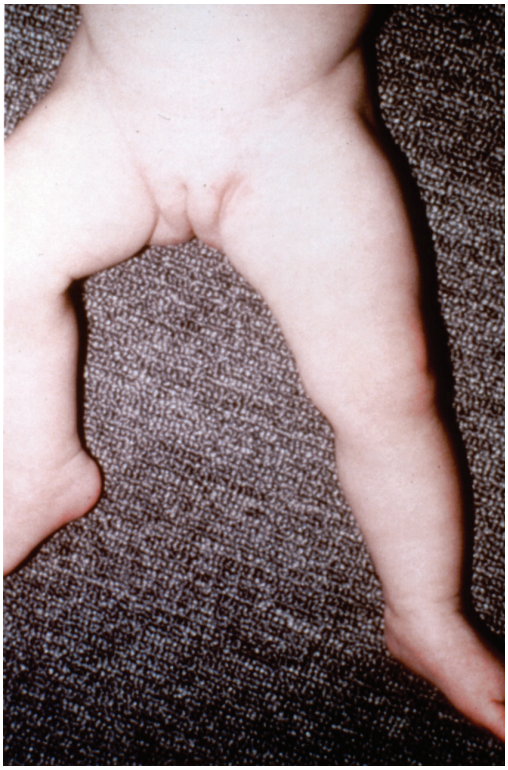

Figure 10.18 Infant with congenital limb deficiency of the leg

Courtesy of Mark Erickson, MD, Children's Hospital Colorado, Aurora, CO.

physiologic feature of almost all newborns and is the usual cause of intoeing in children from birth to 2 years of age. Because this is a physiologic rather than a pathologic condition, spontaneous recovery can be anticipated with normal growth and development.[3,18] Although lateral tibial bowing without a significant shortening of the extremity is considered normal in

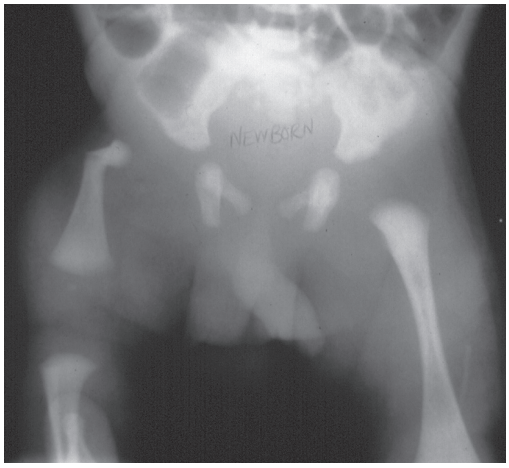

Figure 10.19 X-ray of congenital limb deficiency

Courtesy of Mark Erickson, MD, Children's Hospital Colorado, Aurora, CO.

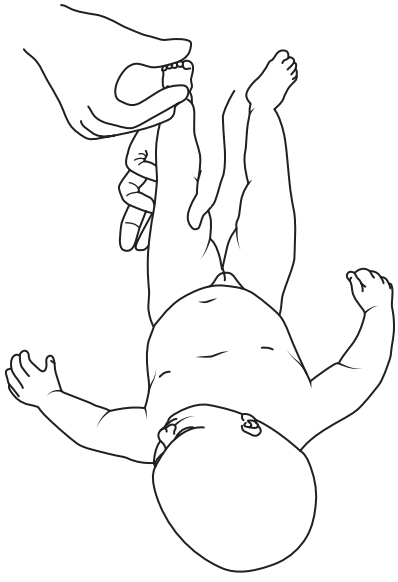

Figure 10.20 Examination for tibial torsion

the newborn, anterior tibial bowing is an abnormal finding, and an orthopedic consultation should be sought.

Ankles and Feet

Examination of the ankles and feet includes observation of the resting position and stimulation for active motion. To stimulate for motion, stroke the sole as well as the dorsal, medial, and lateral sides of the foot. Passive motion of the ankle in dorsiflexion and plantar flexion varies, depending on the infant's position in utero. For example, ankle and forefoot adduction, a positional deformity, can be differentiated from congenital equinovarus (clubfoot) malformation by passively positioning the foot in the midline and dorsiflexing it. A clubfoot or other structurally abnormal foot and ankle will not have a full range of motion and will resist dorsiflexion.

The toes should be examined for number, position, and spacing between them. Overlapping toes in infancy are usually hereditary; conservative treatment measures for mild cases may involve only stretching or stabilizing the toe by taping it in the correct position.[28] The soles of the feet should be inspected as part of the gestational age assessment. Most newborns are flat-footed as the result of a plantar fat pad (a pad of fat in the longitudinal arch of the foot), which gradually disappears during the first year of life.

MUSCULOSKELETAL ANOMALIES

Congenital anomalies of the musculoskeletal system may be evident as the absence of a part, extra parts, or malformed or malfunctioning tissue. Congenital anomalies usually affect the infant's movement, muscle tone, or posture. It is not within the scope of this chapter to discuss all the musculoskeletal anomalies the practitioner may encounter. However, many of the common problems and other special conditions seen in neonates are included in this section.

Anomalies of the Neck

Klippel–Feil Syndrome

Klippel–Feil syndrome is a defect of the cervical vertebrae in which there is both a decrease in the number of vertebrae and a fusion of two or more vertebrae. It can be detected throughout life, but those diagnosed in the newborn period tend to have upper cervical spine involvement as opposed to lower cervical involvement. The condition varies in severity depending on the number of vertebrae that are fused. The cause is unknown, although current theories involve generalized fetal insult, in utero vascular insult, or neural tube abnormalities.[29] In some instances, the syndrome is familial, indicating a genetic transmission.[30] On physical examination, the neonate's neck usually appears shorter than normal, and decreased range of motion is the most frequent clinical finding. Rotational loss is usually more pronounced than is the loss of flexion and extension. The classic signs on physical examination of short neck, low posterior hairline, and limitation of neck motion are seen in less than 50% of neonates with this syndrome.[4] The asymmetric motion may be confused with torticollis (see next section), but an x-ray of the neck will confirm the presence of the Klippel–Feil deformity. Other bony abnormalities, such as Sprengel deformity, congenital scoliosis, and congenital limb deficiencies, are associated with Klippel–Feil deformity.[4,29,30]

Torticollis

Congenital torticollis, or wry neck, is "a spasmodic, unilateral contraction of the neck muscles.[13]" It is an injury sustained to the sternocleidomastoid muscle or to a cervical spine abnormality. It is thought to be the result of birth trauma, intrauterine malposition, muscle fibrosis, or venous abnormality in the muscle.[3,4,29] This anomaly is not usually seen in the immediate newborn period, but a hematoma may sometimes be palpated, or soft-tissue swelling may be noted over the involved sternocleidomastoid muscle shortly after birth. Torticollis usually appears as a firm, fibrous mass or tightness in the sternocleidomastoid muscle

at approximately 2 weeks of age. The mass is 1 to 2 cm in diameter, hard, immobile, and felt in the midportion of the sternocleidomastoid muscle.[3,29] In infants with this condition, the head is tilted laterally toward the involved side, with the chin rotated away from the affected shoulder (Figure 10.21).[3,4,29] Infants with this anomaly should be further evaluated for associated conditions such as metatarsus adductus, hip dysplasia, and congenital anomalies of the cervical spine. If the mass is detected early, most neonates with congenital muscular torticollis can be treated with stretching exercises performed by the parents.[3,29,30] If the mass goes unnoticed, however, the torticollis may not be detected until there is plagiocephaly, or asymmetry of the face and skull development. If the torticollis persists or goes untreated, there is a flattening of the occiput on the opposite side and a flattening of the frontal bones on the side of the lesion.[4,29,30]

Spinal Deformities

Congenital Scoliosis

Scoliosis in the neonate may range from undetectable to very severe. It is not chromosomal or inherited, but rather an embryonic defect. It is a condition in which there is a lateral S- or C-shaped curvature of the spine that is usually associated with a rotational deformity of the spine and ribs. The structural basis for congenital scoliosis is a failure of vertebral formation, segmentation, or a variety of both. The failures can be in any area of the vertebral body (Figure 10.22). Failures of formation indicate the absence of a portion of the vertebra, such as a hemivertebra. Failure of segmentation is the absence of the normal separation between the vertebras. Females are affected more often than males, and their curvature is more likely to worsen. If the defect goes undetected, severe deformities can develop and affect neurologic function as well as cosmetic appearance.[5] Upon diagnosis, the infant should also be evaluated for genitourinary tract anomalies (unilateral renal agenesis being the most common) because there is an increased incidence (20%–30%) of these anomalies with congenital vertebral anomalies.[4] Klippel–Feil syndrome and Sprengel deformity of the scapula are also seen with congenital scoliosis.[29,30]

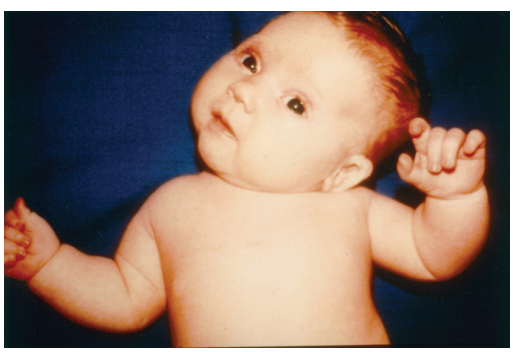

Figure 10.21 Torticollis

Courtesy of Mark Erickson, MD, Children's Hospital Colorado, Aurora, CO.

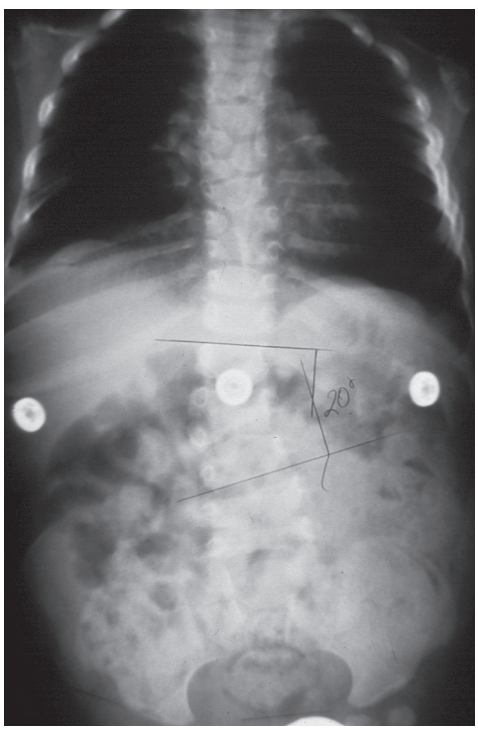

Figure 10.22 X-ray of congenital scoliosis

Courtesy of Mark Erickson, MD, Children's Hospital Colorado, Aurora, CO.

Myelomeningocele

Myelomeningocele is the most severe type of spina bifida. It is a congenital neural tube defect in which the bones of the spine do not completely form. The defect usually presents as a failure of closure at the caudal (tail) end of the vertebral column, permitting the meninges and sometimes the spinal cord to protrude into a sac-like structure (Figure 10.23). Skin disruption is not always present; however, any soft mass noted over the spine or just off the midline must be examined closely to rule out myelomeningocele. Because the functional deficit of the lower extremities is linked to the level of involvement of the myelomeningocele, it is important to examine the muscle function of the lower extremities. Other congenital spinal cord or musculoskeletal defects that may also be seen with myelomeningocele include syringomyelia and hip dislocation.

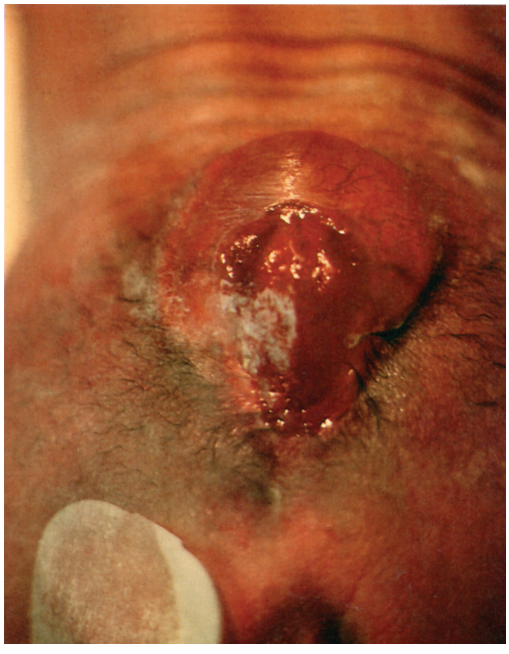

Figure 10.23 Myelomeningocele

Source: From Clark DA. 2000. Atlas of Neonatology. Philadelphia: Saunders, 131. Reprinted by permission.[16]

Upper Extremity Anomalies

Sprengel Deformity (Congenital Elevated Scapula)

Sprengel deformity is one of the more common congenital anomalies of the shoulder girdle. It is characterized by an abnormally small, elevated scapula.[29,31] The elevation may be unilateral or, in 10% to 30% of cases, bilateral.[31] The asymmetry of the shoulder seen with unilateral involvement makes diagnosis relatively easy. On examination, the scapula is noted to be hypoplastic, elevated, and malrotated so that, when palpated, the vertebral border lies superiorly and more horizontal than normal. The angle of the scapula may give the newborn the appearance of a webbed neck or a fullness at the base of the neck.[31] There is usually limitation of shoulder motion, particularly in abduction and forward flexion.[29,31] Internal and external shoulder rotation may be only slightly affected. Sprengel deformity appears to be a familial predisposition, and in 70% of cases another congenital abnormality is present such as congenital spinal problems, renal anomalies, torticollis, and Klippel–Feil syndrome.[19,25]

Cleidocranial Dysostosis

Cleidocranial dysostosis, also known as *cleidocranial dysplasia* or *mutational dysostosis*, is a rare defect that affects the growth of bones in all parts of the body. It can occur either spontaneously or be inherited as an autosomal dominant trait with no predilection of gender or ethnic group. It is characterized by complete or partial absence of the clavicles (Figure 10.24).[15,25,32,33] The chest is narrow and the shoulders hang. It is usually caused by a mutation of the $RUNX_2$ (core-binding factor alpha-1) gene, located at chromosome 6p21. This gene encodes a protein necessary for the correct functioning of osteoblast cells, which regulate bone differentiation.[32] Bilateral absence of the clavicles, which occurs in 10% of cases, or segmental loss of the lateral or middle portion of the clavicles allows for excessive scapulothoracic motion where the newborn's shoulders touch in midline without discomfort.

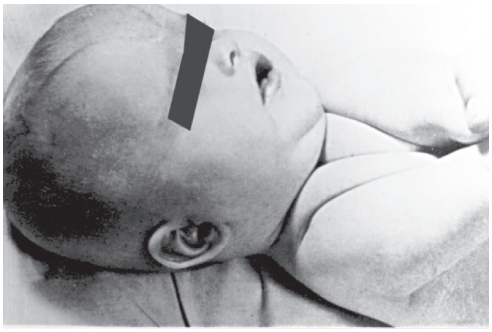

Figure 10.24 Cleidocranial dysostosis

Source: From Swaiman KF, and Wright FS. 1982. *The Practice of Pediatric Neurology.* Philadelphia, PA: Mosby, 441. Reprinted by permission.[33]

Complete absence of the clavicle is usually accompanied by defective ossification of the cranium, large fontanels, and delayed closure of the sutures.[15,32] As the infant ages, there is a delay in the eruption and/or an absence of permanent teeth. Although the deformities may not be cosmetically pleasing, there is usually little functional disability. No treatment of the newborn is required, but genetic counseling is recommended for the parents.[25,32]

Brachial Plexus Palsy

BPP is one of the more common birth injuries detected on the newborn examination, with an incidence of 0.1% to 0.3% of live births.[3] BPP injuries are usually unilateral and are seen more often on the left side.[34] The incidence of BPP has not decreased over the last three decades, which may be partly due to an increase in population birth weights. However, the incidence of lower plexus lesions has decreased significantly with the decline in vaginal breech births.

Most cases of BPP are thought to be the result of prolonged and difficult labor involving traction and lateral flexion of the neck.[17,34] Brachial plexus injury has also been attributed to abnormal in utero forces on the posterior shoulder of the fetus as it passes over maternal bony prominences such as the sacral promontory. Increased in utero pressure and traction have also been proposed as the cause in an abnormal uterus such as a bicornuate or fibroid uterus.[17] Other perinatal risk factors include prolonged labor, large maternal body mass index, large-for-gestational-age fetuses, shoulder dystocia, and those with a breech presentation.[3,4,34] BPP, therefore, is seen most often in large neonates, who, because of their size, are vulnerable to stretching injuries to the components of the brachial plexus.[17,31] Shoulder dystocia in vertex deliveries and difficult arm or head extraction in breech deliveries increases the risk for brachial plexus injuries.[17,18,31] A cesarean delivery does not protect against BPP as 10% of affected infants have a history of cesarean delivery.[3]

There are three types of BPP, each with a different clinical presentation, depending on the site of injury and extent of neural injury:

1. Upper lesion injuries are the most frequent type of brachial plexus injury and involve complete or partial paralysis of the shoulder muscles as a result of injury to cranial nerves 5 (CN V), 6 (CN VI), and occasionally 7 (CN VII). This type of palsy is known as *Erb palsy*, and the clinical presentation frequently is referred to as the *bad shoulder, good hand* scenario.[3,17] An infant with this type of upper arm paralysis holds the affected arm adducted and internally rotated, with extension at the elbow, pronation of the forearm, and flexion of the wrist (Figure 10.25). The grasp reflex remains intact, but the Moro reflex is absent on the affected side.
2. A second type, Klumpke palsy, is an example of a lower lesion injury presenting clinically as the *good shoulder, bad hand* scenario. This palsy involves cranial nerve 8 (CN VIII) and thoracic nerve 1 (T1), with complete or partial paralysis of the forearm and hand muscles. This lower arm paralysis is rare. When the lower plexus is involved, the shoulder is in a relatively normal position with the wrist and hand flaccid, having little or no control.
3. The third type of BPP, complete paralysis of the arm, involves injury to the plexus at all levels. It includes paralysis of the wrist and hand, in addition

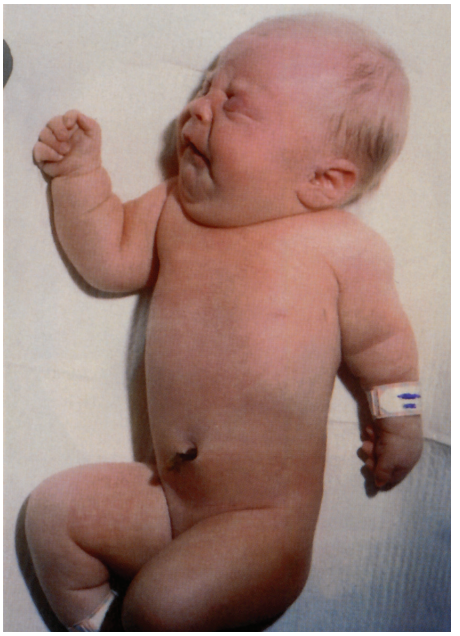

Figure 10.25 Brachial plexus palsy (Erb's)

Source: From Clark DA. 2000. Atlas of Neonatology. Philadelphia: Saunders, 138. Reprinted by permission.[16]

to shoulder and elbow dysfunction.[17,35] Because of the possibility of birth trauma as an etiology for the palsy, skeletal injury may coexist and should be ruled out radiographically.[3]

In cases of BPP in which the nerve roots are not disrupted, infants regain neurologic function within several days as the hemorrhage and edema in the area resolve. Gentle handling and protection of the arm for the first 7 to 10 days will help to avoid additional injury to the plexus. Initial treatment is aimed at preventing contractures of the shoulder, elbow, forearm, and hand during the observational recovery phase by gentle passive range-of-motion exercises. It is not necessary to splint or immobilize the affected arm as it has not proven beneficial to prevent contractures and further stretching of the plexus.[34] Although most infants gain significant functional improvement by 3 months of age, close examination usually reveals tightness of the shoulder on internal rotation, difficulties in supination of the forearm, and abduction of the shoulder.[3,17]

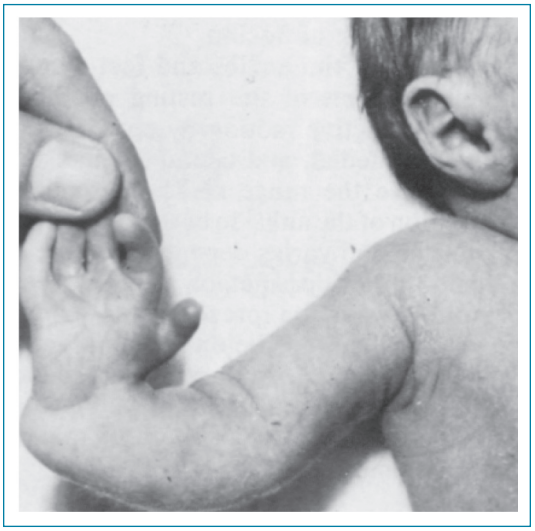

Figure 10.26 Congenital absence of the radius

Source: From Robertson WW. 2005. Orthopedics. In Neonatology: Pathophysiology and Management of the Newborn, 6th ed., MacDonald MG, Seshia MMK, and Mullett MD, eds. Philadelphia: Lippincott Williams & Wilkins, 1432. Reprinted by permission.[26]

If improvement is not noted in the first few months, an electromyography and nerve conduction studies are performed to determine the extent of the damage. Secondary treatment involves nerve repair to restore neurologic function that has not occurred spontaneously. Infants with total plexus involvement require brachial plexus exploration and reconstruction if there is no return of biceps function at 3 months of age. If the arm remains flaccid at 2 to 3 months of age for infants with upper BPP, surgery is undertaken at 3 to 6 months of age.[18,35] In newborns with peripheral nerve disruption or nerve root disruption, early intervention with microsurgery is recommended. Approximately 20% of brachial plexus injuries require surgery, and 90% of these infants achieve useful function of muscle groups above the elbow.[17,35]

Congenital Absence of the Radius (Radial Dysplasia)

Congenital absence of the radius is sometimes referred to as *radial dysplasia* and is easy to recognize. Clinically, the infant presents with the hand and wrist deviated 90° or more (Figure 10.26). The forearm is shortened, with

bowing of the ulna. The thumb is usually absent or hypoplastic. There is marked limited movement of the hand, wrist, and forearm. It occurs in one of 30,000 to one of 100,000 live births and is seen in males more often than females.[29,31,36] Although radial hypoplasia alone may be inherited sporadically, infants presenting with this anomaly should be further evaluated for associations such as VACTERL (**v**ertebral anomalies, **a**nal atresia, **c**ardiac abnormalities, **t**racheo**e**sophageal abnormalities, **r**enal abnormalities, and **l**imb anomalies), Fanconi anemia, and Holt–Oram syndrome.[31,36] Treatment comprises both operative and nonoperative options and is recommended for cosmetic and functional reasons.[29,31,36] A consultation with a pediatric hand surgeon is required.

Lower Extremity Anomalies

Developmental Dysplasia of the Hip

DDH is one of the most significant deformities of the newborn period and the most common neonatal hip disorder. It covers a spectrum of conditions that arise from abnormal development of the hip joint. These conditions range from minimal instability (in which the femoral head remains in the acetabulum) to irreducible dislocation (in which the femoral head loses contact completely with the acetabular capsule and is displaced over the fibrocartilaginous rim). DDH is thought to be caused by lack of acetabular depth, ligamentous laxity limited to the hip joint that is influenced by hormonal and genetic factors, and/or abnormal intrauterine positioning, as in intrauterine breech position.[25,36]

Approximately 30% to 50% of all DDH occurs in infants born after breech presentation.[2,36] Approximately 2.5 to 6.5 infants/1,000 live births develop hip dysplasia, but the true incidence of DDH can only be presumed as it is not always detectable at birth. The overall incidence of detectable dysplasia with evidence of instability has been reported to be as high as one of 100 newborns and with dislocation, one to two per 1,000 newborns.[2,25,37] However, genetic factors and ethnic practices play a key role in the incidence of DDH. African and Asian infants have a very low incidence, wereas Native Americans and Eastern Europeans have an incidence reported as high as 25 to 50/1,000 live births.[25,38] These differences may be due less to genetic predisposition and more to child-rearing practices, such as swaddling, that keep the newborn's hips in adduction and extension.[25,38] Females are more prone to the condition than males, and the left hip is affected twice as often as the right hip.[19,37,38] A family history revealing that a parent had a dislocated hip as a child or that an older sibling has hip dysplasia increases the risk for dislocated hip in the infant.[2,29,37] DDH is also more common in infants with other orthopedic conditions, such as torticollis, and in those with congenital foot deformities, such as clubfoot and metatarsus adductus.[29,36,38]

The clinical manifestations of DDH depend on the severity of the condition. Findings that should lead a practitioner to investigate further include (a) asymmetry of gluteal or thigh folds, (b) limb length descrepancy (Galeazzi/Allis sign), (c) positive Barlow maneuver (hip reduced but dislocatable), and (d) positive Ortolani sign (hip dislocated, but reducible).[36] Early diagnosis and treatment appropriate to the specific anomaly are important to create normal hip anatomy and function. Treatment is easiest and most effective if started in the first 6 months of life. There is no uniformly accepted method for diagnosis of DDH during the newborn period. Sixty percent to 80% of hip deformities identified in children by clinical examination resolve on their own without intervention.[5,39] The most effective diagnostic tool for screening hip joint problems during the first month of life is thought to be ultrasonography. Ultrasounds are completed on infants with high-risk factors, such as females born in the breech position or those with positive Ortolani or Barlow maneuvers.[2] However, the U.S. Preventive Services Task Force (USPSTF) states that 90% of the hip abnormalities identified by ultrasound resolve on their own, and there is insufficient evidence to recommend routine screening of asymptomatic newborns as a means of preventing adverse outcomes.[5,39] Radiographs of newborns with suspected DDH are of limited value because the femoral heads do not

ossify until 4 to 6 months of age.[2] When treatment is initiated early using simple devices such as the Pavlik harness (Figure 10.27) and the von Rosen splint, there is a 95% success rate.[2,18,38] The longer the dislocation remains untreated, the greater the chance of problems in returning the femoral head to its normal position, and the less satisfactory the results. Assessment techniques for and signs of DDH were discussed earlier in this chapter.

Congenital Absence of the Tibia or Fibula

Although congenital absence of a long bone is unusual, when it does occur, the deformity is easily recognized. Tibia or fibula absence may be partial or complete. When a portion of the bone is present, the deformity is likely to be less severe. In the absence of the tibia, the clinical presentation is mild to marked shortening of the lower leg. The knee is unstable and has a flexion contracture. The foot may be normal or fixed in a mild to severe varus position.[40] In absence of the fibula, the clinical presentation is shortening of the involved leg, with bowing of the tibia anteriorly and medially (Figure 10.28). The foot deformity is often severe, with a valgus position.[18,41] Treatment depends upon the severity of the condition and focuses on the problems of foot deformity and leg length discrepancy. These deformities should be seen by an orthopedist early in the neonatal period. Many infants with congenital limb deficiencies frequently have other associated anomalies that represent an inherited syndrome. Because of the high potential for genetic transmission of the disorder, the parents should be offered genetic counseling.[40]

Genu Recurvatum

Genu recurvatum, a rare anomaly with an estimated incidence of one per 100,000 live births, is a congenital dislocation or hyperextension of the knee (Figure 10.29). It is thought to result from a frank breech position in utero (41% of otherwise normal newborns exhibiting this anomaly were breech). It can be the result of a prenatal developmental defect, a specific disease of the muscles or nerves of the leg, or a manifestation of generalized joint laxity, as seen in "floppy baby" syndrome. It can be associated with oligohydramnios, anomalies of the elastic tissues,

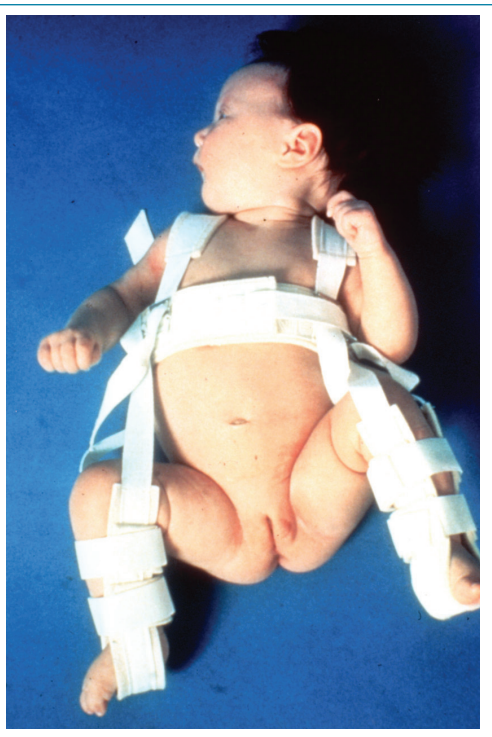

Figure 10.27 Pavlik harness

Courtesy of Mark Erickson, MD, Children's Hospital Colorado, Aurora, CO

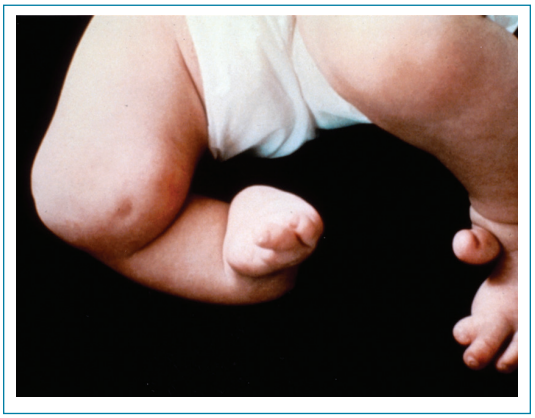

Figure 10.28 Congenital absence of the fibula

Courtesy of Mark Erickson, MD, Children's Hospital Colorado, Aurora, CO.

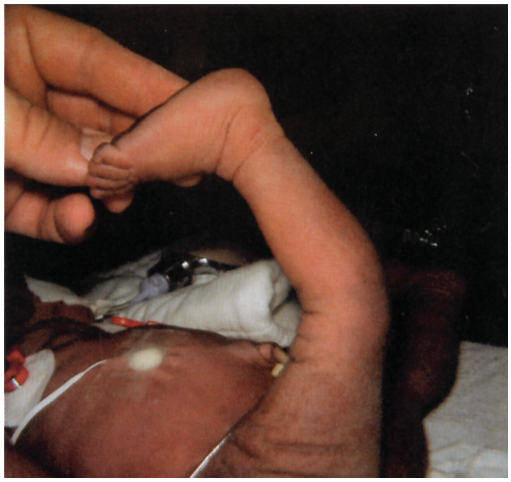

Figure 10.29 Genu recurvatum

Source: Form Clark DA. 2000. *Atlas of Neonatology.* Philadelphia: Saunders, 9. Reprinted by permission.[16]

and fibrosis of the quadriceps muscle along with deficient hamstrings.[36,42] More common in females, genu recurvatum is usually seen bilaterally. Treatment for mild deformities includes physical therapy and progressive reduction of the hyperextension by means of serial splinting. Other infants may require serial casting to hold the knee in flexion.[42] Genu recurvatum associated with dislocation of the knee requires surgery. Because early treatment prevents further deformity or interference with normal function, an orthopedic consultation should be initiated early.

Metatarsus Adductus

The most common congenital foot anomaly, metatarsus adductus is a deformity of the forefoot in which the metatarsal bones are deviated medially. The condition is probably the result of intrauterine positioning. Multiple gestation pregnancies and oligohydramnios, two conditions that decrease the room available for fetal movement, predispose the newborn to metatarsus adductus.[7,11] It is associated with DDH in 10% to 20% of cases.[2,36] Other than the deviation, there are no pathologic changes in the structure of the foot. It occurs equally in both males and females and is seen bilaterally in approximately 50% of newborns who exhibit the anomaly.[36] Metatarsus adductus

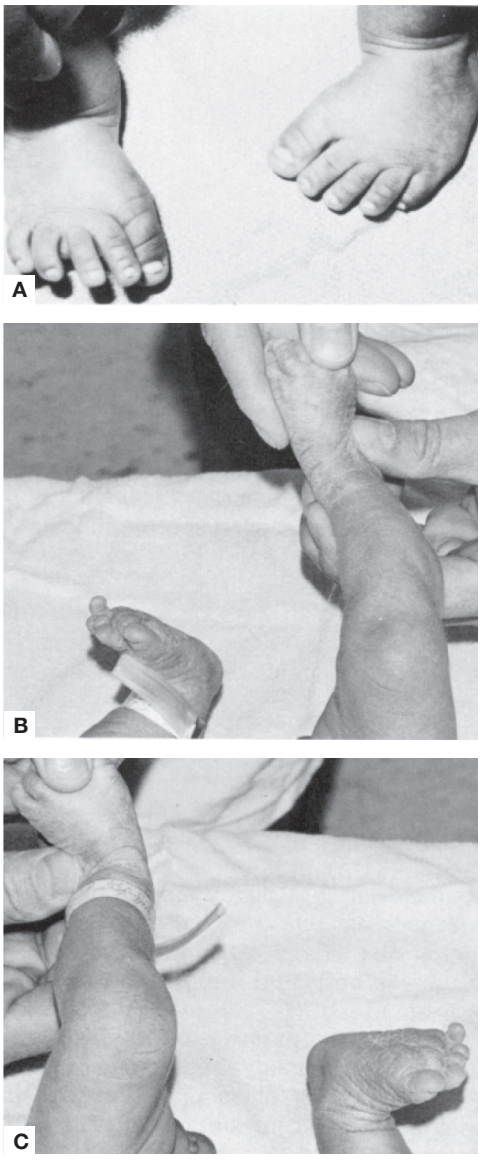

Figure 10.30 Metatarsus adductus (A) Structural metatarsus adductus. (B) Structural metatarsus adductus. The forefoot does not abduct beyond neutral. (C) Positional metatarsus adductus. The forefoot abducts beyond the midline.

Source: Robertson WW. 2005. Orthopedics. In *Neonatology: Pathophysiology and Management of the Newborn*, 6th ed., MacDonald MG, Seshia MMK, and Mullett MD, eds. Philadelphia: Lippincott Williams & Wilkins, 1434. Reprinted by permission.[26]

may be a positional (flexible) deformity with no bony abnormality involved or a structural (fixed) deformity (Figure 10.30). In a positional

deformity, the forefoot is very mobile and can be easily abducted; the heel is likely to be in a neutral position. In a structural deformity, the arch appears to be greater than normal, and there may be a medial crease at its middle portion; the forefoot usually cannot be abducted beyond the midline (neutral position). In infants with severe structural anomaly, the heel (hindfoot) is in a valgus position. Approximately 85% of neonatal metatarsus adductus deformities resolve by 3 years of age and develop normal foot position and function without treatment.[4,11,25,36] Positional deformities will correct spontaneously or can be treated with stretching exercises. In a rigid foot, an orthopedic consultation is necessary for early treatment because manipulative stretching exercises and casting may be required. Surgery is indicated only when casting is unable to produce a flexible foot.[28,29]

Clubfoot (Talipes Equinovarus)

Clubfoot (Figure 10.31) is a complex foot deformity in which the foot turns inward and downward and is readily apparent at birth. It is one of the most common congenital orthopedic anomalies, whose incidence varies with race and gender. In Caucasians, the birth frequency is approximately one per 1,000 live births, with males affected twice as often as females.[5,28,36] The highest incidence is seen in Polynesians at a rate of 6.8/1,000 births. Asian infants are the least likely to present with clubfoot, with it occurring at a rate of 0.6/1,000 live births.[28] The cause of congenital clubfoot is unknown, but it is likely a combination of genetic and environmental factors. It may be associated with chromosomal abnormalities or congenital syndromes. However, most cases are idiopathic and found in normal infants in whom there is no genetic or chromosomal abnormality and no other extrinsic cause can be found. Neural, muscular, and osseous abnormalities are other possible predisposing conditions. Within affected families, the probability is approximately 3% for siblings and 20% to 30% for the offspring of affected parents.[29,36]

Clinical presentation of the deformity is characterized by three primary components: (a) adduction of the forefoot (points medially) and accounts for approximately 95% of idiopathic cases, (b) pronounced varus of the heel, and (c) downward pointing of the foot and toes (equinus positioning). Clubfoot may be unilateral, but involvement is bilateral in about 50% of cases.[28,29] There are variations in the severity of clubfoot. Some are relatively flexible and correctable with conservative measures such as exercises and serial casting. Surgical correction is necessary for severe deformities or when nonoperative treatment methods are unsuccessful.[5,29] Treatment can often be started in the nursery. An orthopedic consultation should be initiated as soon as the diagnosis is made.

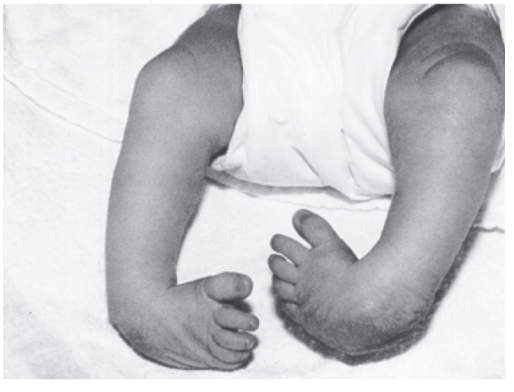

Figure 10.31 Clubfoot

Courtesy of Carol Trotter, St. John's Mercy Medical Center, St. Louis, MO.

Conditions Affecting Upper and/or Lower Extremities

Congenital Constricting Bands

Constricting band syndrome, also known as *amniotic band syndrome; amnion rupture sequence; amniotic deformity, adhesions, mutilations (ADAM) complex;* and *Streeter dysplasia*, presents as a band encircling the arms, legs, fingers, or toes. It can vary from mild, shallow indentations of the soft tissue to severe constrictions causing partial or complete amputation (Figures 10.32–10.35). Occasionally, craniofacial structures are affected, resulting in cleft lip, cleft palate, and other facial

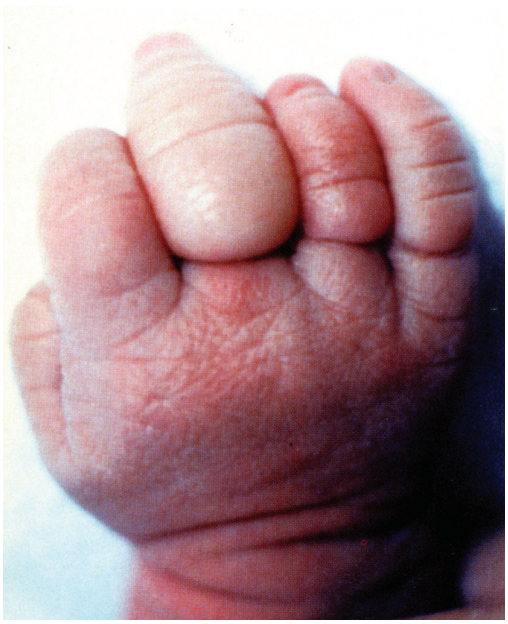

Figure 10.32 Congenital constricting bands (amniotic bands)

Source: From Clark DA. 2000. *Atlas of Neonatology.* Philadelphia: Saunders, 23. Reprinted by permission.[16]

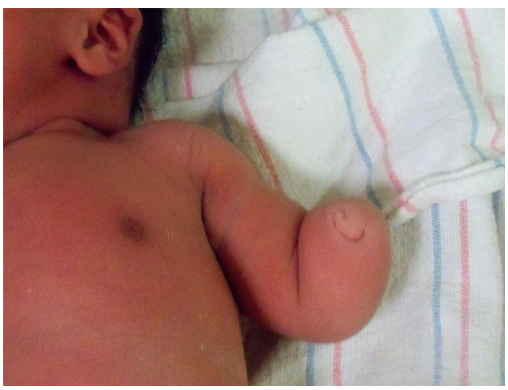

Figure 10.33 Congenital constricting bands (amniotic bands)

deformities.[31] No two cases of amniotic band syndrome are exactly alike and the associated syndromes are highly variable. This deformity is estimated to occur in one per 15,000 live births, with the upper extremities more frequently involved than the lower extremities.[31,36,43] There are no gender or

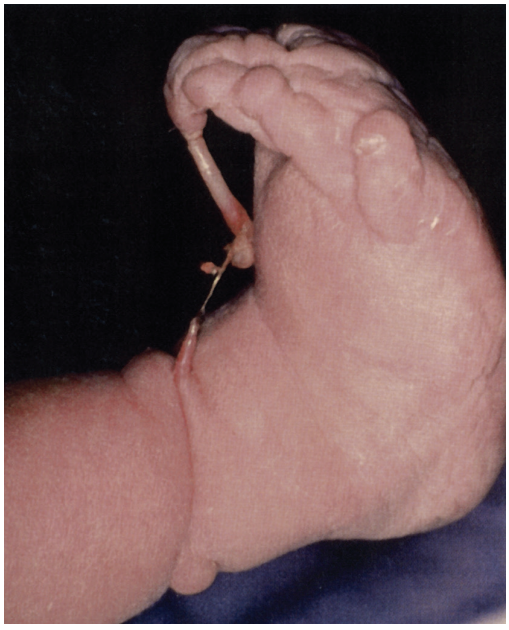

Figure 10.34 Congenital constricting bands (amniotic bands)

Source: From Clark DA. 2000. *Atlas of Neonatology.* Philadelphia: Saunders, 24. Reprinted by permission.[16]

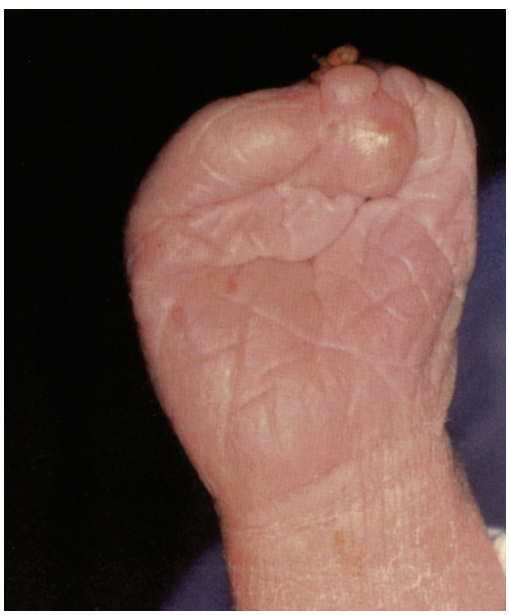

Figure 10.35 Amniotic bands resulting in finger amputation

Source: Clark DA. 2000. *Atlas of Neonatology.* Philadelphia: Saunders, 23. Reprinted by permission.[16]

ethnic predispositions. It is associated with other musculoskeletal deformations in 50% of cases; the most common being clubfoot.[36] The etiology is unknown, but the majority of evidence suggests that there is an abnormal attachment of the amnion to the fetus, because the amnion has either lost its integrity or ruptured and adhered to a fetal body part, causing compression or amputation.[19,36] Amniotic band syndrome can develop at any point during the first 20 weeks of pregnancy but the most severe complications occur when it develops early in the first trimester. Treatment depends on the severity of the condition. Severe bands may need to be treated as an emergency, especially if there is evidence of vascular or lymphatic obstruction. A surgical consultation is needed for these cases. Physical and occupational therapy are needed to ensure optimal use of affected fingers, toes, arms, and legs.[43]

Syndactyly

Congenital webbing of the fingers or toes—syndactyly—is one of the most common congenital anomalies of the upper extremities with an incidence of one per 2,000 to 3,000 live births (Figure 10.36).[25,36] This anomaly affects males twice as often as females and is 10 times more common among Caucasians than among infants of African descent. The frequency of bilateral and unilateral syndactyly is equal. Syndactyly is caused by failure during the sixth to eighth week of gestation of the usual necrosis of skin that normally separates the fingers. Syndactyly appears to be sporadic in most cases, with approximately 10% to 40% familial with variable penetrance. It can occur as an isolated anomaly or be associated with other congenital anomalies such as Apert and Streeter dysplasia.[29] The severity of involvement varies from minimal "bridging" between adjacent fingers and toes to complete webbing of the hand or foot. The more severe the syndactyly, the greater the likelihood of bony abnormalities as well. Syndactyly of the toes does not interfere with function, but may be unacceptable cosmetically. Although surgical treatment is not required, it may be requested by the parents. Treatment for syndactyly of the fingers depends on the severity of the webbing and the presence or absence of bony abnormalities. When multiple fingers are involved, function may deteriorate as the fingers grow. Early correction should therefore be considered, especially if the fingers affected are of unequal length. An orthopedic consultation is needed.

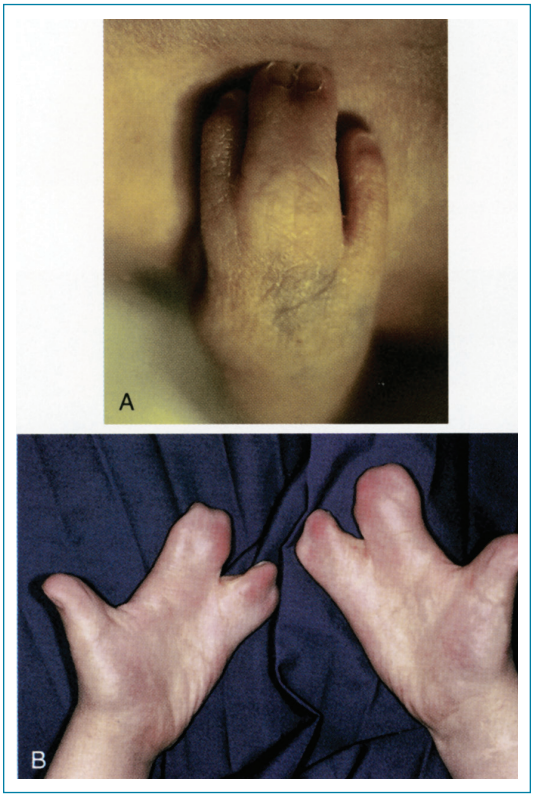

Figure 10.36 Two variations of syndactyly of the fingers

Source: Clark DA. 2000. *Atlas of Neonatology*. Philadelphia: Saunders, 222. Reprinted by permission.[16]

Polydactyly

Extra digits—polydactyly—are common duplication abnormalities affecting both the hands and the feet (Figures 10.37 and 10.38), with an incidence of approximately 1.7 to two per 1,000 live births.[36,44] Postaxial (small finger, small toe) polydactyly has a variable racial incidence. It is seen more commonly in African American infants, with an incidence of

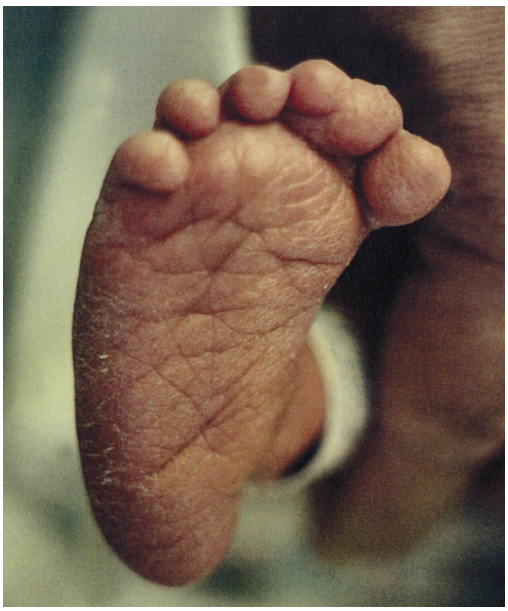

Figure 10.37 Polydactyly of the toes

Source: Clark DA. 2000. *Atlas of Neonatology.* Philadelphia: Saunders, 225. Reprinted by permission.[16]

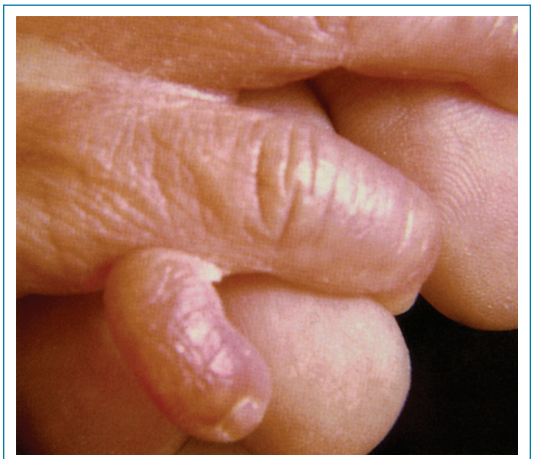

Figure 10.38 Polydactyly of the fingers

Source: Clark DA. 2000. *Atlas of Neonatology.*Philadelphia: Saunders, 222. Reprinted by permission.[16]

3.6 to 13.9 cases per 1,000 live births, whereas the incidence in Caucasian infants is 0.3 to 1.3 per 1,000 live births. Polydactyly is bilateral in 50% of cases and affects males slightly more than females.[44] It occurs more frequently in infants who have a positive family history.[41,44] Polydactyly can occur as an isolated anomaly or be inherited as an autosomal dominant trait. It can also be a manifestation of a multiple malformation syndrome such as trisomy 13, trisomy 21, or Meckel–Gruber syndrome.[14,41] Postaxial polydactyly in African Americans is almost always an isolated malformation. Postaxial polydactyly in Caucasians without a family history is infrequent and associated with chromosomal abnormalities, or other skeletal malformations.[31,36]

The most common type of polydactyly is a floppy digit or skin tag on either the radial or the ulnar side of the hand. Polydactyly may, however, involve the duplication of a normal-looking digit, giving the infant a functional six-fingered hand or a foot with six toes. Treatment depends on the extent of the anomaly. Consultation is required so that therapeutic decisions can be based on functional as well as cosmetic considerations.

Skeletal Dysplasias: Achondroplasia

Skeletal dysplasias are a group of conditions associated with various abnormalities of the skeleton. These conditions are caused by disturbances of bone growth that begin during the early stages of fetal development. Achondroplasia, a form of short-limbed dwarfism (Figures 10.39 and 10.40), is the most common form of skeletal dysplasia with an incidence estimated to be one per 15,000 to one per 40,000 live births and affecting both males and females equally.[45,46] The disorder has an autosomal dominant inheritance pattern with most cases appearing as spontaneous mutations. Approximately 80% of infants with achondroplasia have normal-sized parents. Infants with heterozygous achondroplasia usually have normal intelligence with a normal life expectancy. Although homozygous achondroplasia occurs, it is a lethal condition as a result of respiratory insufficiency caused by an underdeveloped rib cage and neurologic deficits secondary to cervicomedullary stenosis.[47] Achondroplasia is caused by mutations in the gene for fibroblast growth factor receptor 3 (FGFR3) of chromosome 4. FGFR3 is responsible for providing instructions for making a protein that converts cartilage to bone. The mutations

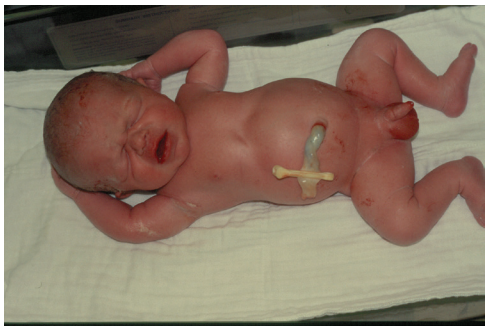

Figure 10.39 Achondroplasia: Flattened nasal bridge, frontal bossing, and shortened limbs

Source: Courtesy of L. Lilien, MD, Phoenix, AZ

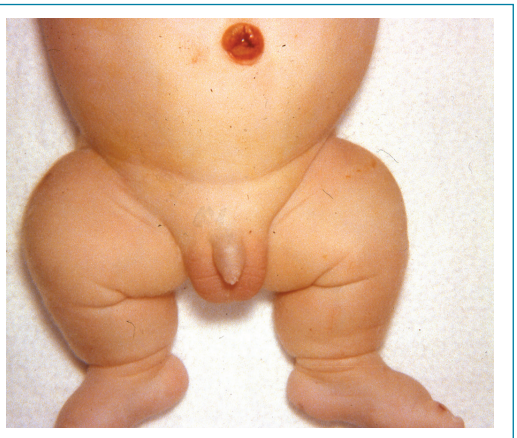

Figure 10.40 Achondroplastic dwarf: Short femurs and tibias

Source: From Clark DA. 2000. *Atlas of Neonatology*. Philadelphia: Saunders, 227.[16]

cause the FGFR3 protein to be overly active, which interferes with skeletal development and disturbances in bone growth.[45–47] Infants with achondroplasia have short upper arms and thighs, limited range of motion at the elbows, an enlarged head with frontal bossing, midface hypoplasia, a depressed nasal bridge and an average-sized torso with a protuberant abdomen. Hyperextensibility of the knees and hands is common. The hands are short and broad with the fingers exhibiting a three pronged appearance (trident configuration) at birth due to an inability to oppose the third and fourth fingers. Infants usually lie with hips abducted and lower legs have some degree of bowing (genu varum). In newborns, mild to moderate hypotonia is a typical finding and contributes to the delay of developmental motor milestones.[45,46]

Because most children with achondroplasia have delayed motor milestones, problems with persistent or recurrent middle ear infections, and bowing of the lower legs, treatment includes frequent monitoring and follow-up care. Although seen less often, infants may have serious health consequences related to hydrocephalus, craniocervical junction compression, upper airway obstruction, or thoracolumbar kyphosis. Anticipatory care should be directed at identifying infants who are at high risk and intervening to minimize risk of complications and prevent serious sequelae. Evaluations by genetic, orthopedic, and neurologic consultants are required for optimal outcomes.[45–47]

SUMMARY

The examination of the musculoskeletal system provides a wealth of information about the overall development of the infant in utero, as well as the newborn's potential for normal development and function. Many of the common congenital anomalies of infancy are found in the musculoskeletal system. Although these abnormalities may not interfere with vital functions (as do those of the respiratory, cardiovascular, or other systems), they are a frequent cause of parental anxiety. The more accomplished the examiner becomes at performing the musculoskeletal examination, the easier it is to recognize deviations from normal, potential problems, and the need to initiate early interventions.

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Neurologic Assessment

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11

The neurologic evaluation is a critical part of neonatal assessment. A single examination may verify the presence of normal or abnormal neurologic responses. Serial follow-up of the abnormal neurologic examination is necessary to validate unusual findings identified on the first examination and to document changes or disappearance over time. Steady improvement in responses and disappearance of abnormal responses during the neonatal period offer a better prognosis than static abnormal responses.

A common approach to assessment of neurologic status is through the systematic testing of specific functions of the nervous system, including the motor system, reflexes, sensory system, and cranial nerves. This approach is followed here. More formal neurologic assessments, designed as clinical and research tools, are available. The neurologic evaluation developed by Amiel-Tison is designed to evaluate neonates at term or corrected term age during the first year of life and emphasizes neuromotor function.[1] Prechtl also developed and validated a neurologic examination for the term neonate.[2] Another comprehensive neurologic examination was designed by Lilly and Victor Dubowitz and is applicable to both term and preterm neonates.[3] The Dubowitz examination places heavy emphasis on evaluation of movement, tone, and also includes some behavioral items evaluated on the Brazelton assessment, further discussed in Chapter 12.

MATERNAL AND FAMILY HISTORY

The neurologic examination should be preceded by a thorough review of the history for genetic or neurologic problems of the family, maternal medical difficulties, as well as the use of medications, alcohol, or illicit drugs. Note any results of chromosome analysis, prenatal ultrasounds, and tests of fetal well-being and maturity. Review the intrapartum course for abnormal presentation, prolonged labor, precipitous delivery, fetal distress, and difficult or operative extraction. Anesthetic agents and medications administered around the time of delivery may also affect the neurologic examination and should be noted. The history is also surveyed for Apgar scores, cord blood pH, presence of depression at birth, difficulties in transition, feeding ability, and gestational age. An accurate assessment of gestational age (Chapter 3) is essential for appropriate interpretation of posture, tone, and reflexes. Neurologic response varies predictably at different stages of maturity.

OBSERVATION

Observing the neonate before disturbing him or her is necessary to evaluate for the presence of dysmorphic features and evidence of birth trauma and/or skin lesions. Posture and activity are also observed.

Evidence of *birth trauma* may include cephalohematoma (see Figure 5.8), a depressed area of the skull, forcep marks (see Figure 4.9), lacerations, abrasions, bruising, petechiae, and localized swelling. If evidence of trauma is found on the face or limbs, spontaneous movement and symmetry of movement should be evaluated to identify possible underlying damage to nerves.

Certain types of *skin lesions* may be significant in the assessment of neurologic abnormalities. Café au lait spots of 1.5 cm or larger or more than three in number may indicate the presence of neurofibromatosis, one of the more common autosomal dominant genetic disorders. In this disease, dysplastic tumors occur along nerves and sometimes in the eyes and/or meninges as well as at other sites in the body and on the skin.[4]

Port-wine nevi involving both eyelids, with bilateral distribution or those that are unilateral but involve all three branches of the trigeminal nerve, are associated with a significantly higher incidence of eye and central nervous system abnormalities.[5] These lesions may indicate the presence of Sturge–Weber syndrome with underlying arteriovenous malformations. Glaucoma and leptomeningeal vessels in the brain that can lead to seizures are other abnormalities.

Areas of skin depigmentation can be significant because they can indicate the earliest manifestation of tuberous sclerosis, a progressive degenerative neurologic disease in which collections of abnormal neurons and glia occur in the subependymal and cortical areas of the brain. The skin lesion is white, macular, and has irregular leaf-like borders. One or several lesions may be present.

If the neonate is awake and crying, attention should be paid to the *quality of the cry*, *symmetry of movement*, and *facial expression*. A loud, lusty cry is usual in the term neonate. A weaker cry may be heard in the premature, depressed, or ill neonate. A high-pitched cry may be heard in infants with neurologic disturbances, metabolic abnormalities, neonatal abstinence syndrome, or infections such as meningitis. Neonates with high-pitched, incessant crying and who are also hyperirritable should raise the concern of possible drug withdrawal, excessive nicotine exposure, or exposure to selective serotonin reuptake inhibitors.[6,7] A catlike cry may be heard with *cri du chat* syndrome, which results from deletion of the short arm of the fifth chromosome. Stridor should raise concern of partial vocal cord paralysis related to cervical nerve damage or to partial webs, stenosis, or malacia of the airway. Lack of movement of an extremity may indicate trauma and nerve damage. Lack of movement of one arm may indicate brachial plexus injury: Erb–Duchenne palsy (damage to the upper spinal roots C5 and C6), Dejerine-Klumpke palsy (damage to the lower spinal roots C8 and T1), or involvement of all the roots that make up the plexus. If respiratory distress or sustained tachypnea is present in the neonate with a brachial plexus injury, the possibility of phrenic nerve damage and resultant diaphragm paralysis should be considered. According to Volpe, approximately 5% of brachial plexus injuries have phrenic nerve injury.[8] Facial asymmetry or unilateral lack of expression is most commonly seen as an isolated finding in infants with facial weakness due to Bell palsy. The etiology may be intrauterine positioning with pressure on the fetal facial nerve from the maternal sacral prominence or secondary to direct trauma from a difficult extraction or forceps delivery. Bruising is often present.[8]

Another cause of facial asymmetry is congenital hypoplasia of the depressor anguli oris muscle. This condition is characterized by an asymmetric facial appearance and is most evident when the neonate is crying. The essential finding is a failure of one corner of the mouth to move down and out. Other functions of the facial muscles are normal. The clinical significance of this disorder is its association with other abnormalities—commonly, congenital cardiovascular anomalies and, rarely, neuroblastoma.[8]

Bilateral facial weakness is seen in neuromuscular disorders characterized by generalized hypotonia and weakness at the level of the muscle, such as congenital myotonic or muscular dystrophy. The clinical presentation includes a tent-like appearance of the upper lip, a partly open mouth, and generalized hypotonia. Congenital facial diplegia syndrome (Möbius syndrome) is a condition in which severe bilateral facial weakness is seen. The upper face is more affected than the lower, the face is expressionless, and the eyes remain partially open. Neuromuscular junction disorders, such as myasthenia gravis and infantile botulism, are other conditions associated with generalized hypotonia, including the face. Other etiologies for generalized facial weakness

include posterior fossa hematoma, cerebral contusion, and neonatal encephalopathy.[8]

The neonate's resting *posture* should be noted and its appropriateness for gestational age gauged. The normal term neonate lies with hips abducted and partially flexed and with knees flexed. The arms are usually adducted and flexed at the elbow (Figures 11.1 and 11.2). The hand is normally loosely fisted, and the thumb may lie in the palm or adjacent to the fingers. At 28 weeks gestational age, the newborn's arms and legs are extended, with little tone. From 28 to 40 weeks gestational age, tone increases in a caudocephalic direction, with increased tone in flexion observed first in the legs (around 32–34 weeks) and later in the arms (around 34–36 weeks). In the premature neonate, the adductor muscles are hypotonic; although flexion is seen as gestational age increases, the limbs are often flat against the bed surface. In this posture, the legs are abducted, and the lateral thigh rests against the surface of the bed. This position, often referred to as the *frog leg* position, is abnormal in neonates of more than 36 weeks gestation. Neonates of 32 weeks gestation or more who lie with their extremities completely extended are demonstrating abnormal postural tone. Depressed postural tone in the arms is suggested by flaccid extension of the arms or by some degree of flexion at the elbow but with open palmar surfaces facing up at 36 weeks or greater. In addition, cortical thumb—a persistent tightly fisted hand in which the thumb is firmly enclosed by the fingers—may indicate neurologic abnormality. When observation identifies any postural abnormality, it should be reconfirmed during the examination of tone and reflexes.

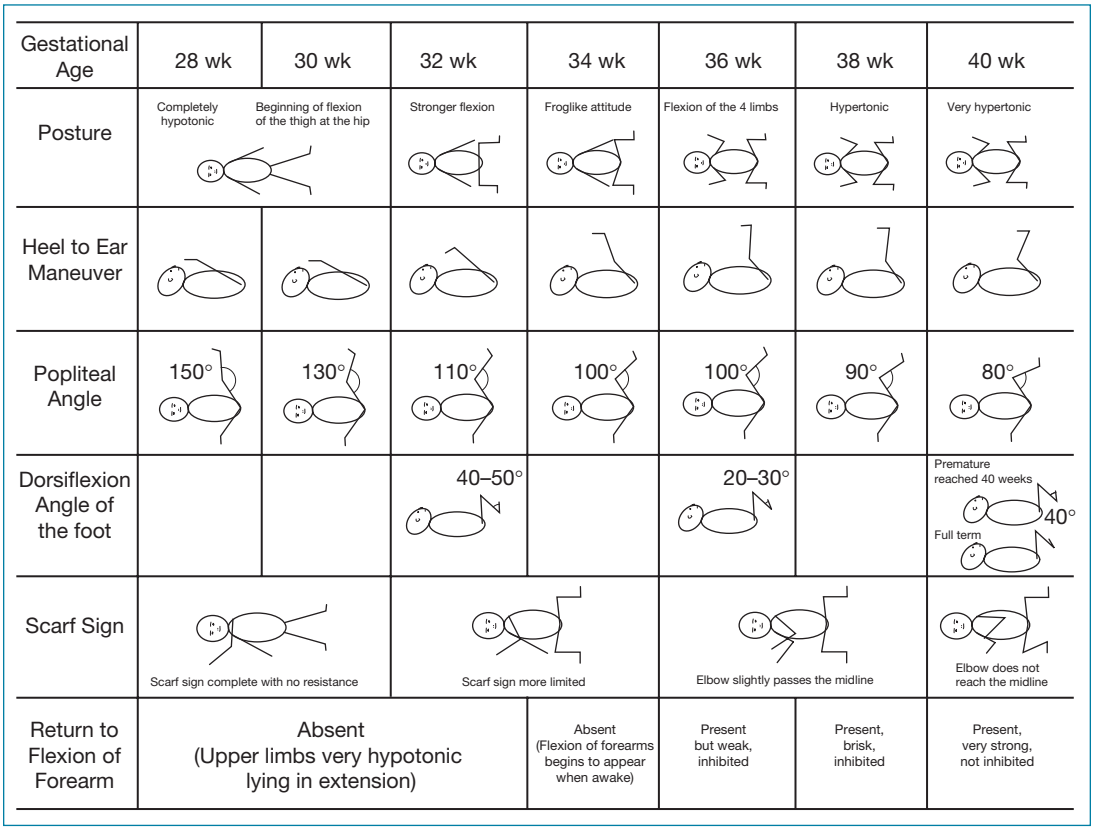

Figure 11.1 Posture and passive tone; tone increases with maturity, showing the ascending direction of tone

Source: From Amiel-Tison C. 1991. Examination of the newborn infant. In Rudolph's Pediatrics, 19th ed., Rudolph CD, et al., eds. New York: McGraw Hill, 178. Reprinted by permission.[9]

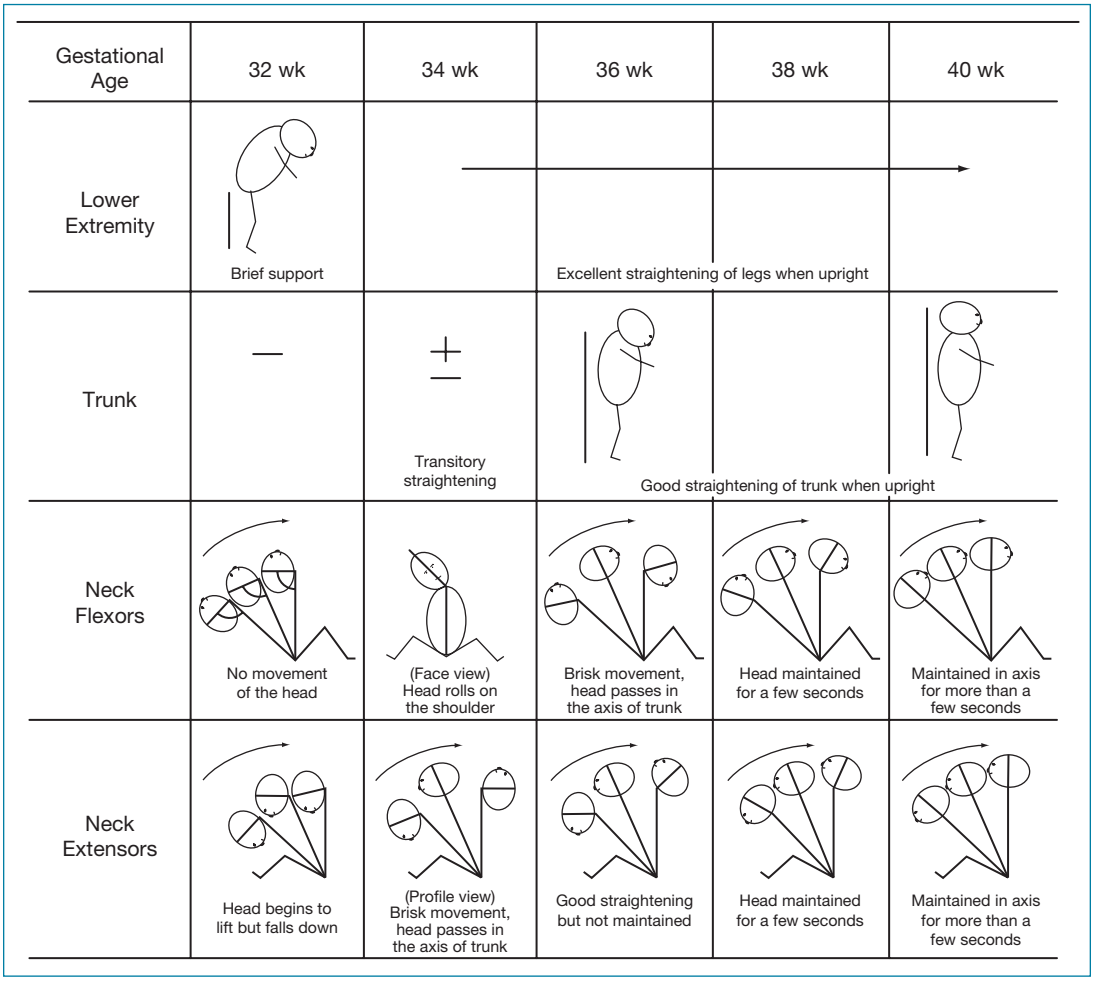

Figure 11.2 Increase in active tone with maturity is illustrated; note the ascending direction of tone

Source: Amiel-Tison C. 1991. Examination of the newborn infant. In Rudolph's Pediatrics, 19th ed., Rudolph CD, et al., eds. New York: McGraw Hill, 179. Reprinted by permission.[9]

The *quality of movements* should also be evaluated in the active neonate. Term neonates move their limbs smoothly. In the preterm neonate, tremors and jitteriness can be normally present. Jitteriness can also often be benign in term neonates and is sometimes seen with vigorous crying. It may also be a sign of disorders such as hypoglycemia, hypocalcemia, drug withdrawal, and neonatal encephalopathy. Jitteriness must be distinguished from seizure activity (Table 11.1). Although the distinction can be subtle in the neonatal period, jitteriness is characterized by rapid alternating movements of equal amplitude in both directions. In contrast, the clonic movements seen during true seizures have a fast and slow component and are not as rapid. Noise, touch, or other environmental stimuli can elicit jitteriness, which can be stopped by flexing or holding the involved extremity. Seizures are generally not initiated by stimulation, nor can they be eliminated by flexing or holding. In addition, jitteriness is not associated with any subtle signs of seizure activity, such as abnormal eye movements.

The neonate's *state* should be noted both before and during the examination, which optimally is performed with the neonate in the quiet, alert state. Timing the examination 30 minutes to 1 hour before a

TABLE 11.1: DISTINGUISHING SEIZURE ACTIVITY FROM JITTERINESS

CLINICAL FINDING	SEIZURE	JITTERINESS
Abnormal gaze or eye movements	Yes	No
Stimulus sensitive	No	Yes
Ceases with passive flexion	No	Yes
Autonomic changes	Yes	No
Predominant movement	Clonic jerking	Tremor

Source: Adapted from Volpe JJ. 2008. *Neurology of the Newborn*, 5th ed. Philadelphia, PA: Saunders, 214. Reprinted by permission.[8]

feeding may increase the chances of the neonate being in this state. Prior to 28 weeks gestation, it is difficult to identify periods of wakefulness. Stimulation may result in eye opening and apparently being alert for short periods. At approximately 28 weeks gestation, there is an increase in the level of alertness, and both stimulated and spontaneous alerting can be seen. The premature neonate has longer sleep cycles than the term neonate. Sleep–wake cycles are more apparent by 32 weeks gestation, and stimulation is usually not necessary to arouse and alert the neonate. By 37 weeks gestation. Increased alertness can be readily observed.

EXAMINATION OF THE HEAD

The status of the fontanels and sutures should be evaluated initially by gentle palpation in the noncrying neonate (Chapter 5). The examiner palpates the fontanel and sutures to determine size and to assess whether they are soft and flat or full and bulging. A full or bulging fontanel with widened sutures may indicate increased intracranial pressure and hydrocephalus. Widening of the sutures alone, with a normal anterior fontanel, may be caused by abnormal ossification seen with intrauterine growth restriction. The head is palpated for other abnormalities, such as cephalohematoma and nondisplaceable sutures.

Head circumference is measured, plotted on a growth chart, and the percentile determined based on gestational age. A neonate with a head circumference greater than the 90th percentile for gestational age and weight, and height below the 90th percentile may have hydrocephaly, macrocephaly, or hydranencephaly. The skull configuration in hydrocephalus is frequently globular. Posterior ballooning of the skull is seen with hydrocephalus caused by Dandy–Walker syndrome, which consists of congenital agenesis of the foramen of Magendie and Luschka with dilation of the fourth ventricle. Abnormalities in skull configuration are also seen in some neonates with craniosynostosis. When a large head circumference and percentile for gestational age are identified, transillumination of the skull can be helpful. In a dark room and after the examiner's eyes have adapted to the reduced light, a rubber-cuffed flashlight or other transillumination device is applied firmly to the skull (see Figure 1.2). A glow of more than 2 cm around the rubber cuff of the flashlight is abnormal and reflects fluid accumulation. Ultrasound, which is readily available in most hospitals, is the most common way to diagnose hydrocephalus in the newborn.

Neonates with small head circumferences (less than the 10th percentile for gestational age) may have microcephaly caused by a chromosomal abnormality, or maternal drug and alcohol intake. Also, intrauterine infection may be present in microcephaly or the small-for-gestational-age neonate. TORCH, which includes toxoplasmisis, other viruses (e.g., congenital syphilis and viruses), rubella, cytomegalovirus (CMV), and herpes simplex virus infections should be considered as causative. Marked molding of the head following birth may give the erroneous impression of microcephaly, especially when the shape of the head is conical.

Although not a routine part of the newborn physical examination, auscultation of the skull for bruits may be of value when arteriovenous malformation or aneurysm is suspected. Flow disturbances resulting from interference with the normal laminar

flow through vessels may cause vessel wall vibrations heard as systolic murmurs and referred to as *bruits*. Bruits may be heard in Sturge–Weber syndrome, usually accompanied by facial hemangioma; it is therefore important to listen for bruits in neonates with this skin lesion. Arteriovenous malformations may lead to unexplained high-output cardiac failure in neonates, and an examination for bruits may assist in making the diagnosis. Auscultation is carried out with the bell of the stethoscope placed over the temporal, frontal, and occipital areas.

MOTOR EXAMINATION

Evaluation of muscle tone involves examination of resting posture, passive tone, and active tone of the major muscle groups. Tone can also be categorized as phasic, which correlates with passive tone and deep-tendon reflexes, and postural, which correlates with active tone. Phasic tone is a brief, forceful contraction in response to a short-duration, high-amplitude stretch. Postural tone is a long-duration, low-amplitude stretch in response to gravity. The two tone types are tested separately and can vary independently.

Phasic Tone

Phasic tone is evaluated by testing the resistance of the upper and lower extremities to movement and by the activity of the deep-tendon reflexes. Resistance of the extremities to passive movements can be evaluated by the scarf sign (Chapter 3) and by arm and leg recoil. Minimal resistance is normal at 28 weeks; resistance increases with maturity. Tendon reflexes are elicited by sharp percussion with the examiner's finger or a small reflex hammer over the tendon. The biceps reflex and the patellar (or knee-jerk) reflex are basic reflexes that can be tested in the neonate. These are most active in the first 2 days after birth and when the neonate is alert. Of all the tendon reflexes, the patellar is the most frequently demonstrated after birth. Innervation for the knee jerk is at the second through fourth lumbar segments. It is tested by tapping the patellar tendon just below the patella while the examiner's hand supports the neonate's knee in a flexed position. The normal response is extension at the knee and visible contraction of the quadriceps. The biceps reflex is innervated at the fifth and sixth cervical nerve roots and is tested by holding the neonate's arm with the elbow in flexion and the examiner's thumb over the insertion of the biceps tendon. The examiner's thumb is tapped with a reflex hammer, and flexion of the biceps occurs.

Weak or absent reflexes may be seen in neonates less than 28 weeks gestational age, in those with neonatal encephalopathy or sepsis, and in those with dysfunction of the motor unit (e.g., the motor neuron, peripheral nerve, muscle, and neuromuscular junction). Neonates with acute encephalopathy frequently have an absence of reflexes initially. As the infant improves, reflexes return, but they may be depressed initially and later become increased or exaggerated. Exaggerated deep-tendon reflexes are also seen in neonates with neonatal abstinence syndrome, selective serotonin reuptake inhibitor exposure, or nicotine exposure. Asymmetric deep-tendon reflexes may reflect either central or peripheral nervous system impairment. *Clonus* is rapid movement of a particular joint brought about by sudden stretching of a tendon. Ankle clonus can be evaluated by holding the anterior portion of the neonate's foot with the hip and knee in flexion and dorsiflexing the forefoot. The response usually consists of several repetitive jerks (beats) of the foot or no movement at all. Sustained clonus (with more than eight to 10 beats occurring) may indicate cerebral irritation.

Postural Tone

Postural tone is best tested by the traction response (or pull-to-sit maneuver), which tests the ability to resist the pull of gravity. The traction response is tested by grasping the

neonate's hands and pulling slowly from the supine to a sitting position. The normal-term neonate reinforces this maneuver by contracting the shoulder and arm muscles, followed by flexion of the neck. As the neonate is pulled to sit, the head leaves the bed almost immediately, lagging only minimally behind the body (Figure 11.3). When the infant reaches the sitting position, the head remains erect momentarily and then falls forward. During traction, flexion occurs in the elbows, knees, and ankles. In the term neonate, more than minimal head lag is abnormal and may indicate postural hypotonia (Figure 11.4).

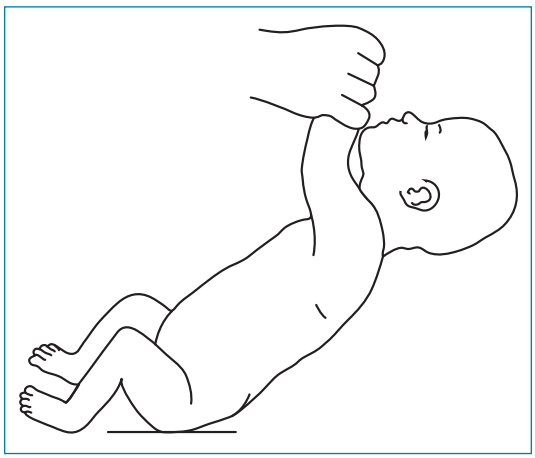

Figure 11.3 Normal traction response seen in the term neonate

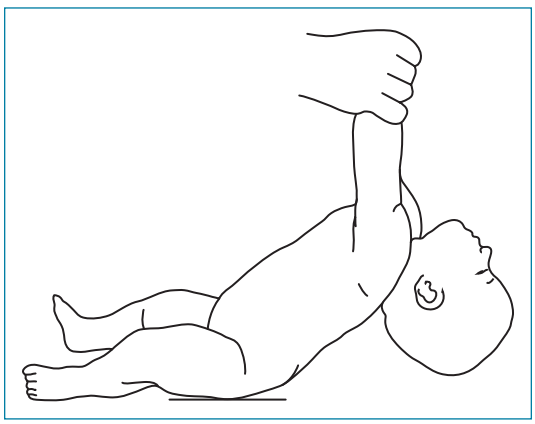

Figure 11.4 Traction response indicating hypotonia

Neck flexion in response to traction is absent in preterm neonates less than 33 weeks gestational age. Because normal postural tone requires the integrated functions of the entire central nervous system, hypotonia (indicating depressed postural tone) may result from disturbances in the central nervous system, the peripheral nervous system, or the skeletal muscles. As with the other components of the neurologic examination, postural tone should be tested with the neonate in an awake alert state. When a large subgaleal hemorrhage or an extremely large caput is present, there may be more difficulty with the performance of neck righting in the pull-to-sit maneuver. It may be difficult to determine whether poor performance represents a mechanical problem caused by extra weight of the head or a neurologic problem related to birth trauma. Normal findings on the evaluation of other reflexes, tone, and activity are reassuring. The infant should be retested prior to discharge.

Testing muscle strength is imprecise in the neonate because differentiating between hypotonia and muscle weakness is difficult. Strength of the upper extremities is gauged by using the pull-to-sit maneuver described previously and the grasp reflex. Strength of the lower extremities is evaluated by observing the infant's stepping reflex, readily elicited by 37 weeks gestational age, and gauging the neonate's ability to support its weight when the feet are against a flat surface.

Movement should be evaluated and the presence of abnormal involuntary movements, such as jerking or jitteriness, noted. At 28 to 32 weeks gestational age, slow twisting movements of the trunk as well as rapid, wide-amplitude movements of the limbs are seen. By 32 weeks gestational age, movements are more flexor and tend to occur in unison. At 36 weeks gestational age, active flexor movements of the lower extremities often occur in an alternating rather than a bilateral symmetric pattern. Term neonates' upper and lower extremities move in an alternating pattern. Typical of the neonatal period are mass movements that occur in

response to environmental stimuli and discomfort. During the first few weeks of life, coarse tremors or brief trembling of the chin may occur normally, as may occasional uncoordinated movements.

Abnormalities of Tone

The motor examination may detect hypotonia or hypertonia. *Hypotonia* is the more common abnormality observed in the neonatal neurologic examination. A focal injury to the cerebrum can result in contralateral hemiparesis involving the face and upper extremities more than the lower extremities. Injury to the parasagittal cerebral region (which may be caused by cerebral perfusion abnormalities) results in weakness of the upper limbs more than the lower limbs. If spinal cord injury occurs, it is frequently in the cervical region and can result in flaccid weakness of the extremities; the face and cranial nerves are usually not affected. Neuromuscular junction diseases, such as myasthenia gravis and infantile botulism, cause generalized weakness and hypotonia. Disorders of the lower motor neurons (e.g., Werdnig–Hoffmann disease) cause flaccid weakness of the extremities with initial sparing of the face and cranial nerves. Fasciculation (spontaneous contraction of a group of fibers in a motor unit) can also be seen in Werdnig–Hoffmann disease and is best observed in the tongue. Inspection of the tongue reveals continuous and rapid twitching movements. Damage to nerve roots results in discrete patterns of focal weakness, the location of which depends on the root involved. One example is the unilateral loss of movement seen in the arm of a neonate with brachial plexus palsy.

Hypertonia is a less common finding in the neonatal period. If present, passive manipulation of the limbs often increases the tone. Opisthotonus (marked extensor hypertonia with arching of the back) is sometimes seen with bacterial meningitis, severe neonatal encephalopathy, massive intraventricular hemorrhage (IVH), and tetanus. Table 11.2 lists patterns of abnormal muscle tone and their possible significance.

TABLE 11.2: PATTERNS OF ABNORMAL MUSCLE TONE IN THE NEONATE

ABNORMALITY	SIGNIFICANCE
Generalized increased or decreased tone	CNS insult or systemic illness
Increased arm flexor tone with increased leg extensor tone	Normal in crying neonate CNS irritability (e.g., hypoxic–ischemic injury, hemorrhage, increased intracranial pressure)
Increased neck extensor tone more than neck flexor tone	Seen in crying neonates Hypoxic–ischemic injury Meningitis Increased intracranial pressure
Tight popliteal angle, increased as compared to leg tone	Intracranial hemorrhage
Asymmetric popliteal angles beyond 40 wk	Hypertonia, hemiplegia

CNS, central nervous system.

Source: From Hill A. 1998. Development of tone and reflexes in the fetus and newborn. In *Fetal and Neonatal Physiology*, 2nd ed., Polin RA, and Fox WW, eds. Philadelphia, PA: Saunders, 2169. Reprinted by permission.[10]

ASSESSMENT OF DEVELOPMENTAL REFLEXES

Developmental reflexes are sometimes referred to as *primary* or *primitive* reflexes because they do not require functional brain above the diencephalon. Although there are many developmental reflexes, it is usual to test only those commonly present in most newborns. The normal timing of appearance and disappearance of developmental reflexes is presented in Table 11.3. Development of reflexes with maturation from 28 to 40 weeks gestation is shown in Table 11.4.

TABLE 11.3: NEONATAL REFLEXES

REFLEX	AGE (WEEKS OF GESTATION; MONTHS POSTNATAL)		
	ONSET (WEEK)	WELL ESTABLISHED (WEEKS)	DISAPPEARANCE (MONTHS)
Suck	28	32–34	12
Rooting	28	32–34	3–4
Palmar grasp	28	32	2
Tonic neck	35	4	6
Moro	28–32	37	6
Stepping	35–36	37	3–4
Truncal incurvation	28	40	3–4
Babinski	34–36	38	12–14

Source: Adapted from Volpe JJ. 2008. *Neurology of the Newborn,* 5th ed. Philadelphia, PA: Saunders, 121–147[8]; and Barness LA. 1991. *Handbook of Pediatric Physical and Clinical Diagnosis.* Philadelphia, PA: Mosby, 386.[11]

Sucking Reflex

The sucking reflex is normally present at birth, even in the premature neonate, although it is weaker with decreasing gestational age. The stimulus consists of touching or gently stroking the lips. In response, the neonate's mouth opens, and sucking movements begin. The examiner's gloved finger can be introduced into the mouth to evaluate strength and coordination of the suck.

Rooting Reflex

To evaluate the rooting reflex, stroke the cheek and corner of the neonate's mouth. The infant's head should turn toward the stimulus, and the mouth should open.

Palmar Grasp

Stimulating the palmar surface of the neonate's hand with a finger should cause a grasp of the finger. Attempts to withdraw the finger should lead to a tightened grasp. When the palmar grasp is tested with both hands, the term neonate can be lifted off the bed for a few seconds. If palmar grasp is weak or absent in a term neonate, cerebral, local nerve, or muscle injury may be present.

Tonic Neck Reflex

To elicit the tonic neck reflex, place the neonate in the supine position and turn his or her head to one side. The neonate should extend the upper extremity on the side toward which the head is turned and flex the upper extremity on the opposite side (Figure 11.5). This is sometimes called the *fencing position*. If the response cannot be obtained or if even slight turning of the head consistently produces a marked tonic neck reflex, these can be important indicators of abnormality.

Moro Reflex

The Moro reflex is a response to the sensation of loss of support. The most effective and reproducible method of stimulating this reflex is to hold the neonate in the supine position with the head several centimeters off the bed. Next, withdraw the hand supporting the head, and the infant's head will fall back into the examiner's hand or against

TABLE 11.4: DEVELOPMENT OF REFLEXES WITH MATURITY FROM 28 TO 40 WEEKS GESTATION

GESTATIONAL AGE (WEEKS)	28	30	32	34	36	38	40
Sucking	Weak and not really synchronized with deglutition		Stronger and better synchronized with deglutition		Perfect		
Grasp	Present but weak			Stronger		Excellent	
Response to traction	Absent		Begins to appear	Strong enough to lift part of the body weight		Strong enough to lift all of the body weight	
Moro reflex	Weak, obtained just once, incomplete		Complete reflex → → →				
Crossed extension	Flexion and extension in a random pattern, purposeless reaction		Good extension; no tendency to adduction		Tendency to adduction but imperfect	Complete response with • Extension • Adduction • Fanning of the toes	
Automatic walking	—	—	Begins tiptoeing with good support on the sole and a righting reaction of the legs for a few seconds	Pretty good; very fast tiptoeing		• A premature newborn who has reached 40 weeks; walks in a toe–heel progression or on tiptoes • A term newborn of 40 weeks gestation; walks in a heel–toe progression on the whole sole of the foot	

Source: Adapted from: Amiel-Tison C. 1991. Newborn neurologic examination. In Rudolph's Pediatrics, 19th ed., Rudolph AM, and Hoffman J, eds. Stamford, Connecticut: Appleton & Lange, 178. Reprinted by permission.[12]

the surface of the bed. The infant's first response is a spreading movement in which the arms are extended and abducted and the hands are opened (Figure 11.6). That response is followed by inward movement and some flexion of the arms, with closing of the fists. An audible cry may accompany this reflex in neonates older than 32 weeks of gestation. Premature neonates have incomplete responses, with abduction of the arms, but without flexion, adduction, or cry. Complete absence of the reflex is abnormal and may occur in depressed neonates. Asymmetry of movements may indicate a localized neurologic defect, such as brachial plexus injury or fractured clavicle.

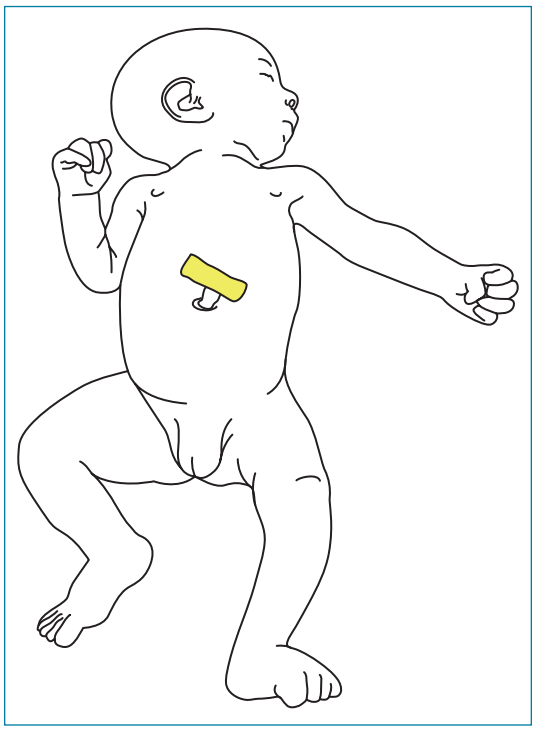

Figure 11.5 The tonic neck reflex

Figure 11.6 The Moro reflex

A loud noise or bumping the side of the incubator or crib often produces a Moro-like response. This is usually a startle reflex—consisting of flexion of the extremities and palmar grasping—but not a complete Moro.

Stepping Reflex

When the neonate is held upright and the soles of the feet are allowed to touch a flat surface, alternating stepping movements can be observed. Stepping is more active 72 hours after birth.

Truncal Incurvation (Galant) Reflex

The truncal incurvation reflex is tested with the neonate in ventral suspension, with his anterior chest wall in the palm of the examiner's hand. Firm pressure with the thumb or a cotton swab is applied parallel to the spine in the thoracic area. A positive response is flexion of the pelvis toward the side of the stimulus.

Babinski Reflex

The response to stimulation of the sole of the foot is usually plantar flexion. In the neonatal period, the Babinski reflex is positive if extension or flexion of the toes occurs. Consistent absence of any response is abnormal and may indicate central depression or abnormal spinal nerve innervation. In children older than 18 months, an abnormal response is extension or fanning of the toes; either response indicates upper motor neuron abnormalities.

ASSESSMENT OF SENSORY FUNCTION

The peripheral sensory system functions include touch and pain. The withdrawal reflex is stimulated to evaluate peripheral sensory function. Touching the sole of the foot with a pin provokes flexion of the stimulated limb and extension of the contralateral limb. Response is present in most neonates of 28 weeks gestational age and older. In neonates, extension of the contralateral limb

varies, and sometimes flexion of both limbs is seen. This assessment is rarely performed as part of a routine neurologic examination but may be performed in cases of myelomeningocele or suspected spinal cord transection to delineate the level of abnormality.

EVALUATION OF THE CRANIAL NERVES

Olfactory—Cranial Nerve I

The neonate's sense of smell is rarely tested because disturbances in olfaction are rarely a feature of neurologic disease in the neonate. The sense of smell is present in neonates, and those of nursing mothers are able to discriminate between their mother's breast pad and that of another woman.[8] Gross evaluation can be done using strong-smelling substances, such as anise, peppermint, or clove oil, and evaluating for grimacing, sniffing, or startle responses (Table 11.5).

Optic—Cranial Nerve II

Visual acuity, visual fields, and a funduscopic examination provide information on the function of the optic nerve. The ability of the neonate's eyes to fix on an object and follow it over a 60-degree arc is evaluated. The object may be the examiner's face, simple black-and-white pictures depicting facial features, or a ball held 10 to 12 inches from the neonate's eyes and moved from side to side. During the examination, occasional nystagmus (an involuntary rapid movement of the eyeball) may be seen in the neonate. Wandering or persistent nystagmus is abnormal and may indicate loss of vision. When a light is introduced into the periphery of the neonate's visual field, the head should turn toward the light. Pupillary size and constriction in response to light are also evaluated. Funduscopic examination is performed, and the character of veins and arteries, the macula, and the optic disk are evaluated. A thorough examination of the optic disk requires pupil dilation. Pupillary abnormalities and possible etiologies are shown in Table 11.6.

TABLE 11.5: TESTING CRANIAL NERVE FUNCTION OF INFANTS

CRANIAL NERVE	FUNCTION
I	Smell
II, III, IV, VI	Optical blink reflex—shine light in open eyes, note rapid closure Size, shape, equality of pupils Regards face or close object Eyes follow movement
V	Rooting reflex, sucking reflex
VII	Facial movements (e.g., wrinkling forehead and nasolabial folds) symmetric when crying or smiling
VIII	Loud noise yields Moro like reflex or startle (until 4 months) Acoustic blink reflex—infant blinks in response to a loud hand clap 30 cm (12 inches) from head (avoid making air current) Eyes follow direction of sound
IX, X	Swallowing, gag reflex
XI	Head turns normally from side to side, shoulder height is equal
XII	Coordinated sucking and swallowing Pinch nose, infant's mouth will open and tongue rise in midline

Source: Adapted from Jarvis C. 2012. *Physical Examination & Health Assessment*, 6th ed. Philadelphia, PA: Saunders, 651. Reprinted by permission.[13]

Oculomotor, Trochlear, and Abducens—Cranial Nerves III, IV, VI

These nerves supply the pupil and the extraocular muscles. Pupillary response to light is tested and should be present at a gestational age of 28 to 30 weeks. Spontaneous movements of the eyes, their size, and

TABLE 11.6: PUPILLARY ABNORMALITIES IN THE NEONATAL PERIOD

ETIOLOGY	PUPILLARY REACTIVITY
Bilateral increase in size	
Hypoxic–ischemic encephalopathy	Reactive early in course; unreactive later in course
Intraventricular hemorrhage	Unreactive
Local anesthetic intoxication	Unreactive
Bilateral decrease in size	
Hypoxic–ischemic encephalopathy	Reactive
Unilateral decrease in size	
Horner syndrome	Reactive
Unilateral increase in size	
Subdural hematoma	Unreactive
Unilateral mass	Unreactive
Third nerve palsy	± Unreactive
Hypoxic–ischemic encephalopathy	± Unreactive

Source: Modified from Volpe JJ. 2008. *Neurology of the Newborn*, 5th ed. Philadelphia, PA: Saunders, 137. Reprinted by permission.[8]

symmetry are evaluated. Presence of ptosis, proptosis, sustained nystagmus, or strabismus are noted.

Movement of the eyes as the position of the head is turned is evaluated. The "doll's eye" maneuver consists of gently rotating the head from side to side and evaluating for deviation of the eyes away from the direction of rotation—for example, in a normal response, when the head is turned to the right, the eyes deviate to the left (Figure 11.7). If the eyes remain in a fixed position regardless of head rotation, brainstem dysfunction may be present. If the eyes move in the same direction as the head is rotated, brainstem or oculomotor nerve dysfunction may be present. When it is not possible to rotate the neonate's head, the semicircular canals can be stimulated by introducing cold water into the ear canal. In this maneuver, the eyes should deviate to the side of the cold water. This response is lost when the pontine centers are compromised.

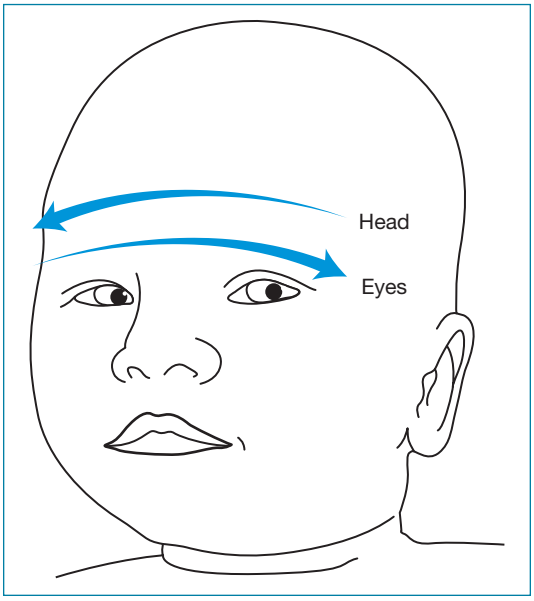

Figure 11.7 The "doll's eye" maneuver

Trigeminal—Cranial Nerve V

The trigeminal nerve supplies the jaw muscles and is responsible for the sensory innervation of the face. The three divisions of this nerve are mandibular, maxillary, and ophthalmic. If unilateral facial paralysis is present, damage to the motor component of this nerve may cause the jaw to deviate to the paralyzed side when the mouth is open. Strength of the masseter muscle is judged by placing a gloved finger in the neonate's mouth and evaluating the strength of the biting portion of the suck. The sensory component of this nerve can be estimated by response to touch (as in the rooting reflex) or by the response to gentle touching of the eyelashes with a piece of cotton. The corneal reflex can be tested by blowing air into the neonate's eye to elicit a blink or by gently touching the cornea with a small piece of cotton.

Facial—Cranial Nerve VII

The facial nerve controls facial expression. Facial symmetry is evaluated in the noncrying, undisturbed neonate, and in the crying neonate. Severe injury to the facial nerve may result in marked facial weakness. If the entire nerve on one side of the face is damaged, the neonate will be unable to wrinkle his brow or close his eyes well with crying, and his mouth will appear to draw to the normal side. Nasolabial creases will also be asymmetric. Causes of facial weakness are listed in Table 11.7.

Auditory—Cranial Nerve VIII

The auditory component of cranial nerve VIII can be grossly tested in the neonate by assessing response to a loud noise. The response may be a blink or a Moro reflex. Noting whether the infant responds to your voice by turning the head toward you during the examination can also be used to estimate hearing. If the infant fails to respond to stimuli on repeat examinations, more accurate hearing tests (e.g., auditory evoked potentials) should be performed. The vestibular component is tested by rotating the neonate from side to side and eliciting the "doll's eye" movement.

TABLE 11.7: CAUSES OF FACIAL WEAKNESS IN THE NEONATAL PERIOD

Cerebral
Hypoxic–ischemic encephalopathy
Cerebral contusion
Nerve
Traumatic nerve damage
Hematoma in posterior fossa
Neuromuscular junction
Myasthenia gravis
Infantile botulism
Nuclear injury
Möbius syndrome
Hypoxic–ischemic encephalopathy
Muscle
Myotonic dystrophy
Muscular dystrophy
Facioscapulohumeral dystrophy
Myopathy
Mitochondrial disorder
Muscle hypoplasia

Source: Modified from Volpe JJ. 2008. *Neurology of the Newborn*, 5th ed. Philadelphia, PA: Saunders, 140. Reprinted by permission.[8]

Glossopharyngeal—Cranial Nerve IX

The glossopharyngeal nerve controls tongue movement. The nerve is assessed/evaluated by inspecting tongue movement, eliciting a gag reflex, and noting the position of the uvula. If weakness of the nerve is present, the uvula deviates to one side.

Vagus—Cranial Nerve X

The motor portion of the vagus nerve supplies the soft palate, pharynx, and larynx. Bilateral lesions of this nerve impair swallowing. Nerve function is evaluated by listening to the cry for abnormalities such as hoarseness, stridor, or aphonia. The ability to swallow indicates a functioning nerve.

Accessory—Cranial Nerve XI

Control of the sternocleidomastoid and the trapezius muscles originates with the accessory nerve. The neonate's head position should be evaluated when the head is turned from one side to the other. Paralysis of the sternocleidomastoid muscle is suggested by difficulty in turning the head to the affected side. Observations of shoulder height are made to evaluate the trapezius muscle function. When the upper fibers of the trapezius are paralyzed, the corresponding shoulder will be lower than the unaffected one.

Hypoglossal—Cranial Nerve XII

The hypoglossal nerve supplies the muscles of the tongue. Assess for atrophy or

TABLE 11.8: CAUSES OF IMPAIRED SUCK AND SWALLOW IN THE NEONATAL PERIOD

Cerebral
Encephalopathy with bilateral cerebral involvement
Nuclear
Traumatic facial nerve damage
Hypoxic–ischemic encephalopathy
Möbius syndrome
Werdnig–Hoffmann disease
Arnold–Chiari malformation
Nerve
Hypoxic–ischemic encephalopathy
Bilateral laryngeal paralysis
Posterior fossa hematoma or mass
Neuromuscular junction
Myasthenia gravis
Infantile botulism
Muscle
Myotonic dystrophy
Muscular dystrophy
Myopathies
Facioscapulohumeral muscular dystrophy

Source: Modified from Volpe JJ. 2008. *Neurology of the Newborn*, 5th ed. Philadelphia, PA: Saunders, 143. Reprinted by permission.[8]

abnormal movements of the tongue, as well as the gag, suck, and swallow reflexes. A weak suck and delayed swallowing are present with damage to the nerve. Other causes of impaired suck and swallow are listed in Table 11.8.

ASSESSMENT OF THE AUTONOMIC NERVOUS SYSTEM

Function of the segmental and peripheral centers of the autonomic nervous system is well established in the term neonate. This system has priority in maturation because it controls the activity of systems and organs essential for life. Vital signs, skin, and sphincters are areas that can be assessed. Observations of trends in temperature, blood flow, heart rate, blood pressure, respiratory rate, and pupillary response to light are made. The anal sphincter is observed to see whether the opening is normal or patulous (distended). The anocutaneous reflex, or "anal wink," is tested by evaluating the response to cutaneous stimulation of the perianal skin. The normal response is contraction of the external sphincter. Bladder sphincter function is more difficult to assess. Constant dribbling of urine or bladder distention and the need to credé the bladder can indicate a neurogenic bladder. A normal variant demonstrating autonomic vasomotor instability in the neonate is the harlequin sign (Chapter 4). When the neonate is lying on his side, the dependent area becomes red, and the upper area appears pale in contrast (see Figure 4.2).

ABNORMALITIES

Neonatal Encephalopathy

Neurologic examination plays a role in the assessment of the depressed newborn. Neonatal encephalopathy results from an insult to the fetus or newborn causing a lack of oxygen and perfusion. It is associated with tissue hypoxia and acidosis. The biochemical definition of asphyxia is acidosis, hypoxia, and hypercapnia. Neonatal encephalopathy—a result of asphyxia—is a condition characterized by recognizable clinical, biochemical, and pathologic features. The spectrum of hypoxic–ischemic encephalopathy can be divided into categories. *Mild encephalopathy* is associated with irritability, jitteriness, and hyperalertness. *Moderate encephalopathy* is characterized by lethargy, hypotonia, a decrease in spontaneous movements, and seizures. *Severe encephalopathy* is characterized by coma, flaccidity, disturbed brainstem function, and seizures. Infants at risk for encephalopathy include those with a low-cord pH, Apgar score of less than 6 at 5 minutes of age, and those who need extended resuscitation continuing

for 10 minutes. Early recognition is important in the infant at risk for encephalopathy. A complete early neurologic examination, and use of bedside amplitude integrated electroencephalograms are necessary as these infants may qualify for specific brain-oriented interventions. The window for these interventions is narrow, so early and complete neurologic evaluation of the infant at risk is vital.[14]

The evolution of severe encephalopathy has been studied and is somewhat predictable.[8] From birth to 12 hours of age, the neonate with severe encephalopathy usually demonstrates stupor or coma, periodic breathing, minimal movement, and seizures. Pupillary and oculomotor responses are intact. By 12 to 24 hours, the neonate's level of consciousness appears to improve, but this apparent improvement is accompanied by apneic spells, weakness, jitteriness, and severe seizures. Between 24 and 72 hours of age, the neonate's level of consciousness deteriorates, and stupor and coma may be associated with respiratory arrest. Pupils become fixed and dilated, and doll's eye response is lost. Those neonates who survive 72 hours or longer usually demonstrate improvement in level of consciousness over days to weeks, but hypotonia and weakness are common. Disturbances in suck, swallow, gag, and tongue movements may impair the ability to feed.

Specific patterns of limb weakness reflect the area of brain injury resulting from asphyxia. In term neonates, injury occurs predominantly to the parasagittal areas, zones between the cerebral arteries that are most affected by changes in blood flow and oxygenation. Weakness of the shoulder girdle and proximal upper extremities results from injury to this area of the brain. Focal ischemic injury in the area of the middle cerebral artery also presents frequently in the term neonate and is demonstrated by hemiparesis. In the premature neonate, decreased arterial blood flow affects the periventricular (PV) area and results in PV leukomalacia. Damage to the PV area results in weakness in the lower extremities.[8]

Maternal Drug Use

Drug use during pregnancy may affect the fetus and the newborn by producing malformations and withdrawal. A group of drugs known as *selective serotonin reuptake inhibitors* is now being implicated in a number of neurobehavioral abnormalities in the term newborn. The signs seen most often include tremors, shaking, agitation, spasms, hyper- or hypotonia, irritability, and sleep disturbances. Medical interventions include supportive care and ventilatory support.[6] Nicotine can also cause neurobehavioral disturbances in the term newborn. Infants exposed to tobacco have increased signs of stress, hypertonicity, and excitability.[7] Other specific drugs and their relationship to central nervous system malformations and abnormal neurologic signs are listed in Tables 11.9 and 11.10, respectively (see Chapter 15).

TABLE 11.9: MATERNAL DRUG USE AND FETAL CENTRAL NERVOUS SYSTEM ABNORMALITY

DRUG	ABNORMALITY
Isotretinoin (Accutane)	Microcephaly, absence of the cerebellar vermis, hydrocephalus, Arnold–Chiari malformation, Dandy–Walker syndrome
Antiepileptic drugs	Microencephaly, anencephaly, myelomeningocele, hydrocephalus
Primidone (Mysoline)	Microcephaly, hydrocephalus, spina bifida, anencephaly
Cocaine	Microcephaly, cerebral infarction, encephalocele
Narcotics	Microcephaly, strabismus
Warfarin (Coumadin)	Microcephaly, hydrocephalus, brain atrophy, Dandy–Walker syndrome
Ethanol	Microcephaly

Source: Adapted from Dodson WE. 1989. Deleterious effects of drugs on the developing nervous system. *Clinics in Perinatology* 16(2): 340–343, 348–353.[15]

TABLE 11.10: MATERNAL DRUG USE AND NEONATAL NEUROLOGIC ABNORMALITY

DRUG	ABNORMALITY
Isotretinoin (Accutane)	Hypotonicity, decreased reflexes, feeding problems, facial nerve paralysis, lack of visual responsiveness, seizures
Narcotics	Withdrawal symptoms; increased activity, tone, and arousal to stimulation
Phencyclidine	Hypertonicity, decreased reflexes, bursts of agitation, rapid changes in level of consciousness
Cocaine	Depressed interactive behavior, poor organizational responses to environmental stimuli

Source: Adapted from Dodson WE. 1989. Deleterious effects of drugs on the developing nervous system. *Clinics in Perinatology* 16(2): 340–343, 350–352.[15]

Neuromuscular Disorders

Components of the central and peripheral nervous systems are responsible for the control of movement and tone. Originating in the cerebral cortex and terminating in the muscle itself, these components compose what is known as the *motor system.* Generally, evidence of neonatal encephalopathy as the cause of abnormalities of the motor system is absent. The main clinical findings in affected neonates are hypotonia and weakness, often with an alert appearance. Examination is directed toward evaluation of the muscle size, tone, tendon reflexes, muscle fasciculation, and muscle fatiguing. Arthrogryposis, characterized by fixed position and limitation of limb movement, may be a major presenting feature of neuromuscular disease in the neonate. Atrophic muscles and decreased tendon reflexes are usually present. Limitation of fetal movement from any condition can lead to arthrogryposis, for example, conditions affecting the brain, anterior horn cells, nerves, neuromuscular junctions, or muscles. Neurogenic causes are the most common and include brain malformations, chromosomal defects, genetic syndromes, and destructive lesions of the central nervous system.[16]

Spinal Cord Injuries

Injuries to the spinal cord are usually seen following deliveries that overstretch the vertebral axis or over-rotate the body in relation to the head. These injuries may occur with cephalic or breech presentations. Clinical manifestations depend upon the level and severity of the injury. Transection injuries are irreversible. Partial or complete recovery is possible with injuries caused by compression or ischemia. Following spinal cord injury, a state of shock and difficulty initiating respiration are common. Lesions above C3 or C4 paralyze the diaphragm. Lower cervical and upper thoracic cord lesions result in flaccidity of the legs and portions of the arms. Lack of perceptible response to pinprick can be demonstrated below the level of spinal cord injury.[17,18]

Defects in Closure of the Anterior Neural Tube

Anencephaly

Anencephaly is the result of defective closure of the anterior neural tube. The defect in the skull begins at the vertex and may extend to the foramen magnum. Dermal covering is absent; hemorrhagic and fibrotic cerebral tissue lies exposed to view. The cranium is underdeveloped, resulting in shallow orbits and protruding eyes. The most common variety of anencephaly involves the forebrain and variable amounts of the upper brainstem. Associated abnormalities (e.g., adrenal hypoplasia and lung defects) are frequently seen.

Encephalocele

Encephalocele is a restricted disorder of neural development involving closure of the anterior neural tube. There is a protrusion of meninges and sometimes cerebral tissue, which is covered by skin. The defect in the skull that allows protrusion is referred to as *cranium bifidum*.

Occasionally, cranium bifidum may be present without protrusion of meninges or fluid and instead appears as a small, tissue-covered opening on the skull. Noticeable tufts of normal hair may surround the defect. Encephaloceles most often occur in the midline of the occipital bone, but they may also occur in the parietal, temporal, or a frontal nasopharyngeal area. They sometimes appear to be protruding from the eye socket.

Severity of clinical findings is related to the location, size, and contents of the sac. Encephaloceles may vary from tiny protrusions to massive protrusions the size of the skull. The sac may contain cerebrospinal fluid, meninges, and cerebral tissue. Severe deficits occur if brain tissue and part of the ventricular system are trapped in the defect. Microcephaly, spasticity, seizures, and cortical blindness are associated with structural defects of the occipital lobe. Skull x-rays are valuable in identifying the defect in the skull (the bifid cranium). Transillumination and ultrasound can be useful in identifying the extent of brain tissue in the sac.

Defects in Closure of the Posterior Neural Tube

Spina bifida, meningocele, and myelomeningocele are defects arising from abnormal closure of the posterior neural tube (Figure 11.8).

Spina Bifida Occulta

A malformation caused by non- or incomplete closure of the posterior portion of the vertebrae, spina bifida is the mildest form of all neural tube defects. It occurs most often in the lower lumbar and lumbosacral area and is covered with skin. The meninges and spinal cord are normal. The presence of tufts of hair, lipomas, or other abnormalities along the spine may indicate the presence of serious underlying defects. A dimple on the spine should be differentiated from a sinus. A dimple is a common finding and rarely indicates an underlying problem. It is usually located just superior to the anal opening. A dermal sinus can occur anywhere along the midline of the back, but is most frequently seen in the lumbar region. Although dimples generally have no clinical significance, sinuses may terminate in subcutaneous tissue, a cyst, or a fibrous band, or may be associated with spina bifida and extend into an open spinal cord.

To inspect the dimple or sinus, try to visualize the skin covering the end of the site. This may be difficult with deep sinuses. Probing the site with instruments is contraindicated; in the case of an open spine, direct trauma to spinal elements as well as possible introduction of bacteria into the defect could occur. With skin lesions, including deep sinuses or dimples, evaluation is indicated. Ultrasound of the involved area is helpful in identifying the vertebral abnormalities and the relationship of the defect to the meninges and cord.

Meningocele

Lesions associated with spina bifida, such as meningoceles, usually involve more than one vertebra. The meninges, covered by thin atrophic skin, protrude through the bony defect. The spinal roots and nerves are normal, and neurologic deficits are unusual.

Myelomeningocele

Myelomeningoceles are lesions associated-with spina bifida in which there is often bilateral broadening of the vertebrae or absence of the vertebral arches. In this type of lesion, the meninges, spinal roots, and nerves protrude. Remnants of the spinal cord are fused and the neural tube is exposed on the dorsal portion of the mass. Most of these lesions occur in the lumbar spine. Abnormalities in neurologic function depend on the level of the lesion (Table 11.11). Generally, the higher the defect is on the spine, the greater the degree of paralysis. Thoracic-level defects may be associated with marked abnormalities in spinal curvature and defects of the hips and lower extremities. Attention should be paid to examination of the motor, sensory, and sphincter functions, and to the reflexes.

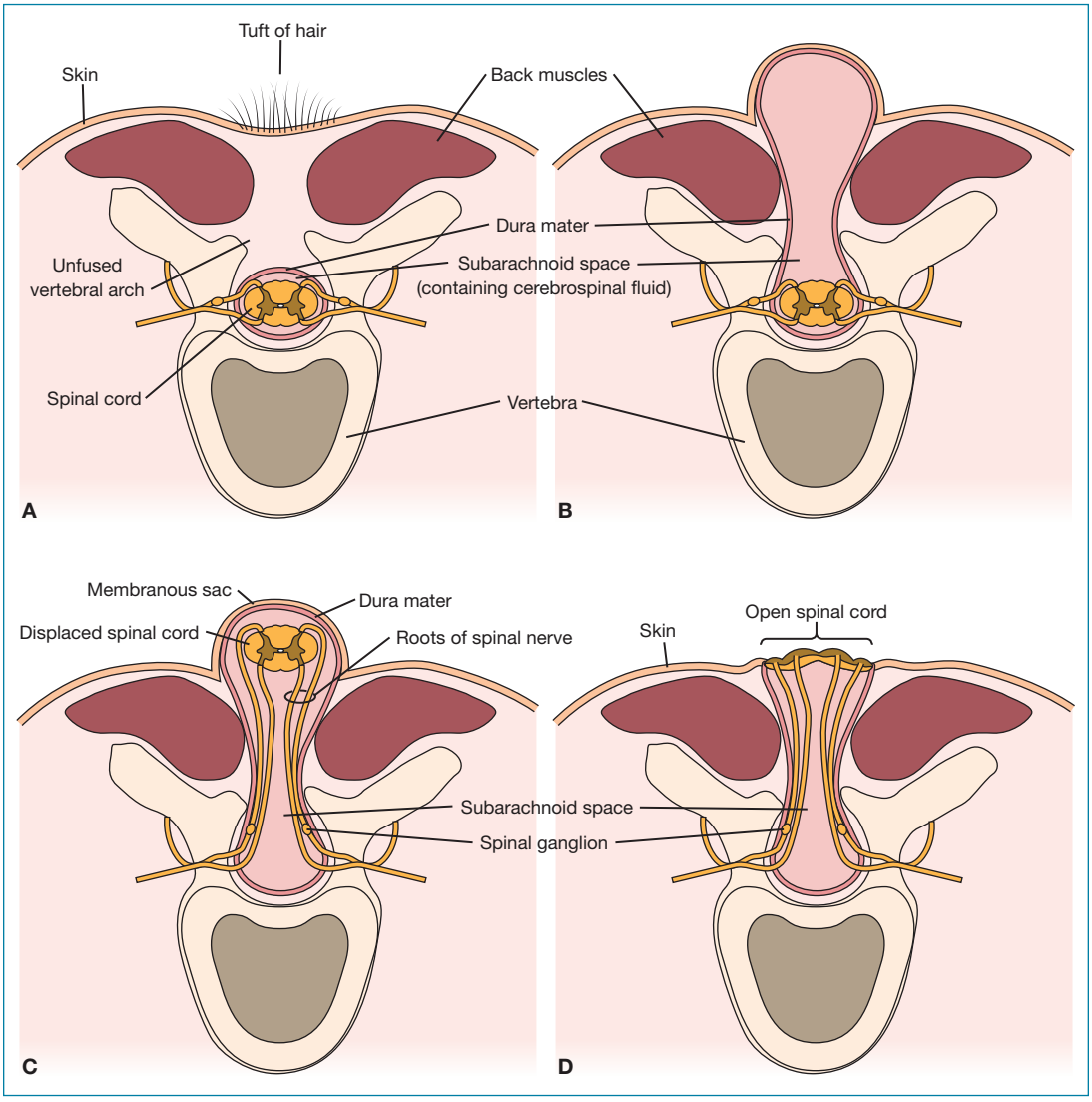

Figure 11.8 Types of spina bifida and the commonly associated malformations of the nervous system commonly associated with them. The Diagrammatic sketches illustrating various types of spina bifida and the associated defects of the vertebral arches (one or more), spinal cord, and meninges. (**A**). Spina bifida occulta. Observe the unfused vertebral arch. (**B**) Spina bifida with meningocele. (**C**). Spina bifida with meningomyelocele. (**D**). Spina bifida with myeloschisis. The defects illustrated in B to D are referred to collectively as spina bifida cystica because of the cyst-like sac or cyst associated with them.

Source: From Moore KL, Persaud TVN, and Torchia MG. 2013. *The Developing Human: Clinically Oriented Embryology*, 9th ed. Philadelphia, PA: Saunders, 399. Reprinted by permission.[19]

Hydrocephalus is a frequent finding associated with myelomeningocele. The status of head size and its percentile for gestational age, fontanel pressure, and width of sutures should be determined initially and periodically. Transillumination of the head following admission can be useful; serial brain ultrasounds are indicated.

Hydrocephalus seen with myelomeningocele is frequently associated with the Arnold–Chiari malformation (inferior displacement of the medulla and fourth ventricle

TABLE 11.11: MYELOMENINGOCELE AND SENSORY, MOTOR, SPHINCTER, AND REFLEX FUNCTION

LESION	INNERVATION	CUTANEOUS SENSATION	MOTOR FUNCTION	WORKING MUSCLES	SPHINCTER	REFLEX
Cervical/ thoracic	Variable	Variable	None	None	None	—
Thoracolumbar	T12	Lower abdomen	None	None	None	—
	L1	Groin	Weak hip flexion	Iliopsoas	None	—
	L2	Anterior upper thigh	Strong hip flexion	Iliopsoas and sartorius	None	—
Lumbar	L3	Anterior distal knee and thigh	Knee extension	Quadriceps	None	Patellar
	L4	Medial leg	Knee flexion, hip adduction	Medial hamstrings	None	Patellar
Lumbosacral	L5	Lateral leg, medial knee and foot	Foot dorsiflexion and eversion	Anterior tibial	None	Ankle jerk
	S1	Sole of foot	Foot plantar flexion	Gastrocnemius, soleus, posterior tibial	None	Ankle jerk
Sacral	S2	Posterior leg and thigh	Toe flexion	Flexor hallucis	Bladder and rectum	Anal wink
	S3	Middle of buttock	Toe flexion	Flexor hallucis	Bladder and rectum	Anal wink
	S4	Medial buttock	Toe flexion	Flexor hallucis	Bladder and rectum	Anal wink

Note: To assess the degree of dysfunction and level of lesion, evaluate the neonate with myelomeningocele for the presence of cutaneous sensation, motor function, working muscles, sphincter control, and reflexes.

Source: Adapted from Volpe JJ. 2008. *Neurology of the Newborn,* 5th ed. Philadelphia, PA: Saunders, 10[8]; and Noetzel M. 1989. Myelomeningocele: Current concepts of management. *Clinics in Perinatology* 16(2): 318. Reprinted by permission.[20]

into the upper cervical canal and elongation and thinning of the upper medulla and lower pons). Inferior displacement of the lower cerebellum through the foramen magnum into the lower cervical canal is also a feature. Hydrocephalus is thought to result from aqueductal stenosis and blockage of the cerebrospinal fluid outflow from the fourth ventricle. Neurologic, neurosurgical, urologic, and orthopedic consultations are commonly required for these patients.

INTRACRANIAL HEMORRHAGE

Primary Subarachnoid Hemorrhage

A common type of neonatal intracranial hemorrhage, primary subarachnoid hemorrhage may occur in both term and preterm neonates, but is more common in the former. Bleeding occurs from vessels within the subarachnoid space, and the blood (hemorrhage) is usually most prominently located over the surface of the cerebral hemispheres

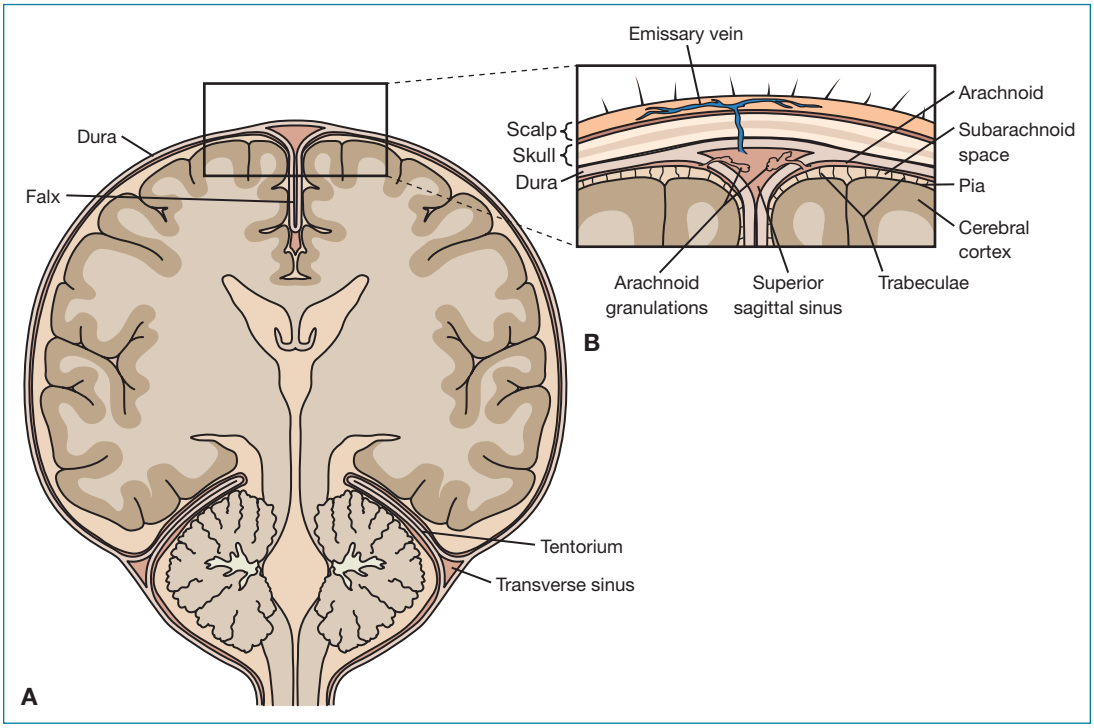

Figure 11.9 The brain and its coverings

Source: From Waxman SG. 2000. *Correlative Neuroanatomy*, 24th ed. New York, NY: Lange Medical Books/McGraw-Hill, 155. Reprinted by permission.[21]

(Figure 11.9). Etiology is thought to be trauma or hypoxia. Complications are rare.

Minor hemorrhages may go undetected because the neonate will generally be asymptomatic. Moderate degrees of hemorrhage may result in seizure activity in a neonate who otherwise appears well. One or more seizures can occur, but the neonate is stable between them.

Rarely, a massive subarachnoid hemorrhage may occur followed by rapid clinical deterioration and death. These neonates usually have histories of severe perinatal asphyxia and some degree of trauma. In those neonates surviving a major hemorrhage in the subarachnoid area, the development of hydrocephalus is possible. Hydrocephalus occurs with major subarachnoid hemorrhages because of decreased spinal fluid absorption by the inflamed arachnoid villi, resulting in adhesions in the subarachnoid space or around the outflow of the fourth ventricle.

Subdural Hemorrhage

The least common type of intracranial hemorrhage, subdural hemorrhage is most often caused by trauma. It is seen more frequently in term rather than preterm neonates. These hemorrhages are caused by (a) rupture of the tentorium, (b) occipital diastasis, (c) falx lacerations, and (d) rupture of superficial cerebral veins.[8]

The dura mater is a fibrous tissue with two layers: an outer periosteal layer and an inner meningeal layer. These layers separate in areas to form sinuses into which major veins drain. One of the layers of separation is called the falx cerebri. The falx divides the cerebral hemispheres. The superior and inferior sagittal sinus and the vein of Galen lie close to the falx. Another area of the dura called the *tentorium* separates the cerebral hemispheres from the cerebellum. The straight sinus, vein of Galen, and transverse sinus lie in close proximity to the tentorium.

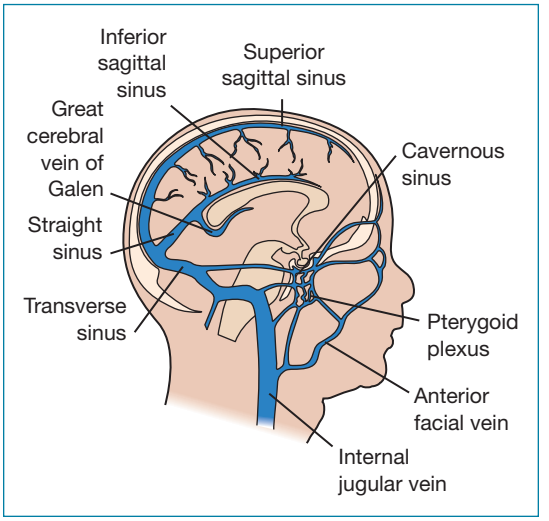

Figure 11.10 Major cranial veins and dural sinuses

Source: From Volpe JJ. 2008. *Neurology of the Newborn*, 5th ed. Philadelphia, PA: Saunders, 486. Reprinted by permission.[8]

Tentorial laceration may result in rupture of the vein of Galen, straight sinus, or transverse sinus (Figure 11.10). A tentorial tear/laceration is usually caused by trauma associated with a difficult delivery and the need for forceps instrumentation/extraction. Clots extend into the posterior fossa and, if large, cause compression of the brain. When the neonate has a large hemorrhage in this area, neurologic disturbances will be present from birth, with signs of midbrain–upper pons compression such as stupor, coma, lateral deviation of the eyes that does not change with the doll's eye maneuver, and unequal pupils with abnormal response to light. *Opisthotonos* may also be present. When bradycardia is seen, it generally indicates severe compression from a massive hemorrhage. As the size of the clot increases, stupor becomes coma, pupils become fixed and dilated, and the signs of lower brainstem compression, such as abnormal eye movements and apnea, occur.

Occipital diastasis is a traumatic injury that results in separation of the cartilaginous joint between the squamous and lateral portions of the occipital bone. This can lead to tearing of the dura and the occipital sinuses with massive bleeding below the tentorium. The same clinical features as described for tentorial laceration occur. Smaller degrees of hemorrhage and hematoma formation in the posterior fossa lead to a different clinical evolution of signs. Initially, the neonate may have no abnormal neurologic signs. These signs begin to develop over hours or days as continued seepage of blood and gradual enlargement of the hematoma occur. Clinical signs of increased intracranial pressure ensue, followed by signs of disturbance of the brainstem, including respiratory depression, apnea, oculomotor abnormalities, and facial paresis. Seizures are also commonly seen.

When *falx laceration* has occurred with bleeding from the superior sagittal sinus, hematoma development in the cerebral fissure occurs. Marked neurologic signs develop only when the clot has extended infratentorially, and then the clinical signs are those described for tentorial laceration.

Rupture of the superficial cerebral veins results in blood collecting over the cerebral convexities. Three neurologic presentations have been associated with hemorrhage in this area. The first and most common that occurs with minor hemorrhages is either hyperirritability and a hyperalert appearance or no clinical signs at all. The second presentation includes signs of focal cerebral disturbances. Seizures are common; hemiparesis with eye deviation to the side opposite the hemiparesis can occur. The doll's eye maneuver remains normal because this is a cerebral lesion, not a brainstem lesion. Dysfunction of the third cranial nerve, the oculomotor, on the side of the hematoma results in a poorly reactive or nonreactive pupil on the side of the lesion. The third presentation is secondary to chronic subdural effusion. The neonate has few or no clinical signs in the neonatal period, but presents months later with an enlarging head.

Periventricular–Intraventricular Hemorrhage

PV–IVH is the most common cause of intracranial hemorrhage seen in the premature

neonate.[8] The incidence of IVH increases with decreasing gestational age. Correlation with gestational age is related to the site of bleeding, the subependymal germinal matrix. In this area of the brain, there is a matrix of poorly supported, thin-walled capillaries located near the caudate nucleus at or slightly posterior to the foramen of Monro. At a gestational age of 28 weeks, the vessels in this area begin to involute, and involution continues so that the subependymal germinal matrix is no longer present by term gestation.

Hemorrhage that starts in the PV germinal matrix can be localized in this area, or it may rupture into the ventricular system and, if large enough, cause distention of the lateral ventricles. Adjacent cerebral tissue can also be damaged as a result of hemorrhage in the germinal matrix, resulting in a parenchymal clot or infarction.

Large IVHs have a high incidence of associated hydrocephalus. Most often, hydrocephalus occurs as a result of inflammation of the arachnoid villi, which absorb cerebrospinal fluid. With hemorrhage, they become inflamed or scarred from blood and particulate matter in the cerebrospinal fluid. Obstruction to absorption of cerebrospinal fluid then occurs. It is less common for obstruction to occur at the outlet of the third ventricle, the aqueduct of Sylvius, when debris and tissue reaction combine to lead to a blockage there.

As stated previously, the germinal matrix is a poorly supported structure with thin-walled vessels that is adjacent to the lateral ventricles. Autoregulation of cerebral blood flow does not occur in sick, premature neonates.[8] This lack of autoregulation renders the vessels of the subependymal germinal matrix vulnerable to blood flow alterations. Hemorrhage can result after a period of increased blood flow, decreased blood flow, increased central venous pressure, or with coagulation abnormalities. Most PV–IVHs occur in the first day of life, and 90% are identified using ultrasound by 72 hours of age.

Three clinical presentations have been described and are thought to be related to severity of hemorrhage. A catastrophic course is the least common but most dramatic presentation. Stupor progresses to coma, shallow respirations progress to apnea, generalized seizures occur, pupils become fixed and nonreactive to light, the doll's eye maneuver is abnormal, the eyes are fixed to vestibular stimulation, and marked hypotonia is present. A falling hematocrit, hypotension, bradycardia, metabolic acidosis, and a bulging anterior fontanel are also accompanying findings.[8]

A less dramatic course has been described in which the neonate presents with signs of a progressively decreasing level of consciousness, hypotonia, lethargy, subtle eye movement abnormalities, and partial response to the doll's eye maneuver.[8] Deterioration occurs over many hours, with periods of apparent stabilization followed by recurrence of abnormal signs. This progression of neurologic symptoms with periods of apparent stabilization may be seen over several days.

The third type of presentation is a clinically silent course in which a screening ultrasound detects the hemorrhage. Rarely, IVH is seen in the term neonate. In these neonates, it is most common for the site of hemorrhage to be the choroid plexus within the ventricle. Trauma, infection, and hypoxic events are thought to be the pathogenic mechanisms.[8]

EXTRACRANIAL HEMORRHAGE

Two types of extracranial hemorrhage can potentially complicate the neonate's course: cephalohematoma and subgaleal hemorrhage. These hemorrhages can lead to anemia and jaundice. Cephalohematoma is associated with the use of forceps during delivery. Subgaleal hemorrhage is associated with vacuum extraction. Cephalopelvic disproportion, prolonged labor, and tetanic contractions are also contributory factors.[8]

A *cephalohematoma* is located below the periosteum and confined by the cranial sutures. The periosteum of the cranial bone limits the potential space available for blood

to expand. Cephalohematoma may contribute to jaundice, but is rarely of clinical significance from a neurologic standpoint (see Figures 5.7 and 5.8).

A *subgaleal hemorrhage* is located between the scalp (galea) and the cranial periosteum. The potential space for blood to accumulate extends from the orbital ridges to the nape.[22] A major portion of the neonate's blood volume can potentially be contained in this space, leading to severe hypovolemic shock, anemia, and death (see Figure 5.7 and 5.9). Clinical signs for early identification are an expanding boggy scalp, pallor, prolonged capillary refill, tachycardia, decreased responsiveness and spontaneous activity, a falling hematocrit, and signs of frank or impending shock. Seizure activity may be a late sign. Incidence of severe subgaleal hemorrhage has decreased due to limitations on the number of vacuum or forceps attempts at operative vaginal deliveries by the American College of Obstetricians and Gynecologists.[23]

MENINGITIS

Neonatal meningitis is associated with a variety of Gram-negative and Gram-positive bacterial organisms, as well as with viruses and fungi (Table 11.12). Neonates with bacterial meningitis often exhibit clinical signs indistinguishable from those of neonatal sepsis, including temperature instability, irritability, poor feeding, and vomiting. Neurologic signs of neonatal meningitis may include irritability, high-pitched cry, lethargy, poor tone, tremors, and seizures. The fontanel may be full, and nuchal rigidity may be present.[25] Congenital viral infections often produce growth restriction, microcephaly, skin lesions, and hepatosplenomegaly.

CMV is among the most common congenital infections that can present asymptomatically or may present with microcephaly, petechiae, purpura, and jaundice on physical examination.[26] Emerging as a perinatal concern are congenital infections caused by Zika virus. The neonate with Zika virus infection may present with seizures, severe microcephaly, contractures, significant early hypotonia, craniosynostosis, and may be small for gestational age. A careful history of travel from the family should be obtained if there is concern for Zika virus infection.[27]

TABLE 11.12: INFECTIOUS AGENTS ASSOCIATED WITH NEONATAL MENINGITIS

Bacteria	**Fungi**
Bacteroides fragilis	Candida
Citrobacter	**Viruses**
Escherichia coli	Cytomegalovirus
Haemophilus	Herpes simplex 1 and 2
Klebsiella	Rubella
Listeria monocytogenes	Varicella zoster
Neisseria meningitides	Enteroviruses
Pseudomonas aeruginosa	**Protozoa**
Serratia	*Toxoplasma gondii*
Staphylococcus aureus	**Other**
Staphylococcus epidermidis	*Treponema pallidum*
Streptococcus Group B	
Streptococcus pneumoniae	

Source: Adapted from Bale JF, and Murphy JR. 1997. Infections of the central nervous system in the newborn. *Clinics in Perinatology* 24(4): 800. Reprinted by permission.[24]

SEIZURES

Seizures are the most frequent sign of neonatal neurologic disorders.[8] They can be characterized by their appearance and the abnormal activity displayed (Table 11.13). Etiology of seizure activity may be central nervous system infection, metabolic derangements such as hypoglycemia or hypocalcemia, hypoxic/anoxic or ischemic–hemorrhagic events, developmental defects, or neonatal abstinence. In some cases, seizures have a familial basis. When seizure activity is identified, rapid evaluation of the cause and prompt treatment should follow.

TABLE 11.13: NEONATAL SEIZURES

SEIZURE TYPE	CHARACTERISTICS
Subtle	Seen in term and preterm infants; apneic spells; tonic horizontal eye deviation, jerking, sustained eye opening, eyelid blinking or fluttering; sucking, drooling, oral buccal movement; swimming, pedaling, rowing movements
Generalized tonic	Primarily seen in preterm infants; characterized by tonic extension of all limbs or by tonic flexion of arms and extension of legs
Multifocal clonic	Seen primarily in term infants; clonic movement migrating from limb to limb in a sporadic pattern (e.g., right leg, then left arm)
Focal clonic	Seen more commonly in term than in preterm infants; well-localized clonic jerking in a conscious state
Myoclonic	Seen in both term and premature infants; synchronous jerks of flexion, single or multiple, affecting upper more than lower limbs

Source: Adapted from Volpe JJ. 2008. *Neurology of the Newborn*, 5th ed. Philadelphia, PA: Saunders, 211–214.[8]

SUMMARY

A careful early neurologic assessment of the newborn is mandatory for optimal management. A normal examination is reassuring to the parents and the examiner. An abnormal examination serves as a guide for attempting to clarify the etiology of abnormal responses, documenting changes over time, and providing optimal care and intervention for the neonate.

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Behavioral Assessment

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The newborn infant is truly amazing and awareness of the infant's capabilities and interactive behaviors have evolved over the past 35 years. Born at term or preterm, the infant has a unique repertoire of behaviors that caregivers can observe to better understand what the infant is trying to accomplish. The infant's behaviors are a window for understanding the infant's developing brain. Assessing the infant's neurologic status and neurobehavioral patterns requires evaluating how the infant responds and interacts with others in context of the environment.

There is growing evidence the premature infant and the developing brain are influenced especially in the vulnerable window of time the infant is cared for in the neonatal intensive care unit (NICU).[1] Although survival rates of extremely premature infants are steadily improving, the incidence of later developmental disabilities for these infants remains high.[2–5] It has been optimistically yet incorrectly proposed that healthy preterm infants without major complications will eventually catch up developmentally to full-term infants. Research suggests as preterm infants mature, they remain increasingly disadvantaged on many neurodevelopmental outcomes.[6–9]

The womb is a dynamic, sensory rich environment. The neonate who is delivered early *experiences a very different sensory environment* in the task-driven, high-tech world of the NICU. To best respond to an infant as caregivers or parents, we must first understand the unique behavioral strategies the infant uses to cope with the intensive care nursery experience. At this point in development, the preterm infant "expects" a sensory experience that is quite different from the actual experiences in the NICU. This mismatch in neurobiological sensory expectations in the context of the developing brain is important to long-term developmental outcomes.[10,11]

Behavioral assessment provides information about the infant's neurobehavioral functioning and allows caregivers to design individualized, developmental care plans for hospitalized infants. Studies by Als and associates and Buehler and colleagues show that appropriate, individualized developmental care of preterm infants decreases the length of hospitalization.[10–12] A meta-analysis of individualized developmental assessment and care demonstrated a statistically significant impact on the requirement for supplemental oxygen and on neurodevelopmental outcome at 9 and 12 months, but not at 2 years.[13,14] Another study indicated an enhancement of developmental outcomes when the infant's behavioral and physiologic cues were supported in the nursery.[15–17] McAnulty and colleagues reported individualized developmental care had enhanced neuropsychological and electrophysiological effects into school age, which were evaluated at 8 years corrected age.[17] Although the latest Cochrane Review related to the integration of developmental care states there is not yet enough evidence to support change of practice, it also states there is no evidence to support that these interventions have negative effects.[18] In fact, there appears to be growing evidence for many of the interventions included in what is called *developmentally supportive care*. What are more difficult to sort out

are the cumulative effects of this type of care on infant and family outcomes. A systematic review examining the Newborn Individualized Developmental Care and Assessment Program (NIDCAP) was published in *Pediatrics* in 2013 and found no differences in outcomes; however, many inaccuracies have been noted in how the review of the science was conducted.[14,19]

Parents and professionals have long searched for accurate indicators of competence, developmental delay, or disability in infants. Neurodevelopment is dynamic, influenced by relationships, the environment, and genetic predispositions. Despite advances in care for preterm infants, it remains difficult to predict adverse neurodevelopmental outcomes accurately.[20] Untimely and/or inappropriate predictions sometimes have caused both infants and families more harm than benefit. Each approach to assessing the newborn represents the need to determine early which infants are progressing normally and which will benefit from therapuetic interventions and follow up. The infant's developmental trajectory is influenced by evolving relationships. Parents are the infant's ideal nurturer and coregulator, enhancing the infant's competence and ability to build trust within the relationship.[21] Parents and infants benefit from the parent's awareness and responsiveness to the infant's behavioral capabilities and temperament. Parental ability to interpret the infant's behavioral cues has been shown to strengthen parent–infant interaction during the first year of life.[15,22–24] When parents have the skills to interpret their infant's cues and respond appropriately, the infant is better able to self-regulate and respond appropriately to the physical and social environment.[10] Providing infants with this foundation for trusting interactions increases their self-regulatory efforts and ability to respond to new situations.

The assessment of neurobehavioral functioning complements and elaborates on the neurologic assessment as described in Chapter 11. To differentiate, the neurologic examination assesses the function of the central nervous system and includes assessment of muscles and reflexes within the context of the infant's state of consciousness, whereas the behavioral examination relies on describing the infant's observable behavior. This behavior pattern is thought to be a reflection of the infant's underlying neurologic status and current level of functioning. A critical window of incredible brain growth and differentiation happens between 24 and 40 weeks postmenstrual age. How the caregiver interacts with the infant during this critical window influences brain development and function.[10]

Newborn behavioral assessments must be understood in the context that all human experiences have psychological, biological, or organic contexts that dynamically influence each other in connection with the physical and social environment. Another important consideration in the foundation for presenting the assessment findings is the acknowledgment that developmental behavior and function are not determined solely by the circumstances at birth. The human brain demonstrates a fascinating capacity to adapt and adjust to difficulties and conditions. Moreover, a child's development is a complex, dynamic process with many influences that occur over time in addition to the structural and functional changes within the infant's central nervous system.

The process of infant behavioral assessment has shifted to embrace a range of competencies building on strengths, rather than deficiencies. The recognition of the infant's functional competence led to the integration of the infant's regulatory efforts (or the infant's capacity to cope with his or her experiences) and the infant's interactive competencies, to be viewed within a social context.[24] The focus is on individual differences and what influences those differences, as well as the conceptualization that these differences affect the caregiver's behavior in addition to the infant's developmental trajectory. A priority for health professionals is to offer helpful and accurate information that can facilitate parent–infant attachment. Assessment of the infant's environment and of family and social interaction must also be included to assess the capacities and needs of infants who may be considered at risk

during the perinatal period.[25] Ideally, the assessment should be repeated at different time intervals to determine the infant's current level of behavioral functioning while identifying strengths and vulnerabilities. Repeating the assessment provides the family with more information about how to best interpret their infant's unique set of cues and behaviors.

APPROACH TO BEHAVIORAL ASSESSMENT

Identifying conditions of risk, describing behavior patterns, and evaluating developmental function are all important aspects of the neurobehavioral assessment. Creditable evaluation relies primarily on the examiner's astute, accurate observational skills. The examiner must observe the infant's ability to organize, recognize signs of disorganization or stress, and modify the interaction based on the infant's behavioral response.

It is important to follow the basic principles of physical examination when assessing the infant's behavior. Perinatal history will provide important information on factors that will affect the infant's ability to interact with the environment. Matters, such as time elapsed since birth, type of labor and delivery, as well as drugs taken during pregnancy and delivery by the mother, must be considered. Gestational age as well as postnatal age and medical status will have a significant impact on both the behavioral findings and the infant's ability to participate actively in the examination. An infant with respiratory distress will exhibit different behaviors than one who is not in distress. Caregiving and activity before the examination must also be considered. Research with preterm infants has demonstrated that the sequence of care and the clustering of caregiving interventions can affect the infant's responsiveness and capabilities.[26,27]

The physical and social environment are important considerations that may affect the outcome of the examination. A warm, calm, quiet, softly lit room provides the best environment in which to observe the infant's responses to stimuli and ability to regulate state. Performing the examination in the presence of the infant's parents or caregivers provides a valuable opportunity for the parent to better understand the infant's behavior and competencies.[28] Other supports that might be needed for some infants to perform at their best include swaddling and containment. These postural supports encourage a tucked, neutral position. The use of rest periods allows the infant to regain energy and focus during the examination. Another strategy to reduce the effects of the sensory environment is to reduce sounds and activity in the infant's immediate environment and thereby decrease stressful stimuli for the infant. These infants are easily stressed. There is often little leeway for the young, small, physiologically compromised infant. The infant who surpasses autonomic regulation thresholds experiences behaviors such as desaturations or bradycardia.

The infant's state of consciousness, or "state," is another important consideration.[29] An infant's state depends on a variety of factors, such as time of last feeding, recent events (e.g., blood tests or circumcision), and the infant's individual sleep–wake cycle. It is important to be aware of these other factors and consider them in relationship to performance during the examination. The response to stimuli and behavioral cues will vary according to the infant's state. Preterm infants exhibit subtle behaviors that are more difficult to identify than term infants. The examiner needs to be flexible to obtain the most valid outcomes and conclusions. It is important to remember while caring for these vulnerable infants that there is only a small window of leeway for state organization; infants can quickly go from being actively engaged in the interaction to being disorganized and stressed to the point of fatigue or withdrawal. Thus, caregivers are always balancing the need for assessing the infant's competence with the knowledge that the NICU sensory experiences may be impinging on the infant's ability to demonstrate appropriate behavioral responses.

In many intensive care nurseries and follow-up clinics, neurobehavioral assessment

is an essential part of the comprehensive care of the high-risk newborn. Evaluating behavior is a useful concept because it facilitates understanding the infant's ability to cope with their experiences.[30] Evaluation also helps to provide an understanding of the infant's interactional capacities, including alerting and orienting to caregivers' faces and objects in the environment. Examples of behavioral assessment of three different infants (well newborn, well preterm infant, and sick preterm infant) are found at the end of this chapter with the descriptions of the instruments that can be used for behavioral assessment. These examples demonstrate the need for the examiner to have a comprehensive understanding of infant behavior and the proper training to assess neurobehavioral functioning. This understanding is necessary for developing a rapport with the infant and administering a developmental assessment.

Behavioral assessment is intended to identify an infant's current level of balance and smooth, integrated functioning. In other words, it is designed to identify in what situations and with what supports the infant exhibits organized behavior. It is also the purpose of the assessment to describe the threshold of disorganization indicated in the infant's behaviors of coping and avoidance. The degree and kinds of stress and intensity of frustration the infant experiences can be indicators of the degree of energy the infant has available. The behavioral assessment must also determine whether there is any leeway in the infant's responses. For example, if the infant can tolerate being moved from the examiner's shoulder (where he is flexed, tucked, and resting calmly) to the lap without an onslaught of disorganized behaviors (such as extended arms and hands, grimaces, fussiness, and irregular breathing), then the infant is organized, even if working hard to achieve that organization. If disorganization is unavoidable, even when sensitive handling is provided, the infant needs a great deal more facilitation before, during, and even after caregiving, with minimal activity during handling.

It is the intent of the behavioral assessment to describe the degree of competence and organization in the infant's behavioral repertoire, as well as when and how behaviors reflect disorganization or stressful responses. Are there changes in color, breathing, or movement patterns as the infant is moved? Or can the infant be moved from one place to another with minimal adjustments and limited cost to behavioral subsystems? What strategies does the caregiver need to employ to help the infant function smoothly? The behavioral assessment provides data that can answer all of these questions. The next several sections outline parameters often included in a behavioral assessment. It is important to note that not all parameters are included in all behavioral assessments. Use of some parameters is dependent on the status of the infant. At times, a chosen tool may bring each parameter used together in a meaningful way. For reliable administration, a thorough understanding of the elements are needed. Once each parameter is discussed, then several different behavioral assessments are described with how they might be best used in the clinical setting.

IDENTIFYING STATES OF CONSCIOUSNESS

State refers to the level of consciousness exhibited by the infant (Table 12.1). State is determined by the level of arousal and ability to respond to stimuli. The infant's behavior, function, and response to the environment will depend upon which baseline state he is in, ranging from deep sleep to vigorous crying. Healthy infants can use their state to exert control over environmental input, but this ability may be limited in the preterm or sick infant. Often the preterm infant is awake with eyes closed; this can be mistaken for sleep. Premature and critically ill infants often have difficulty achieving the entire range of states; for example, a defined state of deep sleep or robust crying. Behavioral assessment begins with evaluating the infant's ability to control state, move smoothly from one state to another, and maintain alertness.[26,31,32]

TABLE 12.1: NEONATAL BEHAVIORAL STATES

STATE	CHARACTERISTICS
Deep sleep	No eye movements No activity Regular breathing
Light sleep	Low levels of activity Rapid eye movement possible
Drowsiness	Variable activity levels Dull, heavy-lidded eyes that open and close
Quiet alert	Wide, bright eyes Attention focused on stimulus
Active alert	Increased motor activity Periods of fussiness Irregular respirations
Crying	Increased motor activity Color changes

State is determined by observing an infant's level of arousal and accompanying behaviors or cues. Several scoring systems have been developed for identifying infant states. Brazelton's system is the most widely used and easiest to follow, particularly for the term infant.[20] For the preterm infant, more definitive state definitions may be useful, such as those developed by Als, Holditch-Davis, and Anderson.[26,33–35] The Anderson Behavioral State Scale (ABSS) has been used often in nursing research because it is easily learned and provides interrater reliability.[33] The ABSS was devised by Gene Anderson specifically for use with preterm infants. It is based on the works of Parmalee and Stern, and Burroughs and coworkers.[36,37] The underlying theoretical framework for this state scale is different from that of other scales. Most other state scales are nominal in nature: the coding is a categorical representation of clusters of behavioral states. These state scales, then, have been designed to capture the *qualitatively* different aspect of behavioral clusters. The ABSS was designed with consideration for the linear relationships among the states, heart rate, and energy consumption. Thus, the differences are *quantitative* or ordinal in nature.[36] The ABSS is particularly useful for preterm infants because it breaks down the typical five or six states into 12 states. This delineation more closely captures behavioral states exhibited by preterm infants because differentiating sleep–wake states is more difficult in preterm infants. For example, the ABSS has four measures of sleep, five measures of awake, and three measures of crying, allowing a more sensitive indication of infant behavioral state.

Interobserver reliability with the ABSS has been easier to establish than with other scales. ABSS scores range from 1 to 12. The infant's sleep is categorized within scores of 1 to 4; the behavior observed with infant's eyes closed (sleep) is used to differentiate the actual score itself. Scores of 1 or 2 are quiet sleep states and represent no body movement; these are considered optimal for recovery and/or growth because of decreased energy expenditures. Scores of 3 or 4 are more active sleep states, with beginning awareness of the environment. Apnea due to disorganized breathing is most likely to be seen in state 3 or 4. Drowsiness with eyes open and closed at times is a score of 5. Scores of 6 or 7 are alert states considered optimal for perception, interaction, and learning. Increasing activity with a degree of alertness are scores of 8 or 9. Scores of 10 to 12 represent increasing levels of agitation and crying. The examiner assigns a state based on ease of identification (the infant clearly exhibits that state). This may be more difficult in preterm or ill infants, whose states may be more fleeting.[27–29]

Sleep States

Deep sleep is characterized by closed eyes with no eye movements, regular breathing, and no spontaneous activity. There is a delayed response to external stimuli and then only a brief arousal, followed by a return to deep sleep. Isolated sucking movements or startles may be noted. Preterm infants may demonstrate a difference between very deep

sleep, still sleep, and deep sleep with startles or muscle twitching.[35]

Light sleep consists of low levels of activity, with greater variability in response to external stimuli (Figure 12.1). Rapid eye movement may be observed. Preterm infants may exhibit irregular respirations. Infants in light sleep may startle or make brief fussing or crying noises. Parents may need support in delaying response to these brief episodes during the light-sleep phase. Active sleep and lower level alert states are seen more often in preterm infants than in term infants, who spend more time in deep sleep and quiet alert states.[33–35]

Transitional State

Drowsiness is characterized by a variable activity level, with smooth movements and occasional mild startles (Figure 12.2). The eyes open and close and appear dull and heavy lidded. The infant will react to stimuli, but the response is often delayed, and the infant may startle easily. From the drowsy state, the infant may either return to a sleep state or move to a more alert state. Caregivers may arouse an infant into a quiet alert state by providing an auditory or visual stimulus. Nonnutritive sucking has also been found to be effective in calming and bringing an infant to an alert state.[37,38]

Awake States

Quiet alert refers to the state in which the infant interacts most with the environment by exhibiting a brightening and widening of the eyes and an alert appearance (Figure 12.3). Attention is focused on available stimuli, whether visual or auditory (Figure 12.4). A minimal amount of motor activity is noted, and respirations are regular. This state provides the greatest opportunity for infant interaction with caregivers. Term newborns commonly experience a period of quiet alertness in the first few hours after birth, providing an opportunity for parents to interact with their infant.

Preterm infants may have difficulty maintaining a quiet alert state for long. Alert periods may be brief and fleeting. The infant may become *"hyperalert,"* with an inability to decrease or end fixation on a stimulus (Figure 12.5). Preterm infants may also appear awake and alert but be unable to involve themselves in interaction.[21,35,39,40] During these periods, they are often noted to use gaze aversion to manage the overwhelming stimuli in the nursery environment.[40–42]

The *active alert* state is characterized by increased motor activity, with heightened sensitivity to stimuli. The infant may have periods of fussiness, yet is consolable. Although open, the eyes are less bright and attentive than in the quiet alert state. Respirations are irregular. The term infant may be able to use self-consoling techniques to return to a quiet alert state (Figure 12.6). The preterm infant will usually become distressed and unable to organize. Interventions such as waiting for the infant to settle, swaddling, containing, and reducing other environmental stimuli can be provided by caregivers to help the infant return to a quiet alert state.[35]

Crying is accompanied by increased motor activity and color changes. The infant is very responsive to unpleasant stimuli, both internal and external. Some infants are able to console themselves and return to a lower state, whereas others need help from caregivers. Preterm infants may exhibit a very weak cry, or their cry is not audible due to intubation. They may demonstrate color changes, alterations in motor activity, and other signs of stress, such as a crying face, apnea, vomiting, or decreased oxygen saturation.[26,29–31] The infant may also extend and straighten his extremities, then lie still, remaining extended without the capacity to return to a neutral, flexed posture as energy is lost, the infant becomes flaccid and appears depleted. The question the examiner must ask when these behaviors are observed is, "What does it take from the environment or caregiver to settle and help the infant recover and return to a modulated, interactive state?" Awareness of the infant's behavior and the ability of the caregiver to support the infant are core components of individualized, developmentally supportive care practices.

Figures 12.1–12.9 Progression of infant through states of light sleep to crying; demonstrating time-out signals with visual stimuli

Figure 12.1 Light sleep

Figure 12.2 Drowsy

Figure 12.3 Quiet alert

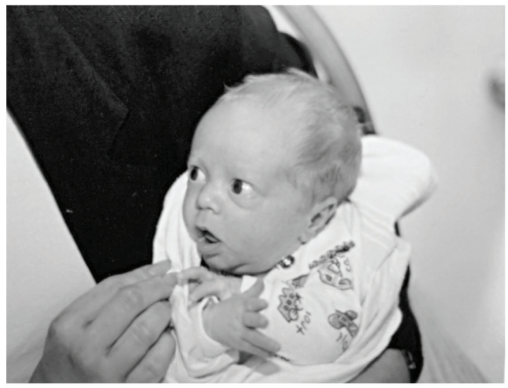

Figure 12.4 Signs of attentiveness

Figure 12.5 Hyperalert response to stimulus

Figure 12.6 Self-consoling behavior; hand-to-mouth movements

Maintenance of State

Although the states can be distinguished from one another, the infant makes frequent transitions among them changing from one state to another several times in the course of the examination. The term infant should display smooth transitions between states and move from sleep to drowsy to wakefulness instead of moving from sleep to agitation with robust crying. Excessive lethargy or irritability is abnormal. The preterm or neurologically impaired infant may exhibit sudden changes between sleep and awake states, but abrupt state changes in the seemingly healthy term infant are a cause for concern.[33,35]

The ability to maintain an alert state with interaction varies among infants. Some have difficulty becoming alert initially and then struggle to maintain this state for any length of time. The infant may need facilitation from a caregiver to sustain alertness. Other infants have trouble filtering out noxious stimuli and progress rapidly to active alert or crying, becoming disorganized. Swaddling or a quiet, darkened environment may help these infants remain alert and focus on a single stimulus.

The examiner should acknowledge the amount of time the infant spends in the quiet alert state or focusing on a stimulus and reinforce the infant's attempts to build positive trusting interactions. Infants who have difficulty remaining alert can be frustrating for caregivers and parents. The examiner may spend time with parents exploring opportunities to support the infant to maintain an interactive alert state. The examiner should consider the situation and ask, "When does the infant look most comfortable?" or "What supports are necessary for the infant to be successful or comfortable?" The examiner then incorporates these individualized strategies, such as postural support, that are appropriate for this particular infant into the interaction with the infant.

Preterm infants have brief periods of alertness, and may have difficulty maintaining this state. Brazelton describes the "cost of attention" as the amount of energy the infant must expend to maintain an interaction.[43] This cost of attention varies, depending on the health and maturity of the infant. Premature or sick infants show fatigue or stress sooner than do healthy term infants.[41,42] The caregiver's supportive responsiveness to the infant's distress is imperative to building trusting relationships.

Signs of stress or fatigue may include color changes, irregular respirations, apnea, changes in tone, irritability or lethargy, and vomiting. The infant may change states rapidly from crying to sleep or may become hyperalert. The examiner must be able to recognize these signs of stress and fatigue and support the infant or discontinue the examination when appropriate. After the infant has had a period of rest, the examiner may be able to begin again. The cost to the infant or the amount of energy expended during the examination should be noted.[27,36] The preterm or ill infant may require completion of the examination in pieces, which is not optimal; yet completion in one time period may be too much for vulnerable infants.

ORGANIZATION

Organization reflects the infant's ability to integrate physiologic and behavioral systems in response to the environment without disruption in state or physiologic functions.[26,44] Physiologic functions include such parameters as heart rate, respiratory rate, oxygen consumption, and digestion. The behavioral system includes state (attention and self-regulation) and motor activity (tone, movements, and posture).

The organized, robust infant maintains stable vital signs, smooth state transitions, and even movements when interacting with the environment. The infant is able to self-console or be consoled easily and can habituate to or block out overwhelming stimuli. The disorganized infant will react to the environment with sudden state changes and will exhibit frantic, jittery movements, color changes, and irregular respirations. Some infants will respond with hypotonia. The ability to maintain organization depends on the infant's maturity level and overall

well-being. Individual temperament may also play a role in organizational ability.

In evaluating organization of motor behavior, the examiner assesses the infant's movement patterns, energy level, and tone. Hand-to-mouth maneuvers in an attempt to self-console are purposeful movements achieved by the mature, well-organized infant (see Figure 12.6). When a cloth is placed over the face of a term neonate, the infant will attempt to remove it by arching, rooting, and swiping at the cloth covering the eyes/face. As during most assessments of behavioral maturity, the preterm infant may have a diffused or delayed response.

Infants who are easily overwhelmed will benefit from care designed to enhance their organizational ability to cope and self-regulate. Clustering care to allow for uninterrupted restorative sleep, arousing the infant slowly, and introducing one stimulus at a time are all interventions that support the infant in being more successful or competent. However, it is important to consider those caregiving tasks that are clustered together and their overall effect on the infant; there is always a cost–benefit ratio to consider.[26,27] Providing postural support to encourage the infant to stretch and squirm to help steady breathing yet return to a neutral, tucked posture is essential for the infant to develop productive coping mechanisms. Being aware of and modifying sensory input is important so as not to increase stress for the high-risk infant. Some infants, however, can tolerate help in modulating their behavior during the examination. Quietly speaking to the infant in a way that provides comfort while you interact can also be a supportive strategy or intervention to obtain a comprehensive impression of the infant's strengths and vulnerabilities.

RECOGNIZING THE SENSORY THRESHOLD

Sensory threshold refers to the level of tolerance for stimuli within which the infant can respond appropriately. After reaching or exceeding this threshold, the infant becomes overstimulated and exhibits signs of stress and fatigue (Table 12.2). Preterm and neurologically impaired infants may have low thresholds compared to healthy term newborns. What would normally be considered routine care (e.g., talking to the infant during feeding) may be overstimulating to an infant with a low sensory threshold. These infants might do better when presented with a single stimulus or modified, muted sensory inputs such as providing a comforting quiet voice or briefly catching the infant with eye contact.

TABLE 12.2: SIGNS OF OVERSTIMULATION (TIME-OUT)

Gaze aversion
Frowning
Sneezing
Yawning
Hiccuping
Vomiting
Mottled skin
Irregular respirations
Apnea
Increased oxygen requirement
Heart rate changes
Finger splaying
Arching
Stiffening
Fussing, crying

The mature newborn has a unique ability to regulate physiologic and emotional response to a variety of stimuli. It is the infant's way of learning to control the effects of the surrounding environment. Evaluating these responses to the environment allows the examiner to design an individualized plan of care that is unique to that infant.[43–45] Knowledge of these responses also facilitates parent competence and involvement.[23,25,46] Infants who are easily overwhelmed may require more frequent breaks during caregiving to adapt and regulate responses.

TABLE 12.3: SIGNS OF APPROACH (ATTENTION)

Quiet, alert state
Focused gaze
Dilated pupils
Regular respirations
Regular heart rate
Rhythmic sucking
Reaching or grasping
Hand-to-mouth movements

Observing Behavioral Cues

An infant's behavior indicates physical, psychological, and social needs. Caregivers who are responsive to these cues develop reciprocal relationships with the newborn. Responding to an infant's behavioral cues also reinforces behavioral organization.[21,35,47] This reciprocity helps to build trust, which is an essential component of emotional development and a key component in the foundation of secure attachment.[48] The infant relies on individualization, responsiveness, and respectfulness, and should be integrated into each interaction.[49]

Signs of approach (or attention) indicate the infant is ready to interact with the caregiver or the environment. Approach behaviors include an alert, focused gaze; regular breathing; and dilated pupils. The infant may also exhibit grasping, sucking, or hand-to-mouth movements (Table 12.3; see Figure 12.6).[34,35]

Avoidance behaviors (time-out signals) indicate the infant is becoming tired, overstimulated, or stressed and needs a break from the stimulus or interaction. Avoidance behaviors include averting the gaze, frowning, sneezing, yawning, vomiting, and hiccupping. The infant may also display finger splaying, arching, stiffening, or crying (Figures 12.7–12.9; see Table 12.2). Color changes, apnea, irregular breathing, and decreased oxygen saturation may also indicate the infant's need for time-out.[33,34,47] A state change may be an avoidance behavior,

Figures 12.1–12.9 Progression of infant through states of light sleep to crying; demonstrating time-out signals with visual stimuli (*Continued)*

Figure 12.7 Sign of overstimulation in response to stimulus

Figure 12.8 Sign of overstimulation in response to stimulus

Figure 12.9 Crying as a response to continued stimulus

as demonstrated by the infant who withdraws or shuts down entirely by falling asleep during repeated or prolonged painful procedures. It is important to remember that while some infants may muster up the energy to protest to a noxious stimulus, they seldom have the autonomic stability to sustain the response. Healthcare providers must use these cues in planning routine care if they are to best support the infant and decrease energy consumption.[41,42]

Reflective strategies can help caregivers better understand their own perceptions of the infant's experience and can be used to strengthen the critical thinking skills needed for comprehensive assessments of vulnerable infants.[50] Reflection provides an opportunity to shift the focus of learning to include self-knowledge as a component of competency.[51] Schön provides a framework for reflection in which the knowledge or act of knowing comes from within the practitioner.[46,52] *Reflection* can also be defined as a process followed to create meaning from our experiences to guide our decision making to take the next steps of action.[50] This mindful awareness of the infant's behavior prepares the caregiver to sustain responsive and respectful interactions. Reflection is a key element of individualized developmental care practices and can be defined as the caregiver's recollection or memory of the interaction, journaling, or talking things through with another person.[53,54] These insights or newly gained knowledge through reflecting on the infant's behavior can be utilized for future interactions to be more supportive of infants' strengths and challenges.[55,56]

HABITUATION

The infant's ability to decrease a response to a repeated stimulus is referred to as *habituation.* When a stimulus is repeated, the infant's initial response will gradually disappear. Habituation provides a defense mechanism for shutting out overwhelming or disturbing stimuli. For example, healthcare providers often habituate to the noxious noise of the intensive care nursery.

Habituation is best assessed when the infant is asleep or in a drowsy alert state.[22,37] The stimulus can involve the visual, auditory, or tactile senses. Visual habituation can be easily assessed by shining a light briefly onto the infant's closed eyes during sleep from 10 to 12 inches away. Repeat the stimulus every 5 seconds to a maximum of 10 times or until the infant ceases to respond (whichever comes first). Note the presence of startles, facial grimaces, blinking, and respiratory changes. If habituation occurs, responses will become delayed and eventually disappear. Infants who are able to habituate successfully to protect their sleep usually do so within five to nine flashes.

The infant's ability to habituate to an auditory stimulus can be tested in the same manner, using an object that makes a noise (e.g., a bell or rattle). Hold the object 10 to 15 inches from the baby and shake the object for about 1 second. Responses may include startles, squirming movements, facial grimaces, and respiratory changes. The following stimuli should be provided when the infant stops responding. Note the infant's ability to decrease her or his responses as the stimulus is repeated or if the infant continues to respond longer than 45 to 60 seconds, indicating he or she may need facilitation from the examiner to stop. As they do with visual habituation, most term infants decrease their reaction after five to nine repetitions.

Habituation to tactile stimulation can be determined by pressing the sole of the foot with a smooth object. Repeat the stimulus every 5 seconds. The infant may begin with a generalized body response, pulling both feet away. The response will gradually decrease to only the involved foot or will disappear altogether.

The ability to habituate varies among infants. Some (including those who are preterm) have difficulty tuning out noxious stimuli.[57] They are easily distracted and then become irritable and disorganized, displaying signs of stress and fatigue. The premature infant's brain development is strongly influenced by random frequent sensory stimulation. Their inability to cope with environmental sights and sounds may make

interactions and feeding difficult. These infants may need to be fed in a quiet, darkened room or presented with one stimulus at a time during their quiet, alert state. Work with parents to reinforce identification of and response to their infant's behavior. This can facilitate a positive relationship between the infant and the family, supporting parental competence.

RESPONSE TO STIMULI

Visual Stimuli

The newborn has the ability to focus on and react to a variety of stimuli in the environment. The examiner should observe and record the infant's response to visual and auditory stimuli. For optimal evaluation, responses should be assessed with the infant in the quiet, alert state.

Two tests for visual response can be performed with the newborn. The first is a response to light. When a light is directed toward the infant's eyes, an appropriate response is a grimace and closure of the eyelids. This response will lessen with habituation. The second test evaluates the ability to fixate on an object and track it. Term infants are able to fixate briefly on an object (e.g., a face or a red ball). The newborn's visual field is fairly narrow, with the ability to focus on objects at a distance of about 10 to 12 inches. Objects closer or farther away will be ignored because newborns have decreased visual acuity that improves with maturation. The term newborn is able to follow or track an object horizontally about 60° and vertically about 30°, often with some head movement.[20,26]

The pupillary reflex develops at approximately 30 weeks gestational age. Without this reflex, premature infants have a limited ability to maintain protective lid tightening and therefore should be protected from bright lights and visual stimulation.[58] As the pupillary reflex matures, preterm infants beyond 30 weeks gestational age demonstrate both response to light and ability to focus on simple patterns.[59] Preterm infants may take longer to focus on an object, as they have less visual acuity than term newborns, which accounts for this variability in response. It is important to continue monitoring light levels, protecting the infant's eyes from direct bright light. In the preterm infant, closed eyelids provide less protection from bright lights than they do in more mature term infants. Behavior responses to visual stimuli should be evaluated for signs of stress and fatigue.[57] An eye examination for the preterm infant may be a noxious procedure for which the infant may need supportive interventions to manage.

Auditory Stimuli

When in the alert state, newborns will respond to an auditory stimulus with brightening of the eyes and face and turning of the head in search of the sound. A rattle, bell, or voice will work well as an auditory stimulus. Keep in mind that a newborn may tune out a noxious auditory stimulus. With the baby's head in midline, initiate the stimulus 6 to 12 inches away from one ear, out of visual range. The infant should open their eyes and turn their head toward the sound; continue by alternating the stimulus on each side with sounds of varying rhythm and intensity based on the infant's responses.

Preterm infants begin to orient (look at and focus) to a soft sound source by approximately 28 weeks gestational age, but often demonstrate sensitivity to obtrusive sounds and physiologic instability in the presence of random loud sounds. The use of soft voice and rhythmic cyclical auditory stimulation can be introduced based on the infant's behavioral response.[60] Careful attention must be paid to limiting environmental sounds because it has the ability to interfere with the responses to the behavioral examination. White and associates provide design guidelines for limiting sound in the NICU.[61]

EVALUATING CONSOLABILITY

Infants' abilities to quiet when in a crying state vary. The well-organized infant

demonstrates observable activities to self-console during the course of examination. These include bringing the hands to the mouth; sucking on the fist or tongue; and using environmental stimuli (visual or auditory), such as a soft human voice, to self-console (see Figure 12.6).[26] Infants who make limited attempts or who show decreased ability to self-console may be more irritable or sensitive to stimuli.[26,62]

Most infants will respond to consoling attempts by caregivers. Irritable infants may be easily disturbed by stimuli from the environment and may be slower to respond (or may not respond at all) to attempts to console them. The examiner should try interventions that can lead to consoling the infant, such as talking softly, providing hand containment or placing a hand over the infant's stomach, flexing the extremities near the trunk to prevent startle activity, holding, rocking, or offering nonnutritive sucking. Decreasing such environmental stimuli as light, sound, and sudden movement may also be helpful. A common mistake is trying several interventions at the same time (e.g., rocking, talking softly, *and* offering a pacifier).[63] A combination of activities may overstimulate some infants. Therefore, limit the interventions to one at a time before using them as a group. If one intervention fails, try a different one, and note which interventions work for this particular infant so they can be used again in the future.

IDENTIFYING TEMPERAMENT

Some babies seem more difficult; others appear to be easier to care for. *Infant temperament* has been defined as the infant's behavioral style; how the infant behaves in relationship to the environment and caregiving interaction.[64] The developing relationship between infant and parent is affected by how the child's temperament is exhibited and perceived. The infant is neurobiologically connected to the parents. When there is synchrony within this dyad, there is said to be "goodness of fit." When synchrony is lacking, the infant is perceived as difficult and/or demanding by the mother/father (primary caregiver), and the asynchrony of the relationship predisposes the infant to long-term negative outcomes. Parents' perceptions of infant temperament are important and should be considered as individualized plans of care are created. Parents' perceptions and beliefs about the attributes of the infant affect how parents care for their infant and the symbiotic relationship that will support the infant's cognitive development. Thus, infant temperament has been measured by asking parents about their infants. How do they perceive the infant? Is the infant calm or demanding? How easily is the infant consoled or is the infant inconsolable? Understanding these dimensions can help the parent acquire realistic expectations for the child's behavior and perceive that he or she can meet them. Consider using reflective strategies, such as journaling or talking about the interaction with the infant, to support the parents in exploring their perceptions and better understanding their infant's behavior.

As stated earlier, *temperament* refers to the way an individual interacts with the environment. Chess and Thomas describe nine behaviors that define variations in temperament.[64] A description of each behavior follows:

> *Activity level* refers to motor activity such as playing, dressing, eating, crawling, and walking. Sleep–wake cycles and their durations are also used in scoring activity level. Some infants are very active, with short sleep cycles; others are less robust.
>
> *Rhythmicity* refers to the regularity of functions such as hunger, sleep–wake patterns, and elimination.
>
> *Approach* or *withdrawal* describes the individual's reaction to a new stimulus such as food, a new toy, or a new person. Approach responses are positive; withdrawal responses are negative reactions to the new situation.
>
> *Adaptability* is the individual's response to new situations once the initial response has passed. Adaptability

examines the ability to adjust to the new situation or environment.

Threshold of responsiveness refers to the amount of stimulation required to generate a response, either positive or negative.

Quality of mood describes the overall mood of the individual or the amount of pleasant, friendly, happy behavior versus unpleasant, unfriendly, or fussy behavior.

Intensity of reaction is the level of energy in a response, whether positive or negative.

Distractibility is the ability of extraneous stimuli to interfere with the individual's current behavior.

Attention span or *persistence* refers to the length of time an individual will pursue a specific activity, especially when obstacles interfere with it.

Based on these behaviors, three categories of temperament can be defined and frequently identified in the newborn:

1. The "easy" baby demonstrates regularity, positive approaches to new situations, adaptability to change, and an overall positive mood.
2. The "difficult" baby has an irregular schedule; trouble adapting to new situations; a low threshold for stimuli; and intense, often negative moods.
3. The "slow-to-warm" infant is characterized by mild intensity, positive or negative moods, and slow adaptation to new situations and people. These infants need repeated, slow exposure to a situation before they will respond positively.

Understanding and supporting an infant to best optimize temperament can help parents create an environment that will maximize their child's positive characteristics and minimize frustration.[58] Parents of a slow-to-warm child can allow extra time to adapt to new situations. The infant with a low sensory threshold may be easier to care for if activity is limited to a stimulus or two at a time.

TERM NEWBORN BEHAVIORAL ASSESSMENT

Perhaps the best known tool for behavioral assessment of the term infant is the Neonatal Behavioral Assessment Scale (NBAS) developed by Brazelton.[43] The tool was designed for use with healthy newborns from about 36 to 44 weeks gestation. A complete behavioral assessment using the NBAS takes about 30 minutes and is best administered by a trained examiner. However, even limited aspects of the examination performed by an untrained examiner can be used to provide helpful information about the infant's neurobehavioral status.[24,58] There are also several studies in which the NBAS has been done in the presence of families to encourage them to see the vast capabilities of their infant.[30,65] The NBAS assesses the newborn's response to 28 behavioral items, each scored on a 9-point scale, and 18 elicited responses, each scored on a 4-point continuum. These items provide information about the newborn's ability to respond and adapt to his environment. Such items as reflexes, state regulation, orientation to visual and auditory stimuli, habituation, motor performance, and interaction with caregivers are assessed. The examination is usually done in one sitting, and items are most often presented in the same order to all infants.

The focus is assessment of the infant and intervention with the family.[22] The parents and nurse work together, using a nonjudgmental approach, to observe and understand the unique behaviors of the infant. Specific items selected from the NBAS by the nurse create opportunities to emphasize the infant's capabilities and to facilitate reciprocal interaction between parents and their infant. Nurses trained in the NBAS can use this assessment in any setting where nurses see newborn babies in the first months of life. The model is based on the assumption that the parents and newborn are dynamically inseparable and that the family and infant require individualized attention. The nurse selects the behaviors and interactions with the infant that will have the most positive impact on establishing a parental–infant

bond. During the examination, the nurse identifies infant strengths and builds on these successful strategies the infant is demonstrating. The nurse/or examiner can also identify the infant's efforts to comfort and self-console as well as how the infant copes with the demands of the experience. The nurse facilitates/guides parents to be most responsive to their infant. Anticipated areas of concern are addressed, and parents are encouraged to understand and communicate with their infant in ways that promote understanding of individual behavioral needs and communications.

The NBAS focuses on motor responses and maturity, interactive skills, visual and auditory orientation, management of sleep–wake cycles, and physiologic integrity and reflexes.[30,65] Self-quieting activity is highlighted, and parents learn how their infant can calm himself by bringing hands to mouth, sucking, looking, and changing position. If a baby cannot self-console, the nurse points out cues that signal stress or disorganization. Parents can then be aware of when their infant is stressed and modulate their reactions. Parents can be shown how to watch for stress cues and how to intervene without either overwhelming the infant with too much stimulation or failing to offer enough intervention to soothe.

The examination is structured with a preferred order of presentation of items. The infant's state plays a major role in the evaluation; therefore, observing the state of consciousness becomes the starting point. This sample evaluation using the NBAS describes some of the major responses of a healthy term newborn, but space does not permit description of the entire examination.

Initially, the examiner evaluates state of consciousness by observing respirations, eye movements, startles, and body movements. A specific scoring system is utilized to describe the initial state and the predominant states throughout the examination. The range and variety of state changes are noted. Once state is observed, habituation is evaluated. Habituation is the degree to which the nervous system reduces or inhibits responsiveness to a repeated stimulus until shutdown. Typically, the examination begins with the infant asleep or in a drowsy state. A flashlight is passed across the sleeping infant's closed eyes up to 10 times, and the degree of response and response decrement is noted. The assessment is repeated with a rattle and then a bell to note the infant's ability to shut out sound after repeated presentations. A typical response would be a startle or dramatic movement to the first stimulus, followed by decreasing responses to each of the remaining nine presentations of the stimulus. A range of responses is possible, but overall, one would like to see the infant react strongly at first and then show a reduction of responses over time. Next, the supine sleeping infant is uncovered slowly and the response to this change is noted. One may see no change, postural changes, color or breathing changes, or state changes. Next, a series of reflexes is evaluated with the infant still in a supine position. First, the reflexes of the hands and feet are evaluated, including plantar grasp, Babinski, heel prick, ankle clonus, passive tone in legs and arms, rooting response, sucking reflex, and glabella reflex. Each item is presented in a specific pattern with a watchful eye for change and variable responses.

Because the infant is supine, the examiner gently undresses the infant prior to assessing the palmar grasp and pull-to-sit reflexes. The infant is then picked up with his or her head facing away from the examiner, and standing and stepping reflexes are tested. The infant is then placed prone on the bed to evaluate the crawling reflex. Next, the infant is picked up to evaluate truncal incurvation. The infant is placed prone over the examiner's hand and raised in the air, and the examiner skillfully strokes along the right and left of the spine to evaluate for hip swing. The infant is then moved through space in a brisk manner, initially in an upright position, then in the horizontal position, and tonic deviation of head and eyes, and nystagmus are assessed by eliciting the infant's vestibular reaction and neurobiological capacities. These maneuvers also allow for observation of the degree and type of balance, and differentiation between relative extensor and

flexor tone and posture. The infant is then held close to the examiner's body to evaluate cuddliness. A cloth is placed over the baby's eyes to evaluate attempts to remove it by swiping with the arms in a defensive movement pattern. Tonic neck reflex and Moro reflex are then tested.

By this time in the examination, the examiner can begin to define what is necessary to help this infant stay in a calm, alert state. When the infant does alert, the social interactive package can be administered. To set the stage for administration of these items, evaluate state, check room temperature and lighting, and have a comfortable chair available. Gently move the infant to the examiner's lap, providing postural supports. Ideally, this change will not disrupt the alertness that has been achieved by the infant. The examiner then presents a red ball, a rattle, and the examiner's face alone, voice alone, and the two together. The responses that are evaluated are the degree of visual fixation, gazing, and any avoidance reactions, such as turning away, or nonresponsiveness. In addition, the examiner may see the degree of recognition of voice and sound in the infant's turning with head and/or eyes to "see" the sound off to the side of each ear.

Throughout the administration of these items, the examiner is vigilant in watching for state changes (e.g., change in the quality of alertness, becoming fussy, or crying). All state changes (if they last at least 15 seconds each) are recorded. The lability of crying and alertness are scored along with irritability. The degree of excitation and ease in consoling the infant are important (self-quieting and consolability). As crying or fussiness develops, the examiner does not immediately stop the cry, but gives the infant a chance to self-quiet, then offers a specific sequence of supports to see what level of support quiets the infant. The examiner watches for any attempts or strategies used by the infant to self-organize and quiet himself or herself (such as hands to mouth, bracing a foot, or sucking). The buildup to a full, intense cry state becomes important so the examiner can see the degree of self-quieting, as well as what it takes to calm the infant. If, at any time, any physiologic instability is observed, the examiner terminates the examination.

Throughout the examination, the examiner is scoring the infant's best performance. The examination moves from presentation of simple to more vigorous stimuli. A typical term newborn may show a variety of responses, from smooth responses that appear to come steadily and without cost or effort to vigorous activity levels with intense upset, demonstrating a need to be comforted. Some infants are more difficult to comfort than others. They may need specific levels of stimulation to elicit their ability to use the comforting resources available to them. "Normal" encompasses a wide range, and all of these responses can fall within normal limits.

Case Assessment of a Full-Term Infant

Jenna is a healthy 3-day-old newborn. Her mother, Helena, had recovered from her delivery and was eager to get to know her baby, but had been told that babies do not see well for the first few weeks. Helena had many questions when the nurse came in to discuss Jenna with her. The nurse noted that Jenna's mother had already learned several things about Jenna: How she liked to suck on her fist, what made her cry, and even how to calm her successfully. When the nurse pointed out Jenna's alertness and explained that Jenna could see her mother even at this early stage, Helena seemed skeptical. The nurse suggested that because Jenna was awake now, Helena should hold her in front of her and move her face out of Jenna's vision, call her name, and talk to her. As soon as Jenna heard her mother's voice, her eyes shifted in the direction of her mother's face. Encouraged to keep talking, Helena was extremely pleased as Jenna continued to look and search until she found her mother's face and voice. As Jenna gazed at her mother, the nurse pointed out that if she kept Jenna's face within 10 to 12 inches of her own face, Jenna could focus and look (Figure 12.10). Helena and Jenna sat gazing at each other, enthralled, for several minutes. After a few minutes, her nurse,

Figure 12.10 Jenna gazes at her mother's face

Figure 12.11 Jenna gazes at her father's face in full alert state

who had remained quiet while letting them get to know each other, explained that Jenna could indeed see and even hear and look for her mother's voice and face. She suggested that this be a game they could play. Helena was absolutely amazed and proud at the same time, marveling at her baby's capacity to interact at such a young age (Figure 12.11).

PRETERM INFANT BEHAVIOR ASSESSMENT

Als and associates developed an instrument for assessing the preterm infant. Their assessment of preterm infant behavior (APIB) examines the interplay of behaviors within five behavioral parameters (subsystems): *autonomic*, which refers to physiologically related changes such as pulse, respiration, skin color, and visceral responses; state, or state of consciousness on the continuum of sleep to wakefulness up to robust crying; *motor*, which assesses tone, posture, and movement patterns; *attention/interaction*, or the ability to attend and respond to the environment; and *self-regulatory*, the infants' efforts to cope with their experiences along with their ability to maintain state and self-console.[39,59] These parameters can be used to assess preterm infants' current level of functioning as well as their ability to cope with the physical and social environment. It is important to note that the APIB is a dynamic, interactive examination that is provided by an examiner who is mindful of an infant's breathing/autonomic stability and thus, the infant is facilitated to exhibit best performance. Once an infant's coping ability and organization are assessed, an individualized plan of care that is unique to the infant can be developed.[39,58,59]

The assessment seeks to describe the unique way in which each individual infant interacts, copes with, and integrates experience from the world around him. The interplay of behaviors is evaluated along with the degree of organization rather than maturation of the central nervous system as the focus; in this model, the preterm infant is seen as being in continuous interaction with the environment. The degree of facilitation by the examiner is also integrated into the scoring.

The APIB assessment is intended to identify an infant's current level of balanced and smooth integrated functioning (competence). In other words, identifying in which situations and with what supports the infant appears to function smoothly and is relaxed and comfortable and that enable the caretaker to promote modulated, organized behavior. The purpose of this assessment is to enable the examiner to describe the threshold of disorganization indicated in the infant's behaviors of defense and avoidance. The degree and kind of stress and intensity of frustration or defense that the infant experiences can be indicators of the degree of energy the infant has available. The assessment

will show whether there is any leeway in the infant's responses. For example, can the infant tolerate being turned over to supine from prone (where he is flexed, tucked, and resting calmly) without demonstrating disorganized behaviors such as extended arms and hands, grimaces, and fussiness, or does the infant lose energy and become depleted with vestibular input of changing positions? What is the relationship between the infant's breathing and movement patterns? Does the infant stretch and squirm in an attempt to steady his or her breathing? Does the infant pause in breathing as he stretches and squirms? If the infant cannot display organized behavior when being handled sensitively, the infant may require greater facilitation before, during, and after caregiving interactions. As with the NBAS, training is required to become proficient at administering the APIB examination. However, an awareness of various behavior dynamics will increase the untrained examiner's ability to assess the infant's well-being and provide appropriate interventions. Training in the APIB is available by contacting Heidelise Als, PhD, Neurobehavioral Infant and Child Studies, Children's Hospital, 320 Longwood Avenue, Boston, Massachusetts 02115.

Case Assessment of a Healthy Preterm Infant

Assessing the organizational function of a healthy preterm newborn follows much the same approach as examining the term infant, yet responses may be very different. In the following example, Alejandra represents a healthy, preterm newborn of an appropriate weight for gestational age when the infant was delivered at 33 weeks and who is now 36 weeks postmenstrual age (PMA). The infant is doing well with minimal respiratory support, is breastfed every 2 to 4 hours, and is bedded in a bassinet.

As the examiner begins the evaluation of this infant, she is positioned supine and loosely swaddled to support flexed, midline postures. Her hips and legs are tucked in close. One of the infant's hands is placed on top of her face, and the other arm is extended along the side of her body. Alejandra is breathing regularly and has stable oxygen saturation levels. Her face is pink, and she is in a deep sleep. Her nicely flexed body position, regular breathing, and good color indicate organized sleep. The examination begins by assessing the habituation responses by passing a flashlight across the infant's eyes, which yields an intense startle at the first shine, followed by brief squirming and a return to sleep. The second and third flashes trigger a slightly delayed but less intense response. The fourth flash yields a cycle of activity starting with the startle, and then all arms and legs extend, followed by a brief stoppage and then a recycling of less intense movements. The next flash yields a facial movement only, and then stillness. Alejandra's breathing is regular, but her face is now paled around her eyes and nose. The next two flashes produce no response. Thus, Alejandra had some difficulty inhibiting the motor arousal with one cycle of response, but was able to eventually settle and sustain sleep and shut out the remaining light stimuli. Using a rattle as an auditory sound stimulus, the examiner repeats a similar sequence of 10 trials. The first response shows a brief movement, with subsequent responses subsiding, and eventually Alejandra successfully inhibits her motor response. The second auditory stimulus of a bell produces no response, and she remains asleep. This means that the sequence can end because she has shutdown successfully. These are appropriate habituation responses for a healthy, preterm newborn.

The next part of the assessment evaluates movement, posture, and tone. The examiner begins by placing Alejandra on her back. As she is touched, her color changes across her face, her hands become blue, and her trunk appears mottled. She makes jerky arm and leg movements in all directions; she awakens and squirms. The examiner places her hand on the soles of Alejandra's feet and contains them, allowing the leg movements to slow, which softens the arm movements. As the examiner bends over the infant and talks quietly, her eyes open and the infant also quiets and fixates on the examiner's face.

Now that Alejandra is calmer and is able to use motor inhibition to quiet her state and movement pattern, the examiner touches the sole of the infant's foot and begins a series of reflexive tests. As Alejandra's feet are touched, she once again becomes active with uncontrolled activity of arms, legs, and trunk, resulting in prolonged tremors. The infant also begins breathing irregularly, and her color changes, so that face and body appear very pale then get quite red. She does not fuss or cry, but it is clear that the examiner's tactile stimulation has aroused the motor system beyond the infant's capacity to regain any control on her own. As Alejandra is placed on her abdomen, she squirms and attempts to regain control. By pressing her feet against the bottom wall of the bassinet and putting her hands to her face, the infant is able to inhibit the squirming and motor movements. Her color and breathing once again come under control.

The examiner loosely wraps Alejandra with her arms out of the blankets and lifts her to assess social skills. The examiner places Alejandra on her lap with her face about 12 inches away from infant. Alejandra opens her eyes wide and actively avoids looking at the examiner, turning her gaze away. The examiner speaks softly and now keeps her face unavailable and out of Alejandra's visual field. Alejandra seems to relax, and her hands open and close rhythmically, her face softens, her mouth begins to purse, and eventually the infant turns her head in the direction of the voice. As Alejandra continues to focus, looking and staying alert, her arms, hands, and head lift toward the examiner in a generalized movement of approach. At the height of this approach, she yawns and sneezes and then settles into a drowsy state. Then the examiner presents the rattle to Alejandra and sees a different response. She startles, frowns, and her face becomes pale. The examiner presents the sound again, but more softly, and the infant turns her head and eyes to the left, in the direction of the sound. Alejandra appears more relaxed and with great effort shifts her eyes to search in the direction of the rattle. Even though her movements remain smooth, she becomes paler and frowns again, indicating the cost of this maneuver to her nervous system.

These responses indicate that although Alejandra can process and respond to the stimulus, it is very taxing for her. Being held wrapped in a blanket and being placed on the examiner's lap help her to respond more competently. She shows her ability to be selective, choosing the animate social voice of the examiner to turn to and avoiding the more difficult inanimate auditory stimulus of the rattle. Alejandra's responses give the overall impression of a well-organized, healthy preterm newborn. Alejandra is capable of communicating within a wide range of behaviors in which she can function smoothly without stress to her overall well-being. Her sleep is stable, and her ability to process both visual and auditory stimuli indicates that she can handle stimuli without becoming upset or irritable. With the onset of a stimulus that is too loud, Alejandra's autonomic system is taxed, and she becomes pale, but the assistance of being wrapped and supportiveness on the examiner's lap, along with a slower presentation of the stimulus, affords Alejandra an opportunity to smoothly regulate her motor and autonomic systems. Alejandra is able to avoid overwhelming input with some degree of specificity by rejecting taxing and costly stimulation.

If Alejandra is provided with a supportive environment, several developmental achievements can be predicted for the next 2 to 3 months. First, her intense sensitivity to activity will lessen, and her autonomic nervous system will mature to a degree so that sleeping and wakefulness will become more regular. Alejandra can open her eyes and interact with her caregivers yet is easily taxed and moves past her threshold for stimulation as she closes her eyes and withdraws from the interaction. She demonstrates the capacity for consistent modulation of the ability to self-regulate as seen with her alertness and interactivity when the stimulation is appropriate. The facilitation and timing of the stimulus by the examiner are pivotal to eliciting the infant's best performance. As Alejandra's organization increases, the examiner should see arousal of her motor system

after tactile stimulation lessens. Alejandra has demonstrated that she can modulate her arousal when the examiner provides containment (putting her hands on the infant's feet, wrapping her, and letting her press her feet against a surface). Alejandra has also shown that her movements become smoother when motor arousal is diminished. Additional signs of motor organization include the ability to bring fingers to mouth and to fold hands under chin and keep them together. The infant is also demonstrating the ability to make a transition from one state to another without becoming upset. Figures 12.12 through 12.18 show when Alejandra is in an organized state and a disorganized state, with some instances of infant cues.

Case Assessments of Immature Preterm Infants

Some infants can show a range of immature or highly sensitive responses to interactions and environmental stimuli. At one extreme of the continuum of preterm functioning is Matt, a hyperreactive infant who reacts to all stimuli, lacking any ability to shut them out and protect himself. Matt appears to be "at the mercy of the stimuli," and his autonomic system pays a severe cost in terms of stress.[60,61] He overreacts to sound or to being touched with a series of startles and limb extensions, followed by uncontrollable flailing, arching, and squirming. Soon Matt is in a challenging cycle that goes on until he either stops breathing or drops his oxygen saturation level or heart rate. This is costly to him because it uses energy and delays his ability to consolidate the regulation of his motor and state systems.[57,61,62] Matt's high reactivity interferes with his growth and may lead to failure to thrive.

Unlike Matt, the hyperreactive preterm infant Lucas is a lethargic, withdrawn, or depleted infant who lies still and does not respond to stimuli.[63,66,67] This preterm newborn appears to want to preserve his fragile autonomic regulation. Because Lucas is past his thresholds and is depleted, he has little energy for interaction. His behavioral responses will be subtle and difficult to interpret as you attempt to interact with him. The challenge for Lucas is that this pattern of withdrawal does not facilitate his ability to develop or activate new pathways to take in new information. Therefore, he develops a rigid pattern of minimal range of behavioral

Figure 12.12 Containment helps the preterm infant to stay organized

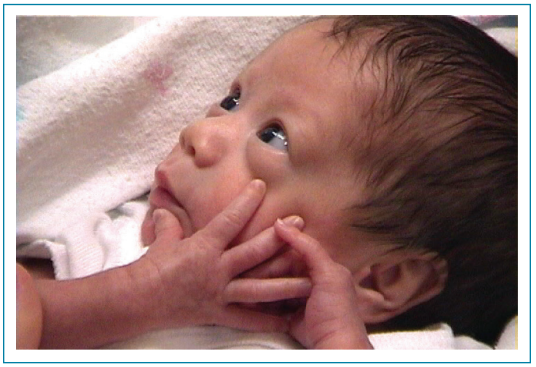

Figure 12.13 Alert state

Figure 12.14 Organized preterm infant

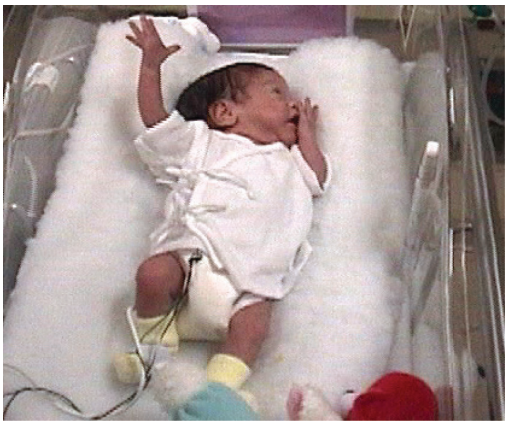

Figure 12.15 Disorganized preterm infant

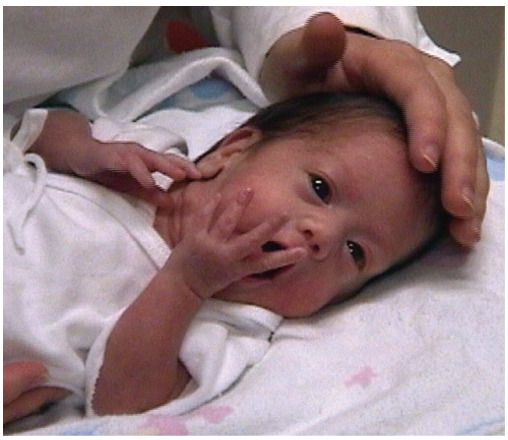

Figure 12.16 Disorganized preterm infant

responses. Autonomic behaviors, such as heart rate and oxygen saturation levels, will often fluctuate. This infant's presentation is very concerning, and his developmental trajectory is guarded.

Some premature infants exhibit a combination of these two types of response patterns, demonstrating an overreactive range of sensitivity at times and later becoming unavailable if overwhelmed by the constant sensory stimuli within the environment. In some disorganized infants, the pattern of protest takes the form of resistant squirms, turning away, pushing away, and avoidance behaviors while being held. These behaviors can be channeled into a more modulated level of interaction if the caregiver slowly introduces caregiving activities, allowing the infant time to ease into them.[68] For example, the caregiver can pause to allow the infant the opportunity to steady his or her breathing or regain energy during an interaction.

An appropriate behavioral assessment describes the infant's abilities to utilize intervention support and delineates the types of facilitation necessary to ensure smooth functioning. As the infant is supported to actively participate in the interaction despite a taxing sensory environment, the caregiver can articulate the infant's neurobehavioral agenda and emerging competence. Recognizing the infant's efforts to cope with stress and regain modulation leads to the infant's evolving behavioral agenda.[33,69]

The ability to understand and reveal the infant's full range of responses becomes essential when parents of a newborn infant begin to read behaviors as meaningful

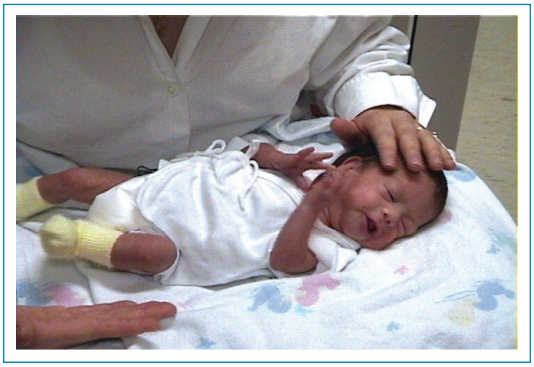

Figure 12.17 Disorganized preterm infant unable to cope with outside stimulation

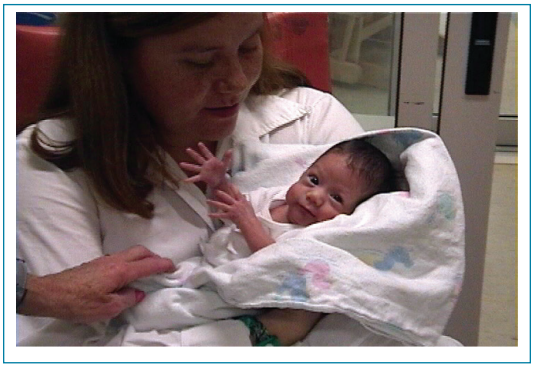

Figure 12.18 Preterm infant signals "time-out"

communications. The nurse's role is to help parents become attuned to the infant's responses and to facilitate their developing relationship. If parents can interpret the behaviors and understand the infant's messages, they can use strategies, such as reciprocity, to enhance the infant's competence. When parents and caregivers can support the infant based on behavioral cues, the infant is more successful and competent and can improve overall level of functioning while caregivers continue to develop their understanding of the infant's unique individuality as further maturation develops.

OTHER EMERGING INFANT ASSESSMENT TOOLS

There are several other infant assessment tools available. One recent systematic review completed by Noble and Boyd found 27 different infant assessment instruments available in the literature for use in the newborn period (up to 4 months corrected age).[70] However, they chose only eight assessment instruments to include in their review; those that had good reliability in prediction, discrimination, and evaluation of change.[68] The Brazelton and APIB as described earlier were among those chosen. Other instruments found to be of high quality included the Test of Infant Motor Performance (TIMP),[71] the NICU Network Neurobehavioral Scale (NNNS),[31] Prechtl's Assessment of General Movements (GMs),[72] Neurobehavioral Assessment of the Preterm Infant (NAPI),[73] Dubowitz neurologic assessment of the preterm and full-term infant (Dubowitz),[74] and Neuromotor Behavioral Assessment (NMBA).[75] The GMs and Dubowitz are well known and described as primarily neurologic system assessments rather than behavioral assessments and as such are discussed elsewhere in this text. Each of the others is appraised in the following with some of the unique examination characteristics, psychometric properties, and clinical utility that might be of interest to those considering use of these assessments. These descriptions are in no way exhaustive but are to be considered an introduction to the different options available for discriminating infant behaviors.

Each of the instruments chosen in the review by Noble and Boyd were found to have adequate content validity[70]; however, the TIMP created by Campbell was found to be suitable to be used as an outcome measure. This could be important if the instrument is being used to demonstrate effectiveness of previously delivered interventions. The TIMP is a standardized instrument for assessing neuromotor development in infants between 34 weeks PMA and 4 months postterm.[76] The TIMP includes 13 observation and 28 elicited items. The TIMP is an age-sensitive measure of motor performance that can differentiate for high and low risk of poor infant motor outcomes and has excellent psychometric properties.[74] Reliable administration of the TIMP requires extensive training. Primarily, physical or occupational therapists become reliable in the scoring, when used for prediction or outcome measures. The NAPI has some similarities to the TIMP; however, it was developed only for use with preterm infants, whereas the TIMP can be used across infant groups. This makes the TIMP useful for comparison across populations.[76] The NAPI was designed to measure the progression of neurobehavioral performance in the preterm infant 32 weeks PMA to term.[71] Training to administer the NAPI is fairly extensive but not as formalized as with some of the other assessment tools. The NMBA has less utility since its scoring is only reliable from 30 to 36 weeks PMA and it was developed to identify preterm infants at risk for developmental delays. It is interesting to note that no formal training is required for the NMBA as opposed to the extensive training required for other instruments. Only the TIMP and the NAPI have published preterm norms, and these two instruments may be better suited for clinical utility than other well-designed infant assessments available.

There has been a surge in the literature over the past few years, addressing the many components and complexities of infant neurobehavioral assessments, especially with the NNNS. This assessment has been utilized in research as well as within

clinical settings and was designed to measure the infant's biobehavioral organizations as a valid biomarker for detecting at-risk infants, with the ability to predict developmental outcomes.[23,77,78] The NNNS examines both neurologic integrity as well as behavioral functioning and scores a full range of infant neurobehavioral performance that was intended to have broad applicability.[23] The standardized nature of the NNNS does not embody optimal performance criteria as a foundational principle and uses the integrity of the infant's innate organization to identify neurobehavioral patterns as an objective format. During the NNNS, the order of the items administered is more structured, and the emphasis to bring out the "best" in the infant is less apparent. The administration of the NNNS is completed in about 20 to 30 minutes and is scored easily by an experienced examiner. The examination is primarily state dependent and follows a relatively invariant sequence. This does not mean that the examiner disregards the infant's behavior and simply completes the maneuvers at the infant's detriment. The examiner is expected to read the infant's states and behaviors to develop a rapport with the infant that ensures the infant's active participation. Measurable antecedents of later developmental outcomes can be detected with neonatal neurobehavior using the NNNS.[78–80] The NNNS assessment is a valid, predictive biomarker for articulating high-risk infant neurobehavioral functioning. The infants neurobehavioral patterns revealed by the NNNS, particularly presence of increased excitability, arousal, hypertonicity, and stress behaviors associated with lower quality of movement scores, orientation and self-regulation, have been shown to be predictive of adverse outcomes.[78] The NNNS can be used with high-risk infants 34 to 44 weeks PMA and is easily administered in the NICU. It is imperative to acknowledge the concept of behavioral regulation, which is considered to be reflective of the infant's level of higher functioning and may be missed with routine neurological examinations.[79] The NNNS is a valuable tool available to nurses and other healthcare professionals who work with preterm infants to provide a strategy for obtaining accurate information about the infant's strengths and vulnerabilities.[79,80] Ideally, the advanced practice nurse or midlevel practitioner completes a behavioral assessment prior to the infant's hospital discharge and incorporates the findings into the infant's medical discharge exam to identify infants most at risk for developmental delays to facilitate appropriate early-intervention services.

SUMMARY

An infant's behavioral patterns may be subtle and difficult to elicit and interpret, yet observing behavior is an integral part of a comprehensive newborn examination. The behavioral assessment allows the examiner an opportunity to evaluate aspects of the infant's neurologic status, and helps establish guidelines for an individualized developmental plan of care for both term and preterm infants. Behavioral assessments can also be used to encourage parents to identify and respond to their newborn's behavioral cues and signals. Behavioral assessment provides an opportunity to consider the infant as a whole individual—an approach that enhances infant competencies and supports the infant–parent relationship.

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Assessment of the Dysmorphic Infant

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Most infants are born healthy, and the first clinical assessment usually reveals no physical abnormality. However, birth defects remain one of the leading causes of infant mortality in the United States. Every 4½ minutes a baby in the United States is born with a birth defect, and as of 2013, one in five infant deaths was attributed to a congenital abnormality.[1]

About 15% of all newborn babies will have at least one minor malformation (e.g., one that does not inferfere with normal functioning), and these often go unnoticed. However, their presence should prompt the practitioner to look for a major malformation, as newborns with one or more minor malformations are more likely to have a major malformation (e.g., causes dysfunction or requires surgical correction). If there is one minor malformation, the incidence of an associated major anomaly is 3%, this increases to 11% in the presence of two minor malformations and to as high as 90% with the presence of three minor anomalies.[2] Fifty percent to 60% of human congenital anomalies are of unknown etiology, and approximately one third are caused by genetic factors. A smaller percentage of birth defects are the result of environmental agents, such as viruses and drugs.[3] Nearly 4,000 malformation syndromes have now been delineated using standard karyotype as well as newer genomic technologies, including microarray and whole genome sequencing.[2,4]

The identification of dysmorphic features during the initial physical examination is a crucial first step in the continuum of care for affected infants. A thorough, systematic approach by a skilled examiner can yield important findings and direct the healthcare team in providing timely and appropriate care for the infant, as well as resources for parents.

MATERNAL AND FAMILY HISTORIES

A complete maternal medical, gynecologic, and obstetric history should be constructed when evaluating the dysmorphic infant. A history of adverse pregnancy outcomes, including multiple miscarriages and stillbirths, can be an important risk factor. Maternal age should be documented because chromosomal anomalies, such as trisomy 21, occur more frequently with advancing maternal age. Pregnancies complicated by medical conditions, such as diabetes mellitus or hypertension, increase the possibility for fetal physical deformities. Prenatal exposure to teratogens, including medications, infections, chemicals, and illicit drugs, must be documented because certain agents cause specific structural abnormalities and functional diseases to be exhibited in the fetus. Critical periods of fetal development, dosage and duration of exposure to the teratogen, and genotype of the embryo must also be taken into consideration.[3] Results of prenatal testing, including multiple marker serum screening, maternal serum alpha-fetoprotein testing, chorionic villus sampling, and amniocentesis, should be recorded to identify an increased risk or a confirmed diagnosis of fetal disorders such as neural tube defects and trisomies. In addition, in 2012, noninvasive blood testing, the MaterniT21 PLUS test, was made available to detect increased

amounts of chromosomal 13, 18, and 21 material as well as an abnormal number of X or Y chromosomes circulating in a pregnant woman's blood.[5]

A comprehensive maternal and paternal family history is also helpful. A number of congenital anomalies and medical conditions can be inherited and therefore place an infant at risk for developing the disorder. Some of these conditions include spina bifida; hydrocephalus; muscular dystrophy; cleft lip; cleft palate; congenital heart defects; polydactyly; clubfoot; congenital hip dislocation; deafness; blindness; childhood cataracts; cystic fibrosis; dwarfism; polycystic kidney disease; and stomach, bowel, or kidney defects.

There are also genetic disorders that occur more commonly within particular ethnic groups. Descendants of Ashkenazi Jewish or French Canadian ancestors may have an increased risk for Tay–Sachs disease, an often fatal disorder marked by degeneration of brain tissue and the maculae of the retinas. Infants of African American ancestry are at an increased risk for inheriting sickle cell disease, a serious condition of red blood cells that are distorted in shape and have a tendency to clump together and occlude blood vessels. Thalassemia, a group of hemolytic anemias, is more prevalent in Mediterranean and Asian populations.

If one or both parents are of Jewish, French Canadian, African American, Mediterranean, or Asian descent or if a medical condition occurs repeatedly in one of the partner's families, the couple may consider genetic testing prior to conceiving a child. If either partner is a carrier for a specific inheritable condition, the significance of the results can then be discussed with the couple's healthcare provider.

PHYSICAL EXAMINATION

The newborn infant should have a thorough physical examination within 24 hours of birth. This first examination may reveal more abnormalities than any subsequent routine examination done. First, the family, maternal, pregnancy, and perinatal histories are reviewed. The examination is performed in an area that is warm and quiet with good lighting. A systematic approach should be used. Although the exact examination sequence is not important, a consistent approach ensures that all aspects are evaluated. Assessment of gestational age should be included. Knowledge of gestational age can be important in the interpretation of physical findings, especially in infants who are noted to have intrauterine growth restriction (IUGR). Every body part should be examined with particular emphasis on size, shape, color, position, spacing, and symmetry.

Appearance and Posture

An inspection is made for deformations and obvious malformations. An abnormal facial appearance or other abnormalities in appearance can indicate the presence of a syndrome. The newborn's posture at rest usually reflects intrauterine position, sometimes called the *position of comfort*.

Skin

The skin is inspected for abnormalities. Areas of abnormal pigmentation, congenital nevi, hemangiomas, macular stains, or other unusual lesions should be noted.

Head

The shape and size of the head are inspected. The presence of abnormal hair, lacerations, abrasions or contusions, scalp defects, unusual lesions, or protuberances should be noted. An asymmetric skull that persists for longer than 2 to 3 days after birth or a palpable ridge along a suture line is abnormal and suggests craniosynostosis. Although occurring in normal infants, craniotabes can be a pathologic finding with syphilis and rickets.

A large anterior fontanel may be associated with congenital hypothyroidism, achondroplasia, hypophosphatasia, chromosomal

abnormalities such as Down syndrome, and with IUGR.[6]

Neck

The neck is assessed for masses, decreased mobility, and abnormalities. Cystic hygroma, the most common lymphatic malformation in newborns, typically presents as a painless mass superior to the clavicle that transilluminates. Redundant skin in the neck may be a feature of some genetic syndromes. Examples include Turner syndrome, in which the neck appears webbed due to redundant skin along the posterolateral line, and Down syndrome, with excess skin posteriorly at the base of the neck.

Face

The face is examined for symmetry. Facial palsies and asymmetric crying facies are most obvious when the newborn is crying and may go unnoticed in the sleeping or quiet infant. Asymmetric crying facies are the result of hypoplasia or congenital absence of the depressor anguli muscle. Only the muscles controlling movement of one side of the mouth are affected, causing asymmetry of the face with crying. However, the muscles controlling movement of the upper face are normal; so when the infant cries, the forehead wrinkles and both eyes close normally. Asymmetric crying facies have been associated with other anomalies, particularly those of the cardiovascular system.[7] Facial palsy may also be secondary to nerve compression during delivery, which can occur as a result of forceps-assisted delivery.

Eyes

If spacing appears abnormal, the distance between eyes can be measured and compared to standard values (see Figure 5.15). This part of the examination is especially important if other dysmorphic features that suggest a syndrome are present. The presence of epicanthal folds is rarely a normal finding and usually suggests a syndrome (trisomy 21). The sclerae should be clear and white. If the sclerae appear deep blue, osteogenesis imperfecta should be considered. Glaucoma is manifested by a large cloudy cornea. Defects in the iris, such as coloboma, should be noted. Cataracts or retinoblastomas will present as a white pupil when the red reflex is assessed.

Ears

The ears are in normal position when the helix is intersected by a horizontal line drawn from the outer canthus of the eye perpendicular to the vertical axis of the head (Figure 13.1).[8] If the helix falls below this line, the ears are low set. An ear is posteriorly rotated if its vertical axis deviates more than 10 degrees from the vertical axis of the head. Malformations of the external ear are often associated with syndromes of multiple congenital anomalies that include renal malformations. The abnormalities may also indicate additional anomalies of the middle and inner ear associated with hearing loss.

Nose and Mouth

A depressed nasal bridge or an extremely thin or unusually broad nose may occur in some malformation syndromes. Clefts of the soft or hard palate are visible to inspection.

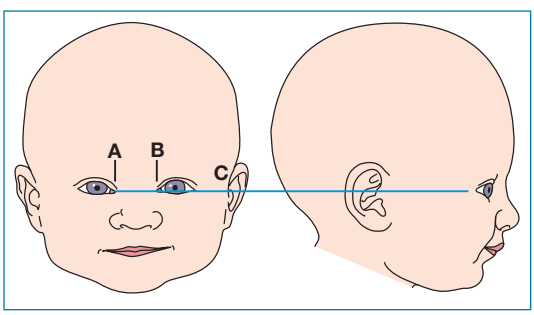

Figure 13.1 Ear placement

Source: From Halderman-Englert CR, Saitta SC, and Zackai EH. 2012. Evaluation of the dysmorphic infant. In *Avery's Diseases of the Newborn*, 9th ed., Gleason CA, and Devaskar SU, eds. Philadelphia, PA: Saunders, 189. Reprinted by permission.[9]

Palpation may be needed to detect a submucosal cleft. Macroglossia, or enlargement of the tongue, can be seen with Beckwith–Wiedemann syndrome.

Chest

The chest is examined for size, symmetry, and structure. A malformed or small thorax may be the result of pulmonary hypoplasia or neuromuscular disorders. Pectus excavatum or pectus carinatum may occur as isolated findings or as part of congenital syndromes. Breast size and position should be noted, because widely spaced nipples occur with some genetic syndromes.

Abdomen

Asymmetry caused by congenital anomalies or masses may first be appreciated by observation. Abnormal, absent, or misplaced kidneys are assessed by using deep palpation (see Figures 9.2 and 9.3). Most abdominal masses in newborns are enlarged kidneys caused by hydronephrosis or cystic renal disease.[6] A single umbilical artery is present in 0.3% of neonates, occurring more frequently in small-for-gestational-age (SGA) infants, premature infants, and twins.[10] Approximately 40% of infants with a single umbilical artery have other major congenital anomalies, predominantly involving the genitourinary system, and have significant mortality. Bourke and colleagues found that in an otherwise normal infant, a single umbilical artery is associated with asymptomatic renal abnormalities in 7% of cases.[11–13]

Genitalia

The genitalia are inspected immediately after birth to identify the infant's gender.

Females

The labia minora should be separated to detect whether the hymen, which normally has some opening, is imperforate. Enlargement of the uterus resulting from an imperforate hymen may be detected as a low midline abdominal mass.

Males

Hypospadias, ventral location of the meatus on the penis, is relatively common. The meatus may be located anywhere from the proximal glans to the perineum, with more severe cases having a more proximal meatus. Infants with perineal or scrotal hypospadias and those with hypospadias of any location accompanied by nonpalpable testes should be evaluated for intersex conditions, including congenital adrenal hyperplasia. Epispadias, dorsal location of the meatus, is uncommon and usually associated with bladder exstrophy.

Ambiguous Genitalia

Signs of ambiguous genitalia include an enlarged clitoris, fused labial folds, and palpable gonads in a phenotypic female and bifid scrotum, severe hypospadias, micropenis, and cryptorchidism (undescended testes; Figure 13.2) in a phenotypic male. These conditions may be caused by abnormalities of sexual differentiation or congenital adrenal hyperplasia. Infants should be evaluated promptly and the appropriate gender assigned as soon as possible.

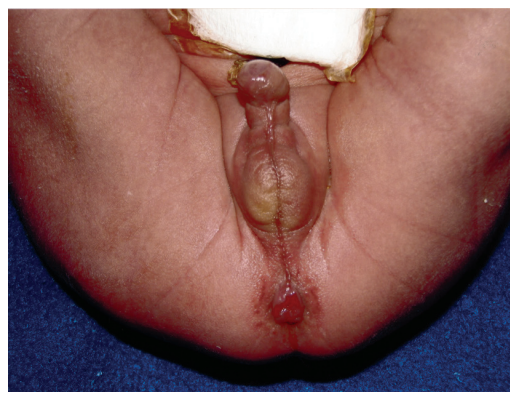

Figure 13.2 Cryptorchidism

Courtesy of Presbyterian/St. Luke's Medical Center, Denver, CO.

Anus

The anus and rectum should be checked carefully for patency, position, and size. On occasion, large fistulas are mistaken for a normal anus, but if one checks carefully, it will be noted that a fistula will be either anterior or posterior to the usual location of a normal anus.[6]

Extremities

The extremities are examined for deformities and movement. The hands and feet are inspected for syndactyly (fusion of digits) and polydactyly (extra digits; Figures 13.3 and 13.4). Syndactyly and polydactyly can be normal variants in a newborn with an otherwise normal examination, may be associated with a strong family history, or may be associated with various syndromes. The presence of a single palmar crease, or simian crease, should be noted. A single palmar crease occurs in 5% to 10% of the normal population and is common in newborns with trisomy 21. Talipes equinovarus (clubfoot; Figure 13.5) is more common in males. The foot is turned downward and inward, and the sole is directed medially. If position can be corrected with gentle force, it will resolve spontaneously. If not, orthopedic treatment and follow-up are necessary. The hips should be examined to detect developmental dysplasia of the hip.[6]

Trunk and Spine

A tuft of hair, discoloration, or hemangioma in the sacrococcygeal area may suggest an underlying vertebral anomaly. Soft-tissue masses along the spine that are covered with normal skin may be lipomas or myelomeningoceles. A dimple without a visible base may indicate the presence of a pilonidal sinus or tract to the spinal cord.

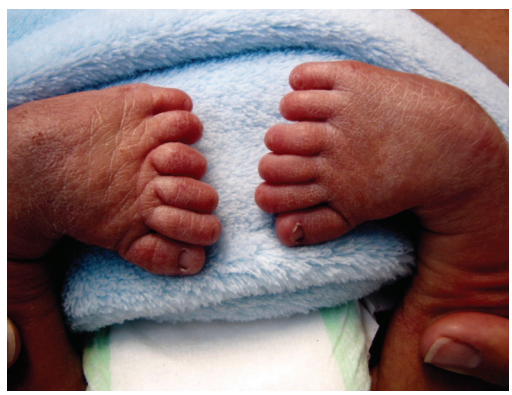

Figure 13.4 Polydactyly of the toes

Courtesy of Presbyterian/St. Luke's Medical Center, Denver, CO.

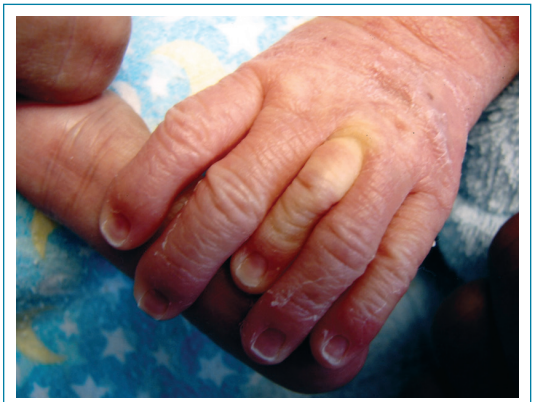

Figure 13.3 Polydactyly of the fingers

Courtesy of Presbyterian/St. Luke's Medical Center, Denver, CO.

Figure 13.5 Clubfoot

In infants with one or more external malformations, a search should also be made for internal malformations, with surveillance that includes an echocardiogram, a renal ultrasound, and a cranial ultrasound. Chromosomal analysis should be completed in infants who have more than one major, multiple minor anomalies or dysmorphic features.

SHARING FINDINGS WITH PARENTS

When an anomaly is identified on physical examination, the infant should be shown to the parents as soon as possible. The physical finding may have been identified antenatally by ultrasound or may not have been expected. Either way, a spectrum of emotional responses from the parents is to be anticipated. Common responses of parents include guilt, intense grief, anger, denial, frustration, and a sense of isolation. It is important to be sensitive to what the parents may be feeling. The defect should be shown to the parents and a factual description given, avoiding opinions or guesses. Genetic counseling should be provided to help parents answer any questions regarding the prognosis for the child and genetic risks for future pregnancies. Medical geneticists and genetic counselors have extensive knowledge of genetic disorders and congenital anomalies and are trained to provide families with psychological and emotional support. However, even a healthcare professional with a basic knowledge of genetics and Mendelian inheritance can be helpful when discussing the physical findings with the parents and can provide answers to general questions.

PROBLEMS IN MORPHOGENESIS

Dysmorphisms are one or more anomalous external physical features that can be indicators of the underlying cause or developmental defect.[4] A congenital anomaly is a structural defect, present at birth, a deviation from normal. Every structural defect represents an inborn error in morphogenesis (dysmorphogenesis). Minor anomalies are unusual morphologic features that have no serious medical or cosmetic consequences to the patient. Almost any minor defect may occasionally be found as an unusual feature in a particular family. Minor external anomalies are most common in areas of complex and variable features, such as the face, ears, hands, and feet. Clinical diagnosis cannot usually be made based on a single defect. A specific diagnosis most often depends on recognition of an overall pattern of anomalies. Single minor anomalies are present in about 15% to 20% of newborns; 90% of infants with three or more minor anomalies also have one or more major anomaly, requiring significant surgical or cosmetic intervention.[3] Therefore, recognition of both minor and major anomalies is equally important.

Based on the developmental process involved in their formation, patterns of anomalies can be classified into four categories: malformation, deformation, disruption, and dysplasia. A malformation results from an abnormal morphogenesis (dysmorphogenesis) of the underlying tissue, which is attributed to a genetic or teratogenic factor. Malformations arise from intrinsic defects in genes that specify a series of developmental steps.[4] Often, but not always, a malformation in one part of the body is associated with malformations elsewhere. Examples of malformations are congenital heart defects or neural tube defects. Malformations occur in all gradations, the manifestations ranging from nearly normal to more severe, and have a recurrence risk of 1% to 5%.[14]

Deformation is an alteration in form, shape, and/or position of a normally formed body part by biomechanical forces that distort the normally developing structure.[4,6] It usually occurs in the fetal period, not in embryogenesis, and is a secondary defect. Congenital hip dislocation and clubfoot are examples of deformations that can be caused by intrauterine constraint. Most deformations apparent at birth either resolve spontaneously or can be treated using external fixation devices to correctly position the affected part. Most deformations have a very good prognosis with a very low recurrence risk.

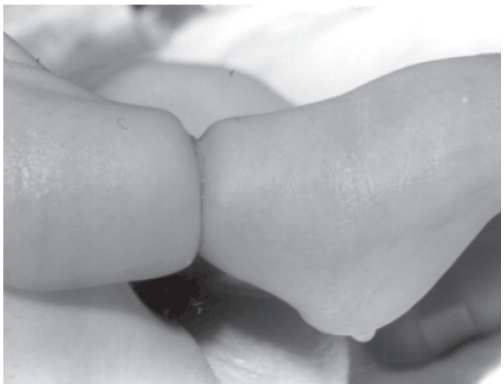

Figure 13.6 Constriction defect from amniotic band

Courtesy of J. Hernandez, MD, The Children's Hospital, Denver, CO.

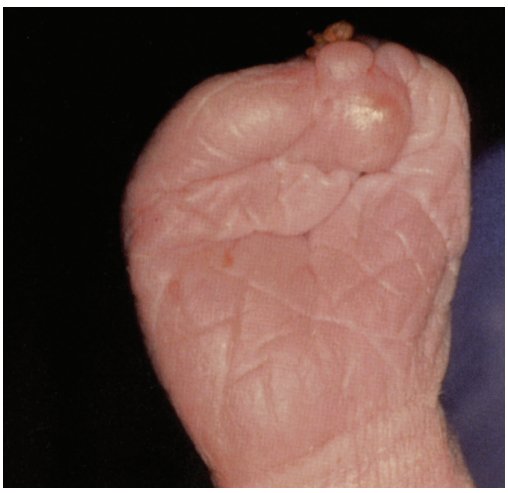

Figure 13.7 Amniotic bands resulting in finger amputation

Source: From Clark DA. 2000. Atlas of Neonatology. Philadelphia, PA: Saunders, 23. Reprinted by permission.[15]

Disruptions result from an extrinsic insult or destruction of originally normal fetal tissue.[4,6] They are a secondary malformation. Usually, a body part rather than a specific organ is affected. Such disruptions may be vascular, infectious, or mechanical in origin. One example of this is disruption of normally developing tissues by amniotic bands (Figures 13.6 and 13.7).[4] Disruptions are more difficult to treat than deformations because they involve actual loss of normal tissue. Disruptions are generally sporadic with a low recurrence risk.

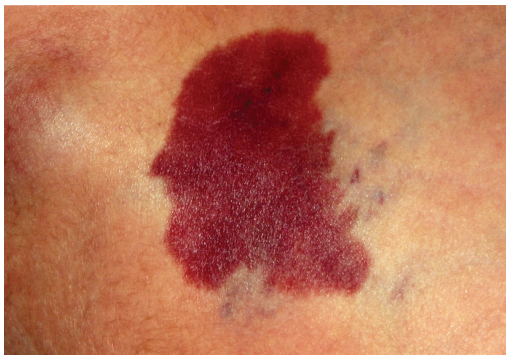

Figure 13.8 Hemangioma

Dysplasia is a primary defect involving abnormal organization or differentiation of cells into tissue that results in clinically apparent structural changes.[4,6] This can be localized, for example, a hemangioma (Figure 13.8), or generalized, such as achondroplasia (dysplasia of skeletal tissue). Dysplasias are usually not correctable, and the affected individual experiences the clinical effects of the underlying cell or tissue abnormality for life.[9] Malformations and dysplasias are primary events in embryogenesis; disruptions and deformations occur secondarily. The concepts of malformation, deformation, disruption, and dysplasia are useful clinically to assist in recognition, diagnosis, and treatment of congenital anomalies. However, given the constellation of congenital anomalies, a neonate may present with combinations of these patterns of anomalies. The occurrence of congenital anomalies can further be divided into several categories: syndromes, sequences, associations, and teratogens.

GENETICS

The nucleus of the human cell contains chromosomes, structures that include DNA and transmit genetic information during cell division and human development. Each human being has 46 chromosomes—22 pairs of

autosomes and a pair of sex chromosomes (XX or XY) that determine gender. The chromosomes contain genes, the biologic units of inheritance. Genes control the physical, biochemical, and physiologic traits passed along to children from their parents. Genetic abnormalities are divided into three categories. Those that[14]

- Influence gene dosage (chromosomal abnormalities such as trisomies)
- Involve mutations in the genes themselves (over 6,000 rare single-gene disorders)
- Create a vulnerability to developmental errors that are then influenced by environmental factors (multifactorial inheritance disorders such as isolated malformations or schizophrenia)

The gene mutations that cause greater than 6,000 individually rare disorders can be further classified into four categories: autosomal dominant, autosomal recessive, X-linked, and mitochondrial mutations. Each individual receives two sets of chromosomes, one from each parent. Each pair of chromosomes contains a pair of genes, or alleles, that normally work together. A mutant gene is one that has altered in such a way that it can produce an abnormal trait.

Diseases caused by autosomal dominant genes are rare. A single mutant gene is dominant if it masks the effect of its paired gene and causes an obvious abnormality. The risk of the single mutant gene being passed on is 50%, but autosomal dominant disorders have a wide range of expression and will present in varying degrees between affected individuals due to influences of the normal paired gene as well as the genetic and environmental background of the individual. Examples of autosomal dominant disorders are retinoblastoma and neurofibromatosis.

Autosomal recessive disorders are also rare, although the number of carriers for these diseases can be high. These disorders are inherited from normal parents who both have the same recessive mutant gene. In most cases, both parents of an affected individual are heterozygous carriers of the disease. Typically, one fourth of their offspring will be normal heterozygotes, one half will be normal carrier heterozygotes, and one fourth will be homozygotes who have the disease. An example of an autosomal recessive disease is cystic fibrosis.[16]

Genes located on the sex chromosomes cause X-linked disorders. The Y chromosome does not appear to carry any disease-causing genes. X-linked dominant traits are rare, but X-linked recessive diseases occur more commonly. A single copy of a mutant gene on the X chromosome will be expressed in the male because he has no normal partner gene. His daughters will all be carriers because they will receive his X gene, and his sons will all be normal because they receive his Y gene. Because females receive an X chromosome from each parent, they can be homozygous normal, homozygous for the X-linked disease, or heterozygous. Fifty percent of male offspring of X-linked recessive women will be affected, and 50% of her daughters will be carriers. Examples of X-linked disorders are Turner syndrome and Klinefelter syndrome.[17]

Mitochondrial mutation disorders result from insufficient energy production in critical tissues. Most of these disorders present after the child is born, usually with visual loss, seizures, encephalopathy, progressive myopathy, or diabetes. The human egg is the source of mitochondria for all offspring and is therefore inherited only from the mother. Males with disorders caused by mitochondrial mutations have no risk of passing along the disorder to their offspring. Females, however, have a risk that approaches 100%. Female offspring of affected women will inherit some abnormal mitochondria, but may not manifest the disease.[14]

SYNDROMES

A syndrome is a collection of anomalies involving more than one developmental region or organ system or a pattern of multiple anomalies thought to be pathogenetically related.[9] Chromosomal syndromes are the

malformation syndromes usually diagnosed in the neonatal period. The most common of these are trisomy 21, trisomy 18, trisomy 13, and 45,X. With the advent of the human genome project, more information is now available regarding chromosome structure. Once thought to be associations, DiGeorge and Beckwith–Wiedemann have now been found to have chromosomal abnormalities as an underlying etiology and are more correctly categorized as syndromes.[18]

Trisomy 21 (Down Syndrome)

The incidence of trisomy 21 is one per 650 to one per 1,000 live births, making it the most common pattern of malformation in man.[14] Down syndrome can usually be diagnosed at birth or soon after by its dysmorphic features, which produce a distinctive phenotype. Principal features include hypotonia, poor or absent Moro reflex, hyperextensibility of joints, excess skin at the nape of the neck, flat facial profile (Figure 13.9), low-set ears, slanted palpebral fissures, and single transverse palmar (simian) creases (Figure 13.10). Associated anomalies include congenital heart defects (30%–40%); increased incidence of duodenal atresia, esophageal atresia, and imperforate anus; and significant hearing loss (90%). Most of the features of trisomy 21 can occur as isolated features in normal infants. It is the combination of features forming a recognizable pattern that permits early diagnosis.[19]

Trisomy 18 (Edwards Syndrome)

The incidence of trisomy 18 is approximately one per 5,000 to one per 7,000 live births.[17] There is a 4:1 preponderance of females to males. The Edwards syndrome phenotype is as distinct as Down syndrome, but because it is less common, it is less likely to be recognized clinically. Trisomy 18 syndrome (Figure 13.11) is highly lethal, with 50% mortality within the first several weeks of life. Only 5% of affected infants will survive the first year, and they will have severe mental deficiencies.[17] Physical findings include prenatal and postnatal growth deficiency, micrognathia, overlapping digits, complex congenital heart disease, low-set

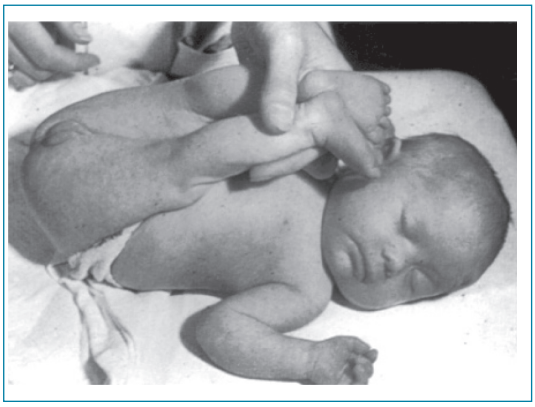

Figure 13.9 Trisomy 21 (Down syndrome)

Courtesy of J. Hernandez, MD, The Children's Hospital, Denver, CO.

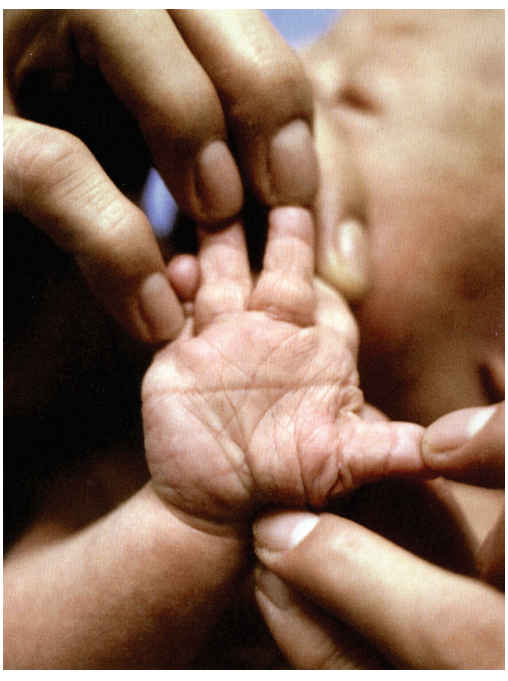

Figure 13.10 Single palmar crease

Source: From Clark DA. 2000. Atlas of Neonatology. Philadelphia: Saunders, 31. Reprinted by permission.[15]

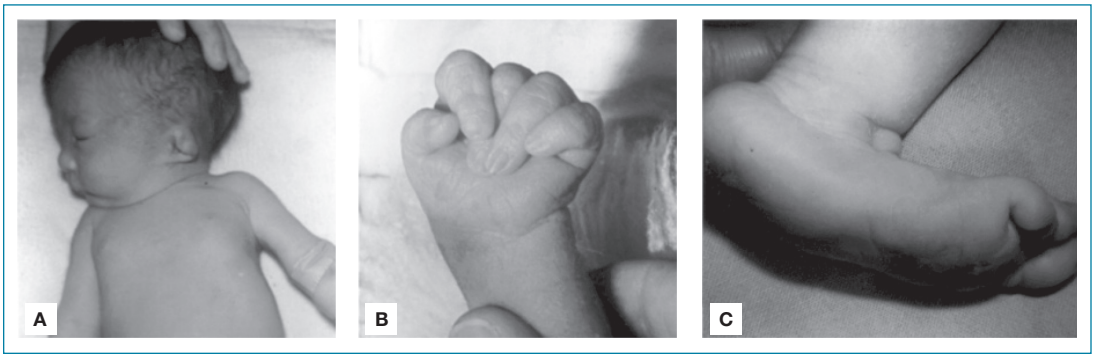

Figure 13.11 Trisomy 18 (**A**) Prominent occiput; short sternum; micrognathia; malformed, low-set ears. (**B**) Overlapping fingers. (**C**) Rocker-bottom feet.

Courtesy of J. Hernandez, MD, The Children's Hospital, Denver, Colorado.

ears, rocker-bottom clubfeet, and generalized hypertonicity. Associated anomalies include tracheoesophageal fistula or esophageal atresia, hemivertebrae, omphalocele, myelomeningocele, and radial dysplasia.[17]

Trisomy 13 (Patau Syndrome)

The incidence of trisomy 13 is approximately one per 10,000 live births.[14] Trisomy 13 (Figure 13.12) is highly lethal with a mean life expectancy of 130 days.[6] This malformation pattern is quite distinguishable and clinically recognizable. Physical findings include ora-facial clefts, microphthalmia or absence of the eyes, low-set ears, rocker-bottom feet, moderate microcephaly, polydactyly, scalp cutis aplasia, and congenital heart disease. Associated anomalies include cleft lip and palate, cystic kidneys, holoprosencephaly, and other severe central nervous system (CNS) malformations. The identification of multiple midline defects is a way to recognize trisomy 13.[14]

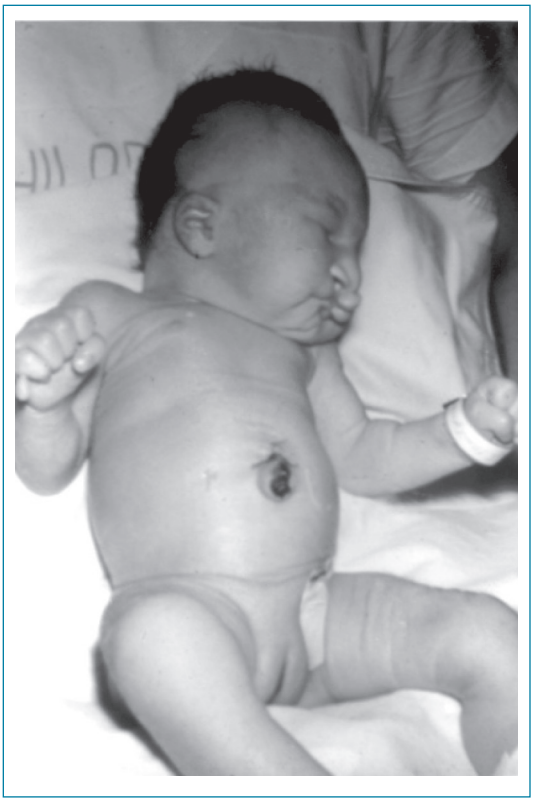

Figure 13.12 Trisomy 13

Courtesy of J. Hernandez, MD, The Children's Hospital, Denver, CO.

45,X (Turner Syndrome)

The incidence of monosomy X or Turner syndrome is approximately one in 2,500 live-born females.[14] Ninety-five percent of conceptions are miscarried or stillborn. The 45,X syndrome (Figure 13.13) is usually compatible with survival if the fetus reaches term gestation. Females with Turner syndrome can often be identified at birth or before puberty

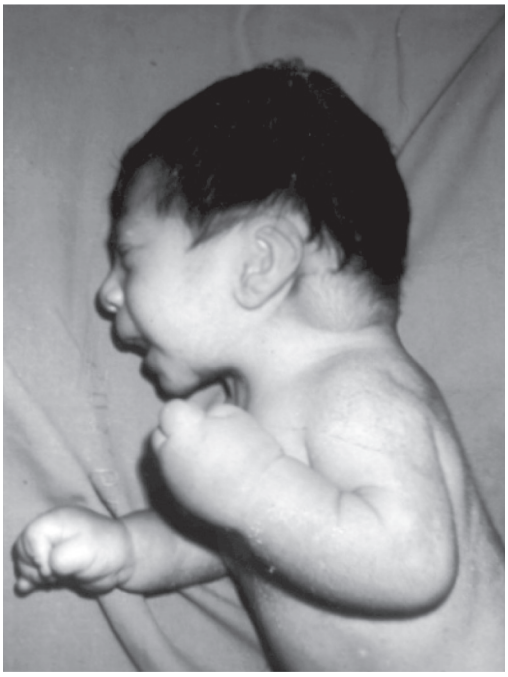

Figure 13.13 Turner syndrome Lymphedema (hands), webbed neck, low posterior hair line, low-set ears

Courtesy of J. Hernandez, MD, The Children's Hospital, Denver, CO.

by their distinctive phenotypic characteristics. Physical findings include small stature, short webbed neck, lymphedema of the hands and feet, frontal prominence, low posterior hairline, and broad chest with widely spaced nipples. Associated anomalies include congenital heart defects, structural kidney defects, and gonadal dysgenesis.[14]

DiGeorge Syndrome

DiGeorge syndrome is a chromosomal deletion of 22q11.2 characterized by structural or functional defects of the thymus, conotruncal heart defects, hypoparathyroidism, and secondary hypocalcemia.[17] DiGeorge syndrome is detected in approximately one per 5,000 live births.[6] Symptoms vary from patient to patient. Physical findings of DiGeorge syndrome include cardiac anomalies, usually conotruncal in nature, such as truncus arteriosus or aortic arch anomalies (approximately 75%), cleft palate (approximately 70%), immunodeficiency due to thymic hypoplasia (approximately 75%), and craniofacial features that include microcephaly, abnormally shaped ears, prominent nasal root with bulbous nasal tip, and hooded eyelids. However, some neonates have no identifying craniofacial features. Associated findings include renal anomalies, hearing loss, significant feeding problems, and hyperextensibility of hands and fingers. Hypocalcemia is a prominent laboratory finding secondary to absence or hypoplasia of the parathyroid glands and thymus. Etiology has been associated with prenatal exposure to alcohol and isotretinoin (Accutane). There is significant neonatal morbidity and mortality associated with the cardiac defects, immunodeficiency, and seizures related to hypocalcemia.[20]

Beckwith–Wiedemann Syndrome

Beckwith–Wiedemann syndrome is caused by a mutation or deletion within the chromosome 11p15.5 region. An estimated one in 13,700 newborns are affected. Beckwith–Wiedemann syndrome is usually identifiable at birth because the infant will be large for gestational age, and have refractory hypoglycemia, a large tongue, creases on the earlobe, and an omphalocele. Hyperplasia of a limb or one side of the face or trunk may be present at birth. Classified as an overgrowth syndrome, affected infants are considerably larger than normal (macrosomia) and continue to grow and gain weight at an unusual rate during childhood. Associated anomalies include renal malformations and cardiomyopathy. Polyhydramnios and a high incidence of prematurity are also common historical findings. Early diagnosis and aggressive treatment of hypoglycemia may prevent mental deficits.[21]

CHARGE Syndrome

CHARGE syndrome is an acronym for **c**oloboma, **h**eart anomaly, choanal **a**tresia, **r**estricted growth and development, **g**enital

anomalies, **e**ar anomalies and/or deafness caused by mutations on the CHD7 gene located on chromosome 8q1[22] Not all features need be present for a diagnosis to be made, and the extent of involvement of each system is widely variable.[9] CHARGE syndrome is diagnosable when three or four major criteria or two major and three minor criteria are present. Occurrence of CHARGE syndrome is one per 12,000 live births.[22] CHARGE syndrome often presents as a medical emergency because of the presence of choanal atresia, serious heart defects, and swallowing difficulties. Associated anomalies include cleft lip and palate as well as unilateral facial palsies. Most patients have some degree of mental deficiency or CNS defect and visual or auditory anomalies that further compromise cognitive function.

SEQUENCES

A sequence is a pattern of multiple anomalies derived from a single known or presumed structural defect or mechanical factor followed by a cascade of secondary effects.[6] The most common nonchromosomal deformation or disruption sequences diagnosed in the neonatal period are Potter oligohydramnios sequence, amniotic band sequence, arthrogryposis, and Pierre Robin sequence.[6]

Potter Oligohydramnios Sequence

The incidence of Potter sequence is one of 3,000 to one of 9,000 live births.[16] Almost all of these infants die in the neonatal period due to pulmonary hypoplasia. Potter sequence is caused by severe oligohydramnios (Figure 13.14). Renal agenesis, polycystic kidneys, urinary tract obstruction, or chronic leakage of amniotic fluid may be the cause of oligohydramnios. This results in intrauterine constraint of the fetus and pulmonary hypoplasia. Physical findings include refractory respiratory distress, frequently with concomitant pneumothoraces, clubfeet, hyperextensible fingers, large ears, low inner eye folds, and a beak nose. Anuria is typically present in the newborn. Associated anomalies include congenital heart defects, Eagle–Barrett syndrome (prune belly syndrome [absent abdominal musculature, urinary tract abnormalities, and cryptorchidism]), esophageal and duodenal

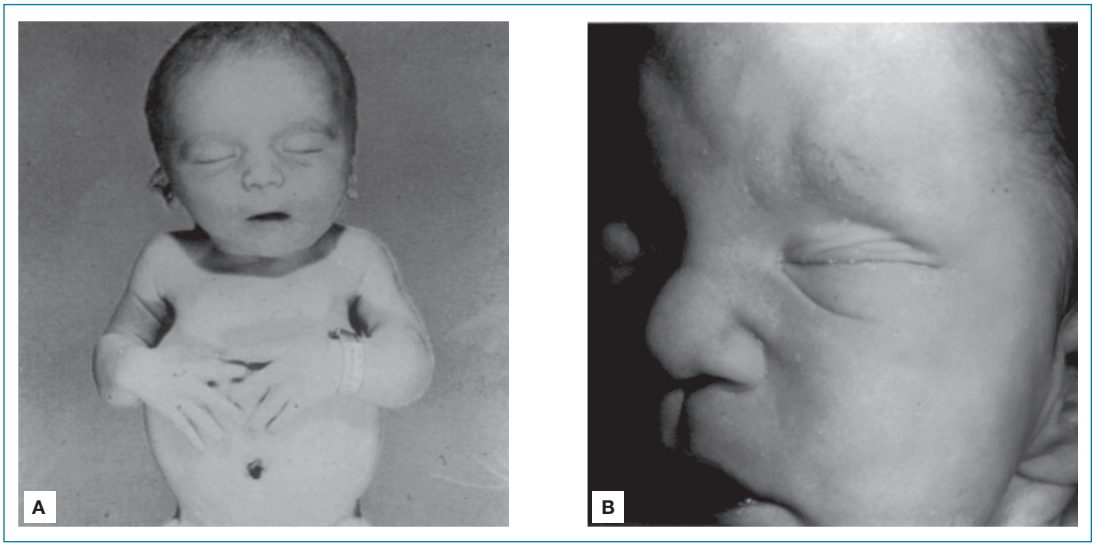

Figure 13.14 Constraint deformities (**A**) Secondary to Potter sequence: narrow, flared thorax, folded ear. (**B**) Typical Potter facies: Flattened nose, ear anomalies, furrowed brow.

Courtesy of J. Hernandez, MD, The Children's Hospital, Denver, CO.

atresias, imperforate anus, and Pierre Robin sequence. Diagnosis is usually confirmed by renal ultrasound and autopsy findings of urinary tract abnormalities.[23]

Amniotic Band Sequence

The incidence of amniotic band sequence is approximately one in 8,000 to one in 11,000 live births.[17] Early amnion rupture occurs, and small bands of amnion encircle developing structures, usually limbs, leading to constrictions, intrauterine amputations, and/or umbilical cord constriction (see Figures 13.6 and 13.7). In addition, deformational defects occur secondary to decreased fetal movement, the result of tethering of a limb by an amniotic band. The decreased fetal movement may result in scoliosis or foot deformities. No two affected fetuses will have the exact same features, and there is no single feature that consistently occurs. Anomalies of the extremities include congenital partial or irregular amputations, constriction rings, and distal swellings. Craniofacial anomalies can include microcephaly, encephaloceles, and facial clefts. Examination of the placenta and membranes is diagnostic.[24]

Arthrogryposis (Multiple Joint Fixations)

Arthrogryposis occurs in approximately one of 8,000 live births.[17] Physical findings include joint contractures, extensions, and dislocations. Joint contractures can be secondary to intrinsic factors affecting the fetus such as early onset of neurologic, muscle, and joint problems or to extrinsic factors such as fetal crowding and constraint. Neurologic abnormality is the most common cause of arthrogryposis. Non–joint-related anomalies may indicate that the arthrogryposis is part of a multiple defect syndrome. Affected infants should also be assessed for scoliosis (Figures 13.15 and 13.16) and hip dislocation.

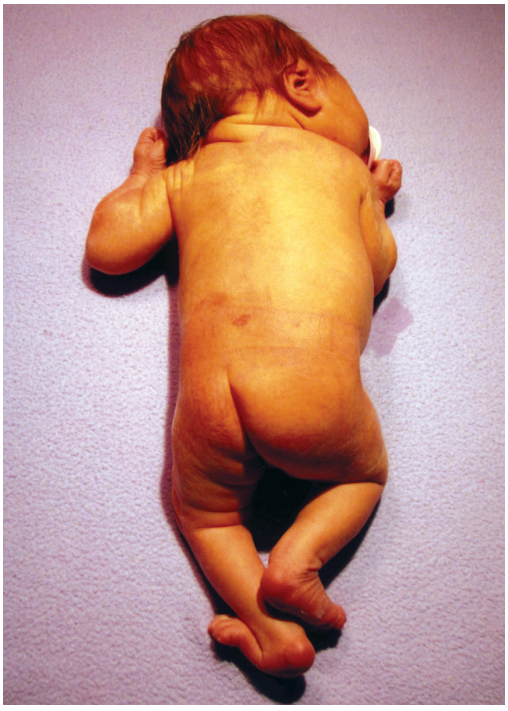

Figure 13.15 Scoliosis

Courtesy of Presbyterian/St. Luke's Medical Center, Denver, CO.

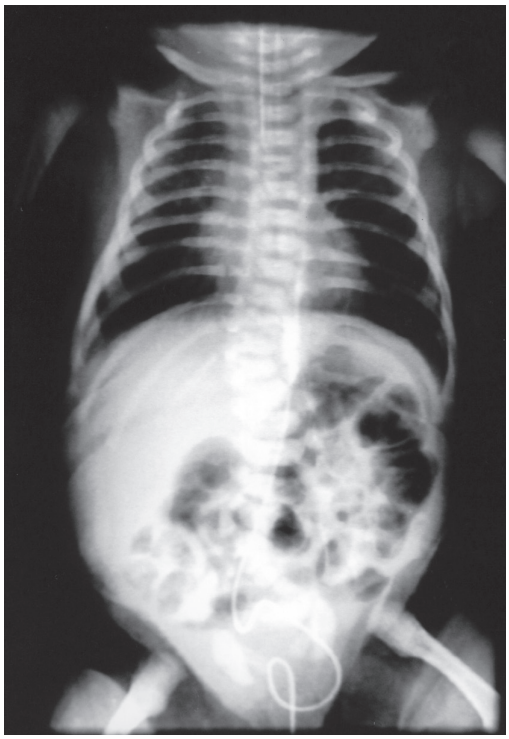

Figure 13.16 X-ray of scoliosis

Pierre Robin Sequence

Pierre Robin sequence occurs in approximately one of 8,500 live births.[6] The initiating defect of this sequence is severe hypoplasia of the mandible, causing the tongue to be posteriorly located, resulting in severe upper airway obstruction and cleft palate. Physical findings include micrognathia, cleft palate, and low-set ears. Respiratory distress secondary to upper airway obstruction may be present. Many syndromes have the craniofacial features of Pierre Robin sequence. If noncraniofacial primary malformations are present, then other diagnoses should be considered.[25]

ASSOCIATIONS

Association refers to a nonrandom occurrence of multiple malformations for which no specific or common etiology has been identified.[8] The most usual association is VATER/VACTERL.

VATER/VACTERL Association

VATER/VACTERL is an acronym that includes **v**ertebral anomalies, **a**nal atresia, esophageal atresia with or without **t**racheo**e**sophageal fistula, and **r**adial and/or **r**enal dysplasia. **C**ardiac defects, single umbilical artery, **l**imb abnormalities, and IUGR are also nonrandom features of this pattern of anomalies. VATER/VACTERL occurs in 1/5,000 live births, and the etiology is unknown.[6] Diagnosis requires exclusion of other similar disorders, including chromosomal syndromes. Most infants diagnosed with VATER/VACTERL have normal brain function and thus merit vigorous attempts toward rehabilitation.

TERATOGENS

Although the human embryo is well protected in the uterus, maternal exposure to teratogens may cause developmental disruptions. A teratogen is any agent external to the fetus that causes a structural or functional disability postnatally. Teratogens can be drugs and chemicals, altered maternal metabolic states, or infectious agents (Table 13.1). Known teratogenic factors cause 5% to 10% of congenital anomalies. Susceptibility to a teratogen is determined by the embryologic stage of development when exposed. Each part, tissue, and organ of an embryo has a critical period during which development can be disrupted (Figure 13.17). The most critical period in development is when cell division, cell differentiation, and morphogenesis are at their peak.

Fetal Alcohol Syndrome

Alcohol is thought to be the most common teratogen to which a fetus may be exposed. The incidence of this disorder in the United States is difficult to estimate, but the risk that an alcoholic woman will have a child with fetal alcohol syndrome is 35% to 40%.[6] Common features include short palpebral fissures; epicanthal folds; a flat nasal bridge; a long, simple philtrum; a thin upper lip; small hypoplastic nails; irritability in infancy; and growth deficiency. Associated anomalies are cardiac defects, ventricular septal defect being the most common, and microcephaly. Long-term effects include mental deficiency and behavioral problems.[6,14]

Fetal Cocaine Exposure

Cocaine has been suggested as a teratogen. Infants exposed to cocaine in utero characteristically are SGA and present with hyperirritability, feeding problems, and abnormal sleep patterns. No definitive physical findings have been established with maternal cocaine use, with the exception of an increased incidence of genitourinary tract anomalies.[6]

Anticonvulsants

Phenytoin (Dilantin) and valproic acid are commonly prescribed for management of maternal epilepsy; however, both

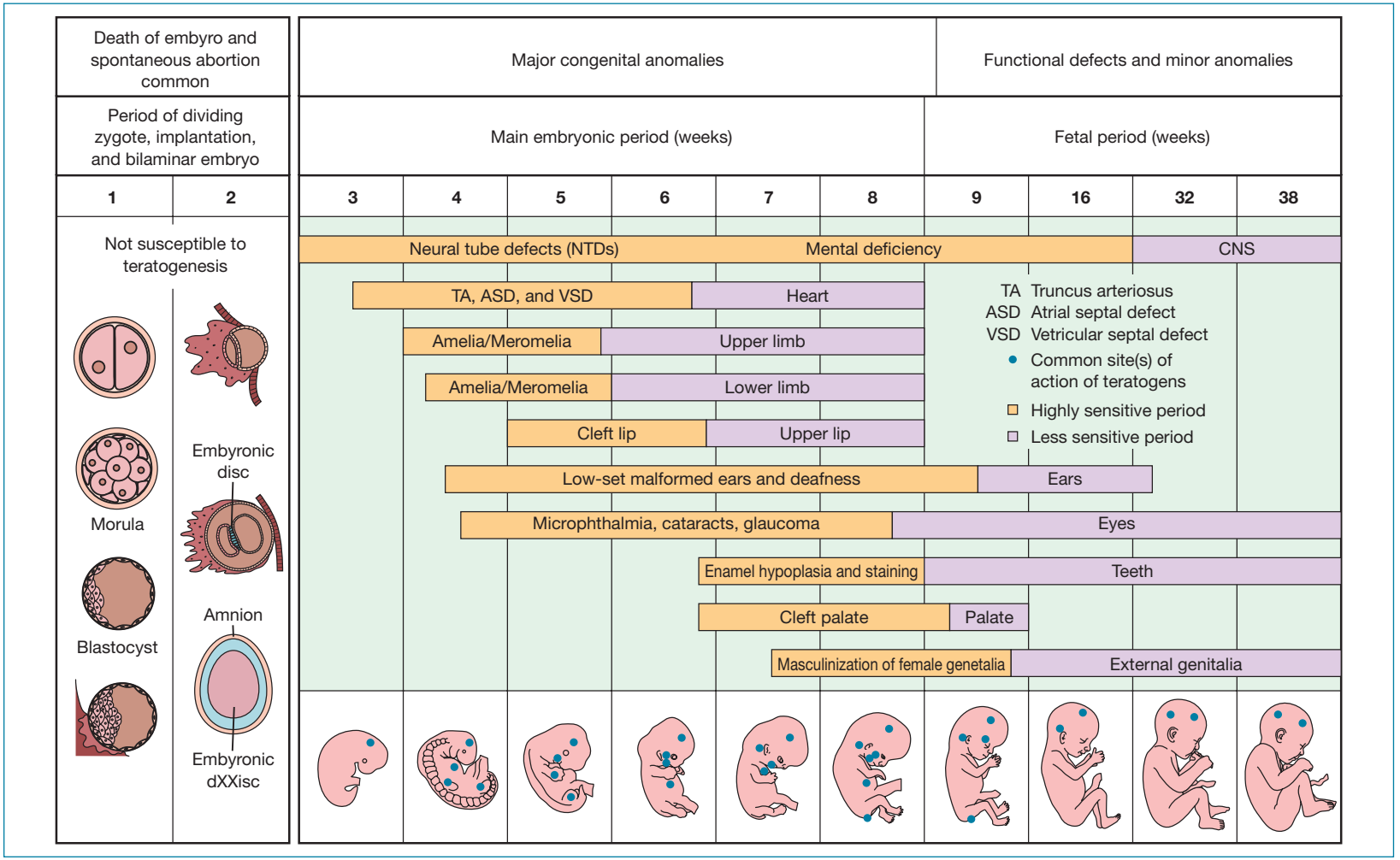

Figure 13.17 Embryonic and fetal development

Source: From Moore KL, Persaud TVN, and Torchia MG. 2013. Human birth defects. In *The Developing Human: Clinically Oriented Embryology*, 9th ed. Philadelphia, PA: Saunders, 489. Reprinted by permission.[3]

TABLE 13.1 SOME TERATOGENS KNOWN TO CAUSE HUMAN CONGENITAL ANOMALIES OR BIRTH DEFECTS

AGENTS	MOST COMMON CONGENITAL ANOMALIES
Drugs	
Alcohol	*Fetal alcohol syndrome*: IUGR; mental deficiency, microcephaly; ocular anomalies; joint abnormalities; short palpebral fissures
Aminopterin	IUGR; skeletal defects; malformations of the CNS, notably meroanencephaly (most of the brain is absent)
Androgens and high doses of progestogens	Varying degrees of masculinization of female fetuses: ambiguous external genitalia resulting in labial fusion and clitoral hypertrophy
Cocaine	IUGR; prematurity, microcephaly, cerebral infarction, urogenital defects, neurobehavioral disturbances
Diethylstilbestrol	Abnormalities of the uterus and vagina; cervical erosion and ridges
Isotretinoin (13-cis-retinoic acid)	Craniofacial abnormalities; NTDs, such as spina bifida cystica; cardiovascular defects; cleft palate; thymic aplasia
Lithium carbonate	Various defects usually involving the heart and great vessels
Methotrexate	Multiple defects, especially skeletal, involving the face, cranium, limbs, and vertebral column
Misoprostol	Limb abnormalities, ocular and cranial nerve defects, autism spectrum disorder
Phenytoin (Dilantin)	*Fetal hydantoin syndrome*: IUGR, microcephaly, mental deficiency, ridged frontal suture, inner epicanthal folds, eyelid ptosis, broad depressed nasal bridge, phalangeal hypoplasia
Tetracycline	Stained teeth, hypoplasia of enamel
Thalidomide	Abnormal development of limbs (e.g., meromelia [partial absence] and amelia [complete absence]), facial defects, systemic anomalies (e.g., cardiac, kidney, and ocular defects)
Trimethadione	Developmental delay, V-shaped eyebrows, low-set ears, cleft lip and/or palate
Valproic acid	Craniofacial anomalies; NTDs, cognitive abnormalities, often hydrocephalus, heart and skeletal defects
Warfarin	Nasal hypoplasia, stippled epiphyses, hypoplastic phalanges; eye anomalies; mental deficiency
Chemicals	
Methylmercury	Cerebral atrophy, spasticity, seizures, mental deficiency
Polychlorinated biphenyls	IUGR, skin discoloration
Infections	
Cytomegalovirus	Microcephaly, chorioretinitis, sensorineural hearing loss, delayed psychomotor/mental development, hepatosplenomegaly, hydrocephaly, cerebral palsy, brain (periventricular) calcification

(*continued*)

TABLE 13.1 SOME TERATOGENS KNOWN TO CAUSE HUMAN CONGENITAL ANOMALIES OR BIRTH DEFECTS (*continued*)

AGENTS	MOST COMMON CONGENITAL ANOMALIES
Herpes simplex virus	Skin vesicles and scarring, chorioretinitis, hepatomegaly, thrombocytopenia, petechiae, hemolytic anemia, hydranencephaly
Human parvovirus B19	Fetal anemia, nonimmune hydrops fetalis, fetal death
Rubella virus	IUGR, postnatal growth retardation, cardiac and great vessel abnormalities, microcephaly, sensorineural deafness, cataracts, microphthalmos, glaucoma, pigmented retinopathy, mental deficiency, neonatal bleeding, hepatosplenomegaly, osteopathy, tooth defects
Toxoplasma gondii	Microcephaly, mental deficiency, microphthalmia, hydrocephaly, chorioretinitis, cerebral calcifications, hearing loss, neurologic disturbances
Treponema pallidum	Hydrocephalus, congenital deafness, mental deficiency, abnormal teeth and bones
Varicella virus	Cutaneous scars (dermatome distribution), neurologic anomalies (limb paresis [incomplete paralysis], hydrocephaly, seizures, etc.), cataracts, microphthalmia, Horner syndrome, optic atrophy, nystagmus, chorioretinitis, microcephaly, mental deficiency, skeletal anomalies (hypoplasia of limbs, fingers, toes, etc.), urogenital abnormalities
Venezuelan equine encephalitis virus	Microcephaly, microphthalmia, cerebral agenesis, CNS necrosis, hydrocephalus
Zika virus	Severe microcephaly with the skull partially collapsed, decreased brain tissue, damage to the back of the eye (e.g., pigment changes, scarring), limited range of motion to joints, hypertonicity[27]
Radiation	
High levels of ionizing radiation	Microcephaly, mental deficiency, skeletal anomalies, growth restriction, cataracts

CNS, central nervous system; IUGR, intrauterine growth restriction; NTDs, neural tube defects.

Sources: Adapted from Moore KL, Persaud TVN, and Torchia MG. 2013. Human birth defects. In *The Developing Human: Clinically Oriented Embryology*, 9th ed. Philadelphia, PA: Saunders, 488. Reprinted by permission.[3] Centers for Disease Control and Prevention. 2018. Congenital Zika syndrome and other defects. Retrieved from https://www.cdc.gov/pregnancy/zika/testing-follow-up/zika-syndrome-birth-defects.html[26]

are teratogens. Fetal hydantoin syndrome is characterized by a typical facies (broad, low nasal bridge, hypertelorism, epicanthal folds, ptosis, and prominent, malformed ears), low-set hairline, and nail hypoplasia. Cleft lip and palate and umbilical and inguinal hernias are associated anomalies.[17] Fetal valproate syndrome features consist of a prominent or fused metopic suture, epicanthal folds, mid-face hypoplasia, and broad, low nasal bridge with short nose and long philtrum. Congenital heart defects, genitourinary anomalies, and club feet are associated anomalies.[6]

Infants of Diabetic Mothers

Maternal altered metabolic states can lead to a higher risk for abnormalities in the newborn. Poorly controlled maternal diabetes mellitus with persistent hyperglycemia and ketosis, particularly during embryogenesis, is associated with a two- to three-fold higher incidence of birth defects.[3,17] Infants of diabetic mothers (IDMs) present with anomalies in approximately 1/2,000 births. Common anomalies include holoprosencephaly (failure of the forebrain to divide into hemispheres), meroencephaly (partial absence of the brain),

sacral agenesis, vertebral anomalies, congenital heart defects, limb defects, and renal anomalies. Improved diabetic control during gestation dramatically decreases the incidence of diabetes-related malformations, but does not reduce it back to the level of incidence for a mother without diabetes.[3,6]

Infectious Diseases

Congenital anomalies also may be associated with certain infections during pregnancy. The common and best-understood infections are represented by the acronym TORCH, which stands for **t**oxoplasmosis, **o**ther agents (including syphilis), **r**ubella, **c**ytomegalovirus, and **h**erpes simplex. TORCH infections may present with similar clinical findings: IUGR; hepatosplenomegaly; rash; CNS manifestations such as microcephaly, chorioretinitis, and intracranial calcifications; jaundice; and low platelets.[27]

RESOURCES

The World Wide Web is a powerful tool to utilize when searching for information regarding birth defects, including specific conditions, diagnosis, prevention, screening, research, and national organizations. An abundance of reliable and up-to-date information from expert sources can be accessed in a short time. Each of the sources listed in the following section provides links to alternate websites if additional information is desired.

Online Resources for Birth Defect Information

Centers for Disease Control and Prevention
http://www.cdc.gov/ncbddd/birthdefects/index.html
Gene Tests
http://www.ncbi.nlm.nih.gov
March of Dimes Birth Defects Foundation
http://www.marchofdimes.com/professionals
Medline Plus
http://www.nlm.nih.gov/medlineplus/birthdefects.html
National Birth Defects Prevention Network (NBDPN)
https://www.nbdpn.org
National Human Genome Research Institute
https://www.genome.gov
National Institute of Child Health and Human Development
http://www.nichd.nih.gov
National Newborn Screening and Genetics Resource Center
http://genes-r-us.uthscsa.edu/
The Teratology Society
http://www.teratology.org/
Online Mendelian Inheritance in Man
http://www.ncbi.nlm.nih.gov/omim
The Genetic Alliance
http://www.geneticalliance.org/

SUMMARY

The approach to the evaluation of the dysmorphic infant is multifaceted and begins with a thorough history and physical examination. With experience, the examiner's identification of physical findings on the continuum of normal to abnormal is enhanced. A general knowledge of genetics and common disorders is helpful when counseling parents. Multiple resources, including geneticists, genetic counselors, and Internet websites, are available to healthcare professionals and parents who are involved in providing care to the dysmorphic infant.

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Pain Assessment in the Newborn

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14

Over the last several decades, there has been an increased awareness of the importance of assessing and managing pain in hospitalized newborns. One only has to spend time in the neonatal intensive care unit (NICU) to observe how frequently newborns encounter postoperative, procedural, and disease-related pain throughout their hospital stay. Although neonates cannot specifically communicate pain, they do display physiologic and behavioral cues that caregivers can use as objective and valid indicators of the infant's pain experience.[1] Caregivers must know the potential causes of pain and use a high index of suspicion when gauging the infant's response cues for the presence, absence, or intensity of pain.[2] The assessment of the infant's response cues is an essential first step to optimally addressing the infant's pain.

APPROACH TO PAIN ASSESSMENT

Pain assessment in the newborn and especially the preterm neonate presents a challenge to even the most skilled clinician. When the examiner depends on a pain score to identify pain consistently and accurately, this can lead to undertreated neonates because of the number of variables that affect physiologic and behavioral responses to painful stimuli. Pain assessment tools are useful insofar as a normal baseline is known and the score is interpreted based on a change from baseline parameters. If a patient is admitted in pain or baseline parameters are abnormal, the pain score may be misinterpreted and undertreatment or overtreatment may occur. More information is required for interpreting pain in neonatal patients.

Infants needing neonatal intensive care may or may not respond to pain with clear signals. There are many reasons, such as low energy reserves, sedation, paralysis, and vague or unclear behavioral cues due to illness and/or prematurity. Therefore, a number of factors must be considered in assessing neonatal pain. A complete approach to assessment is required for the neonatal nurse to adequately determine the presence of pain, interpret the findings, and take action as an advocate for the patient (Figure 14.1). The process involves data collection, a systematic approach to evaluation, and a method of documentation.

Data Collection

Assessing pain in infants depends on systematically collecting various types of data, including the following (Figure 14.2):

- Demographic
- Historic
- Physiologic
- Behavioral
- Risk factors for pain

For successful interpretation and intervention, nursing judgment and communication, both written and verbal, are vital.[4] Without the whole process of collection, interpretation, and intervention, pain may go unobserved and untreated.

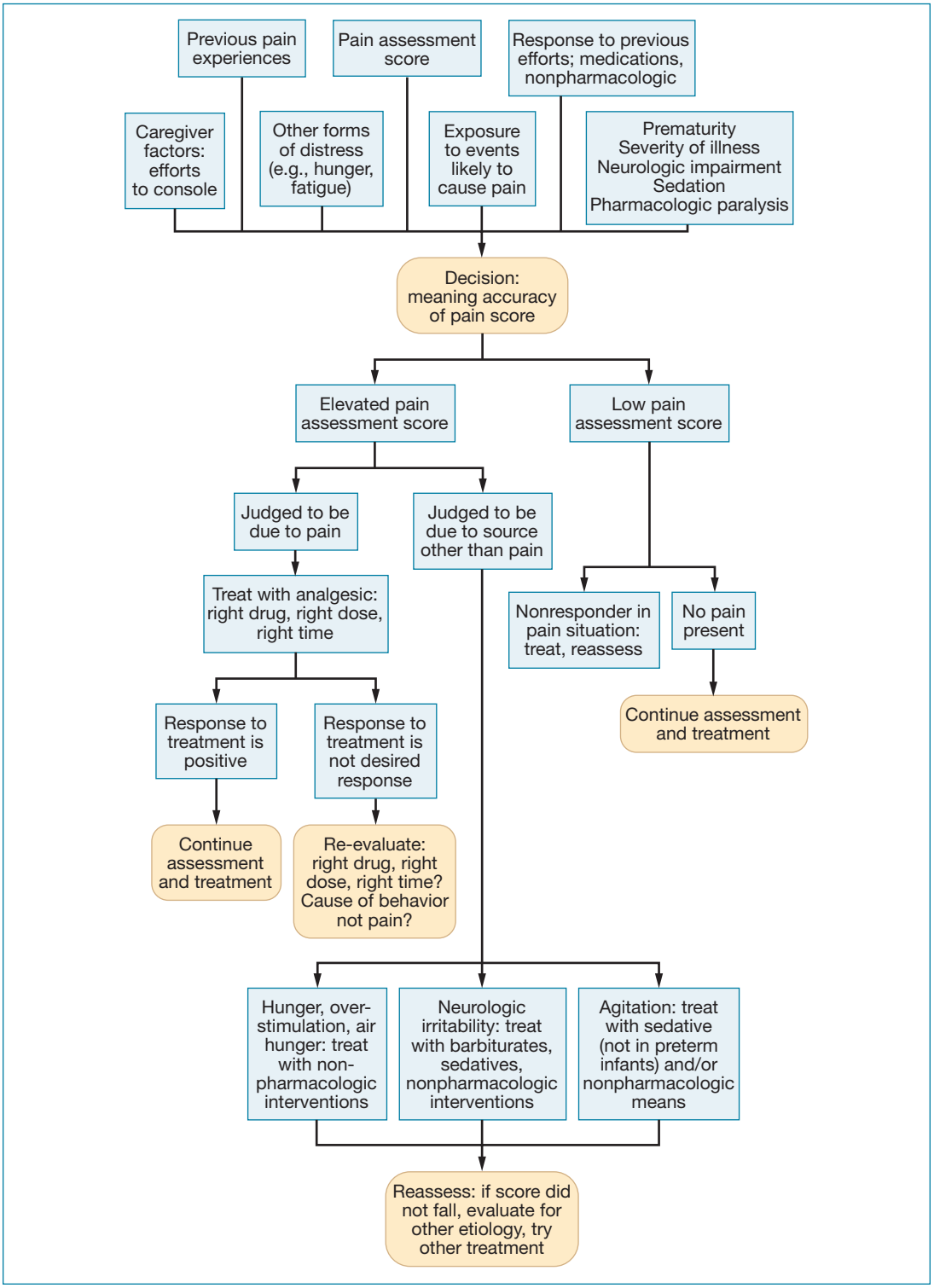

Figure 14.1 Pain assessment algorithm, generalized

Source: Adapted from Hummel P, and van Dijk M. 2006. Pain assessment: Current status and challenges. *Seminars in Fetal & Neonatal Medicine* 11(4): 242. Reprinted by permission.[3]

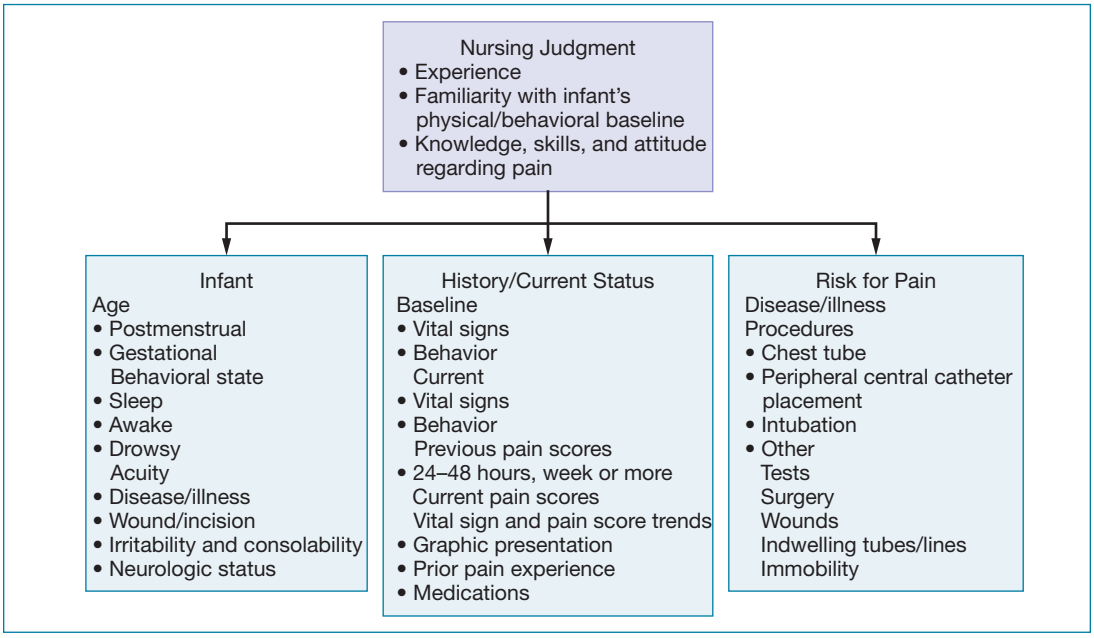

Figure 14.2 Data collection for pain assessment

Systematic Approach to Evaluation

Data collection is only part of the process; it is what a nurse does with the data that will determine whether pain is viewed as being present. Sound nursing judgment comes from education about neonatal pain, experience, and a belief that pain assessment is essential for quality care in the NICU. Once the critical component of nursing judgment is present, the nurse can provide a convincing argument for pain intervention to medical staff. An overall approach to pain assessment that is systematic and provides guidance for nursing practice is shown in Table 14.1.[5,6]

TABLE 14.1 INFANT PAIN ASSESSMENT PROCESS

1. Acknowledging pain cues/signs
2. Hypothesizing the reason for distress
3. Analyzing assessment data using nursing judgment
4. Evaluating effectiveness of comfort measures
5. Assessing consolability following comfort measures
6. Speculating on the pain intensity
7. Providing pain medication when indicated
8. Reassessing at appropriate intervals for analgesic effectiveness or medication tolerance

Source: Adapted from Fuller BF. 1998. The process of infant pain assessment. *Applied Nursing Research* 11(2): 62–68.[5]

In a quasi-experimental study, 24 pediatric nurses were asked to assess pain after viewing videotapes of infants (age range: birth to 12 months) and reading written clinical information. Their pain assessments were compared with assessments by 60 pediatric nurses who viewed the same videotapes but without the written background data. The nurses who only viewed the videotapes rated the pain level as significantly lower than those who also read background clinical information. These nurses were similar in age and experience, demonstrating that clinical information is important for the identification of pain, particularly in nonverbal patients. Nurses from the "video only" group also judged infants from 0 to 3 months as having lower pain levels than those 10 to 12 months.[7] Therefore, judgment of pain requires the nurse to have complete information, especially for infants who demonstrate weak pain signals.

Documentation

Documentation of pain data should be done in a way that helps the caregiver analyze the infant's current pain responses and trends over time. This information allows the nurse to better communicate with the medical team by demonstrating the trend of pain scores and vital signs that may support the need for additional pain management. A single pain score provides immediate information about pain at one moment in time (Figure 14.3), but does not give an overview of fluctuating scores over time; adequacy of pain control; gradually increasing pain; or emerging tolerance to a particular medication, dose, or dose interval. Individual scores are useful when an acute event causes elevated pain scores. However, gradually increasing scores may be missed until the score is high enough to rate intervention. By that time, pain management may be more difficult. A documentation design that can be used in electronic format must be capable of plotting pain scores and vital signs over time (e.g., 12 hours, 24 hours, 1 week). Subtle changes are easier to see if the documentation method allows for an overview of vital signs along with pain scores, as is the case when using a graphic display (Figure 14.4). Nurses familiar with the infant's baseline assessment of vital signs and behaviors will be able to note the subtle variations over time, which may lead to better identification and management of pain.

	0800
Vitals	
Temp	98 (36.7)
Temp src	Axillary
Pulse	160
Resp	38
BP	81/56
MAP (mmHg)	61
BP Location	Left leg
HeRO Documentation	0.45
Oximeter	
SpO2	99
PIPS (Premature Infant Pain Scale)	
Gestational Age	1
Behavioral state	1
HR baseline	160
Sat baseline	99
HR Max	171
Oxygen sat minimum	99
Heart Rate	1
Oxygen sat	0
Brow bulge	0
Eye squeeze	0
Nasolabial furrow	0
PIPS Score	3
Is PIPS Score >7? If Yes a Reassessment is needed:	

Figure 14.3 Example of a single pain evaluation in an electronic medical record. BP, blood pressure; HR, heart rate; MAP, mean arterial pressure.

ASSESSMENT OF PAIN

Pain assessment relies on the careful observational skills of the examiner. Pain can be assessed using behavioral measures or physiologic measures, but the best assessment is done when a multidimensional approach is employed.

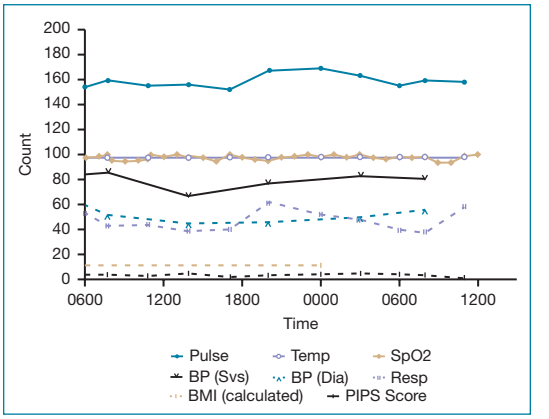

Figure 14.4 Graphic display example of 48 hours trend of pain assessment with vital signs from an EMR. EMR, electronic medical record.

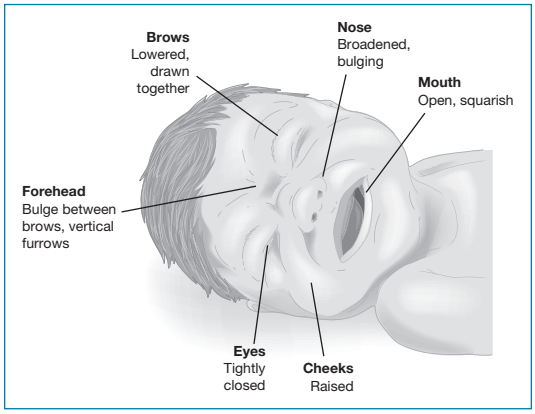

Figure 14.5 Characteristic of "cry face"

Behavioral Indicators

The clinician can observe the infant's facial activity, crying, and body movements to assess for the possibility that the infant is experiencing pain. A "cry face" (Figure 14.5) consisting of subtle changes in the infant's facial expressions such as brow bulge, eye squeeze, and nasolabial furrow is the most specific indicator of acute pain in newborns.[8–11] Gestational age and behavioral state will have a significant impact on the infant's facial expressions, with younger and sleeping infants having a diminished or delayed response.

The clinician can also observe and listen to the infant's cry. Different types of cry—such as high pitched, harsh, intense—may communicate the urgency or severity of distress.[12-13] Absence of cry or objective signs of pain does not necessarily mean that the infant is not in pain, but may only signal that the infant's response capability has been depleted.[11, 14-17]

Individual body movements can also provide helpful information about the infant's pain experience. Infants, particularly healthy term newborns, may use reflexive limb withdrawal in response to noxious stimuli to signal pain.[18] Because of inadequate muscle strength, posture, tone, and movement compared to term newborns, preterm neonates display less vigorous and robust pain responses.[14,15,19] Other observed behaviors exhibited by preterm neonates include increased flexion and extension of arms and toe and finger splay.[20–22]

Physiologic Indicators

Infants may also communicate pain through physiologic changes. The clinician can often observe vital sign changes on the infant's cardiorespiratory monitor to assess an infant's pain. Newborns will acutely respond to pain of handling or procedures with increases in heart rate and blood pressure while oxygen saturation decreases.[3,23] However in prolonged pain, these physiologic parameters may not be valid indicators for assessment.[3]

Assessment Tools

Pain assessment is best accomplished using a published pain assessment tool with known reliability and validity that has been demonstrated to be clinically useful and feasible in the clinical setting. The clinician should also consider choosing an instrument based on a similar infant population, setting, and type of pain.[24] In order to best assess trends over time, the clinician should use the same tool over consecutive assessments when evaluating pain in a given newborn.

A multitude of pain assessment instruments exist to assess and manage pain in hospitalized newborns.[24,25] To improve the science around pain assessment, researchers should focus on validating or refining existing tools rather than continuing to develop new tools. The four most commonly used tools in the newborn are the Premature Infant Pain Profile (PIPP) developed by Stevens and colleagues, the Crying Requires Increased Oxygen, Increased Vital Signs, Expressions, Sleeplessness (CRIES) developed by Bildner and Krechel, the Neonatal Infant Pain Scale (NIPS) developed by Lawrence and colleagues, and the Neonatal Pain Agitation and Sedation Scale (N-PASS) developed by Hummel and colleagues.[26-30]

The PIPP, which measures acute pain, has undergone rigorous psychometric testing.[31] Although the PIPP may be presumed to be valid only with preterm neonates,

it has been tested in neonates ranging in age from 24 to 40 weeks gestational age. The PIPP incorporates two contextual factors that may account for an infant's less robust pain responses, which can result from immaturity or behavioral state. By scoring infants who are younger or asleep higher on the PIPP, the adjusted scores do not penalize those known to be less capable of mounting a robust response to noxious stimuli. The PIPP contains two physiologic indicators (e.g., heart rate and oxygen saturation) and three facial indicators (e.g., brow bulge, eye squeeze, and nasolabial furrow). Physiologic and behavioral indicators are fairly straightforward to score when scoring procedural pain, but are often more challenging if used to score ongoing pain. If no clear 15-second baseline period is available for scoring, the clinician must sometimes judge baseline parameters using either preoperative vital signs or estimated vital signs before the known painful event. Although total scores vary between 18 and 21, depending on the infant's gestational age, scores between 7 and 12 usually signify mild to moderate pain requiring nonpharmacologic comfort measures and possibly a mild analgesic. Scores greater than 12 indicate moderate to severe pain requiring pharmacologic pain intervention in addition to comfort measures. The PIPP has been revised (PIPP-R) to address construct validity and feasibility, allowing the caregiver to more easily apply the PIPP-R (Figure 14.6) in the clinical setting.[32]

Infant Indicator	Indicator Score				Infant Indicator Score
	0	+1	+2	+3	
Change in HR (bpm) Baseline: _____	0–4	5–14	15–24	>24	
Decrease in oxygen SAT (%) Baseline: _____	0–2	3–5	6–8	>8 or increase in O_2	
Brow bulge (seconds)	None (<3)	Minimal (3–10)	Moderate (11–20)	Maximal (>20)	
Eye squeeze (seconds)	None (<3)	Minimal (3–10)	Moderate (11–20)	Maximal (>20)	
Nasolabial furrow (seconds)	None (<3)	Minimal (3–10)	Moderate (11–20)	Maximal (>20)	
				Subtotal Score:*	
Gestational Age (weeks + days)	>36 weeks	32 weeks–35 weeks, 6 days	28 weeks–31 weeks, 6 days	<28 weeks	
Baseline Behavioral State	Active and awake	Quiet and awake	Active and asleep	Quiet and asleep	
				Total Score:**	

*Subtotal for physiological and facial indicators. If subtotal score is >0, add GA and BS indicator scores.

**Total Score should equal Subtotal Score + GA Score + BS Score.

Scoring instructions

Step 1: Observe infant for **15 seconds at rest** and assess vital sign indicators [highest heart rate and lowest O_2 SAT] and behavioral state.

Step 2: Observe infant for **30 seconds after procedure** and assess **change** in vital sign indicators (maximal HR, lowest O_2 SAT* and duration of facial actions observed).
*If infant requires an increase in oxygen at any point before or during procedure, they receive a score of 3 for the O_2 SAT indicator.

Step 3: Score for corrected gestational GA and BS if the subtotal score >0.

Step 4: Calculate total score by adding **Subtotal Score + BS Score.**

Figure 14.6 Premature Infant Pain Profile—Revised. BS, behavioral state; GA, gestational age, HR, heart rate; SAT, saturation.

Source: Adapted from Stevens BJ, et al. 2014. The Premature Infant Pain Profile—Revised (PIPP-R): Initial validation and feasibility. *Clinical Journal of Pain* 30(3): 239.[32]

CRIES (Figure 14.7) is another instrument that has been used extensively to assess postoperative or prolonged pain in neonates between 32 and 60 weeks gestational age.[27] CRIES is an acronym for the five parameters it measures: **c**rying, **r**equires oxygen to maintain saturation greater than 95%, **i**ncreased vital signs, **e**xpression, and **s**leepless. Infants previously requiring oxygen are more difficult to score using the CRIES instrument. It provides no specific guideline for infants previously requiring oxygen, so nursing judgment is required to systematically adjust scores in this category to account for increases in oxygen levels above baseline values. In addition, if the CRIES score is used for procedural pain assessment, baseline vital signs taken immediately before the procedure can be used for scoring the category of "increased vital signs." Total scores for CRIES range from 0 to 10, with scores less than 4 indicative of mild pain requiring nonpharmacologic pain relief measures and scores 5 or greater consistent with moderate to severe pain requiring pharmacologic intervention in conjunction with comfort measures.

The NIPS (Figure 14.8), like the PIPP, was originally developed to assess procedural pain in preterm and term newborns, but literature also validates its utility with postoperative pain.[33-35] The NIPS examines five behavioral parameters (e.g., facial expression, crying, arms, legs, and state of arousal) and one physiologic parameter (e.g., breathing pattern). Total score ranges from 0 to 7. Scoring of the NIPS does not contain physiologic parameters requiring cardiorespiratory monitoring; therefore, this tool is particularly useful in assessing pain in healthy term infants. Although guidelines for pain interventions based on total score are not provided by the developers of the NIPS, all pain instruments in neonates are based on the premise of increasing pain intensity. Therefore, in tools without scoring guidelines for pain management, when pain scores reach the midrange of the total possible points for that tool (e.g., approximately 4 or greater with the NIPS),

	Scoring Criteria for Each Assessment			
	0	**1**	**2**	**Infant's Score**
Crying	No	High pitched	Inconsolable	
Requires O_2 for saturation >95%	No	<30%	>30%	
Increased vital signs*	HR and BP within 10% of preoperative value	HR or BP 11–20% higher than preoperative value	HR or BP 21% or more above preoperative value	
Expression	None	Grimace	Grimace/grunt	
Sleepless	No	Wakes at frequent intervals	Constantly awake	
			Total score†	

* BP should be done last.

† Add scores for all assessments to calculate total score.

Key: BP = blood pressure; HR = heart rate.

Figure 14.7 CRIES: Neonatal Postoperative Pain Assessment Score

Source: Copyright S. Krechel, MD, and J. Bildner, RNC, CNS. Neonatal pain assessment tool developed at the University of Missouri–Columbia. Reprinted by permission.

	Before Time*		During Time*					After Time*		
	1	2	1	2	3	4	5	1	2	3
Facial expression 0: Relaxed 1: Grimace										
Cry 0: No cry 1: Whimper 2: Vigorous										
Breathing patterns 0: Relaxed 1: Change in breathing										
Arms 0: Relaxed/restrained 1: Flexed/extended										
Legs 0: Relaxed/restrained 1: Flexed/extended										
State of arousal 0: Sleeping/awake 1: Fussy										
Total										

* Time is measured in one (1) minute intervals.

Figure 14.8 Neonatal Infant Pain Scale

Source: Copyright 1989, Children's Hospital of Eastern Ontario, Ontario, Canada. Reprinted by permission.

the clinician may infer that the infant is experiencing moderate to severe pain and that pharmacologic intervention for that pain is warranted.

The N-PASS measures acute pain, prolonged pain, and level of sedation in neonates between 23 and 40 weeks gestational age.[29,30] The N-PASS contains four behavioral indicators (crying/irritability, behavior state, facial expression, extremities/tone) and one physiologic indicator (vital signs). Like the PIPP, points are added to the pain score based on the infant's gestational age to compensate for the preterm neonate's limited ability to behaviorally or physiologically communicate pain. Total pain scores vary depending on the infant's gestational age, but scores greater than three usually signify pain requiring nonpharmacologic comfort measures and/or pharmacologic pain intervention. The sedation portion of the N-PASS requires an assessment of response to stimuli and can be useful in titrating opioid requirements based on level of sedation desired. Sedation is scored between 0 and 2 for each behavioral and physiologic criterion, then summed and noted as a negative score (0 to -10). Scores of -10 to -5 are considered deep sedation, whereas light sedation scores range from -5 to -2.

SPECIAL POPULATIONS

The evidence base on pain assessment is still limited for extremely low-for-gestational-age (ELGA), neurologically impaired, pharmacologically paralyzed or sedated, and other special populations of infants.

ELGA Infants

Gestation at birth and postmenstrual age both affect pain response, with more robust responses to pain stimuli noted with advancing gestation and postmenstrual age.[36] The ELGA infant's lack of response does not

necessarily indicate a lack of pain perception, but may be due to factors such as exhaustion, minimal energy reserves, protective apathy (resulting from the infant's repeated attempts to communicate that he is in pain with no response from caregivers), or disease progression.[14,37,38]

The preterm infant's pain response is affected by severity of illness, previous pain experiences, number of painful procedures, and medications. Cry is not a sensitive indicator for pain in ELGA infants because many are intubated, on continuous positive airway pressure (CPAP), or lack the energy for crying.[38]

Preterm and term infant responses to pain differ from those of 2- and 4-month-old infants, who show no difference between each other in pain responses. Preterm infants demonstrate longer latency for cry and have higher pitched cries; term newborns show more tautness of tongue than either preterm or older infants. Two- and 4-month-old infants have more vertical mouth position than preterm or term newborns, who display more horizontal mouth action.[39]

Facial actions in both preterm and term infants have been demonstrated to show more sensitivity and specificity to pain than physiologic parameters, and some behaviors, such as crying, may be influenced by a variety of conditions such as hunger, agitation, a stressful environment, and repositioning.[36,40]

So many factors influence healthcare physicians and staff in effectively recognizing pain in ELGA infants within the context of a clinical environment. Context in the NICU includes unit culture, workload, experience of the team, patient acuity, process efficiency, and general knowledge of neonatal pain behaviors that evolve with development and maturity. It is essential to understand the caregivers' experience, knowledge, and attitudes regarding pain. Caregivers express uncertainty in their assessment of ELGA infants because of variable and sometimes very subtle indications of pain. More research is needed on pain responses in special populations, such as the ELGA infant, to assist healthcare staff in recognizing and treating pain in vulnerable infants.[41]

Neurologically Impaired Infants

A survey of direct care staff suggests there is a generalized perception that, as the level of neurologic impairment increases from mild to moderate to severe, the infant experiences progressively less pain compared to infants without impairment.[42,43]

Stevens and colleagues conducted a study using Delphi methodology to gain group consensus among 14 pediatric pain experts on pain indicators thought to be characteristic of infants at risk for neurologic impairment (low, moderate, severe risk). The highest level of agreement among experts on pain indicators in infants at risk for neurologic impairment was on brow bulge, facial grimace, eye squeeze, and inconsolability. The expert panel also agreed that, for the severe risk group, heart rate changes and decreased oxygen saturation were important indicators of pain.[44]

Another study found infants at a postmenstrual age of approximately 32 weeks with grade IV intraventricular hemorrhage or cystic periventricular leukomalacia by ultrasound showed no signs of altered pain response compared with matched controls. The infants with parenchymal brain injury demonstrated significantly more tongue protrusion upon heel lance.[45]

In a prospective cohort study, pain was compared in healthy newborn babies with babies with Down syndrome who had developmental problems. The Down syndrome babies expressed pain more slowly and the response was less clearly defined than in those babies without Down syndrome. Babies with Down syndrome had significantly lower oxygen saturation levels after the puncture ($p < .001$). When pain was finally detected in these babies, it took twice as long for them to recover to baseline values than the control group ($p < .001$). Oxygen saturation and pain response after a painful procedure was not affected by skin-to-skin contact. Nurses need to keep in mind that Down syndrome babies are neither quick nor robust in their response to pain. It is essential to remember that pain control is still necessary for these babies

to reduce pain and possibly aid in faster recovery from procedures.[46]

Abnormal pain responses later in infancy may become apparent as neurodevelopmental abnormalities that blunt or alter biobehavioral reactivity emerge.[47] A study, published in 2008, by Gibbins and colleagues found that behavioral and physiologic pain responses in at-risk infants were diminished at 6 months of age compared to the neonatal period.[48]

Pharmacologically Paralyzed or Sedated Infants

Infants receiving moderate to heavy sedation or paralytic agents cannot mount a behavioral response to pain. Risk factors for pain should be carefully considered, and physiologic measures of pain should be used in the absence of behavioral indicators.[49,50] Unfortunately, there are no available pain instruments for use with this special NICU population. However, although not currently available for bedside use, new and emerging technologies, such as near-infrared spectroscopy (NIRS), amplitude-integrated electroencephalography, functional magnetic resonance imaging (fMRI), skin conductance, and heart rate variability assessment, are being explored and may be future technology used to assess pain in these infants.[25,51]

Infants With Persistent or Chronic Pain

Prolonged or persistent pain is not well described due to limited research, especially in the preterm population. Most of the research on neonatal pain consists of an acute pain stimulus (heel lance) or postoperative pain.[52] In one study, 22 ventilated preterm infants, randomized to receive morphine or a placebo, were assessed for ongoing or persistent pain by clinical staff, including nurses and doctors. Infants on morphine therapy were correctly identified by staff members 71% of the time. Pain-related facial expressions (grimacing), high activity levels, poor response to handling or routine care, and insufficient ventilatory synchrony were associated more with infants on placebo than those receiving morphine.[53]

Neonatal staff often form a general impression when assessing persistent pain in neonates. Indicators most commonly used by staff include physiological indicators, facial expression, body posture and movements, response to handling, sleep patterns, and subjective opinion. Neonatal staff report more difficulty in identifying persistent pain and comfort than do parents observing their own baby.[54]

It has been suggested that neonatal pain that lasts longer than 7 days should be treated as chronic pain. Over time, chronic pain can blunt the physiologic response to pain. An infant with acute pain may show dramatic changes in vital signs, such as heart rate, whereas an infant with chronic pain may have little change in heart rate variability.[55]

Initial validation with the EDIN (Échelle Douleur Inconfort Nouveau-Né [Scale of Pain and Discomfort Newborn]) scale for assessing prolonged pain in preterm infants is promising, but requires additional research.[56] A study by Ancora and colleagues showed that gestational age affected EDIN scores of infants exposed to prolonged pain.[57] EDIN scores were lowest in the extremely low-birth- weight (ELBW) infants as compared to infants who were gestationally more mature. Five behavioral indicators with increasing scores indicating prolonged pain comprise this scale: facial activity, body movements, quality of sleep, quality of nurse–infant interaction, and consolability (Figure 14.9).

More subdued responses may be seen in infants with chronic pain, or a complete shutdown may become apparent when pain exceeds the infant's ability to respond, as with any overwhelming stimulation.[36,52] These reactions may be caused by the ongoing nature of persistent or chronic pain, during which the usual pain signals do not result in relief, thus resulting in a negative feedback loop.

Indicator	Score/Description	Results
Facial activity	0: Relaxed facial activity 1: Transient grimaces with frowning, lip purse, and chin quiver 2: Frequent grimaces, lasting grimaces 3: Permanent grimaces resembling crying or blank face	
Body movements	0: Relaxed body movements 1: Transient agitation, often quiet 2: Frequent agitation but can be calmed down 3: Permanent agitation with contraction of fingers and toes and hypertonia of limbs or infrequent, slow movements and prostration	
Quality of sleep	0: Falls asleep easily 1: Falls asleep with difficulty 2: Frequent, spontaneous arousals, independent of nursing, restless sleep 3: Sleeplessness	
Quality of contact with nurses	0: Smiles, attentive to voice 1: Transient apprehension during interactions with nurses 2: Difficulty communicating with nurses, cries in response to minor stimulation 3: Refusal to communicate with nurses, no interpersonal rapport, moans without stimulation	
Consolability	0: Quiet, total relaxation 1: Calms down quickly in response to stroking or voice or with sucking 2: Calms down with difficulty 3: Disconsolate, sucks desperately	
	TOTAL SCORE =	

Figure 14.9 EDIN Pain Scale—Assessment of prolonged pain in premature infants

Source: Adapted from Debillon T, et al. 2001. Development and initial validation of the EDIN scale, a new tool for assessing prolonged pain in preterm infants. *Archives of Disease in Childhood. Fetal Neonatal Edition* 85(1): F37. Reprinted by permission.[56]

Infants Exposed to Psychotropic Medications

Infants exposed prenatally or postnatally through breast milk to selective serotonin reuptake inhibitors (SSRIs) and benzodiazepines demonstrate significantly decreased facial action and less cardiac reactivity during heel lance than those not exposed to psychotropic medications. Mean heart rate was significantly lower during recovery from heel lance for the SSRI-exposed infants.[47,58]

Infants Undergoing Opioid Weaning

Screening for pain should continue during opioid weaning. One might suspect recurring pain in infants with increased neonatal abstinence scores who frequently need extra doses of opioids above the weaning dose for symptom management. Although alternate dosing of opioids and benzodiazepines has not been studied in neonates during weaning, this combination might diminish the pain response in infants still experiencing pain. Pain screening with a validated tool in combination with neonatal abstinence scoring can alert the clinician to the presence of pain because some indicators differ among instruments. For example, facial grimacing, a sensitive indicator of pain, is not on abstinence scoring tools.

Iatrogenic withdrawal differs from neonatal abstinence in that dependence results from medications to alleviate pain. Therefore, pain may be inadequately treated if medications are weaned before the cause of pain diminishes. Healthcare providers must be able to distinguish between iatrogenic withdrawal and neonatal abstinence syndrome.[59]

Infants at End of Life

A study by Fortney and Steward reported documentation of pain scores and symptoms of discomfort in neonates at end of life is

frequently inadequate. Inadequate medical record documentation may lead to undertreatment of pain throughout the dying process.[60] Currently, there is no appropriate tool sensitive to neonatal pain at the end of life. Choice of an assessment tool depends on the individual patient (e.g., a preterm infant might be best evaluated using the PIPP to account for gestational age). Often, infants who are terminally ill do not have the energy to express pain behaviorally. Significant impairment of the terminally ill infant's communication ability could compromise comfort. Physiologic indicators may be the only available parameters, and these might be affected by the dying infant's condition. Therefore, it is reasonable to use physiologic indicators, risk factors for pain, and an infant's general condition to help determine pain at this time.[49]

There is great variation in the provision of analgesics to infants during and after ventilator withdrawal, with many infants receiving no analgesia. Infants with major chromosome abnormalities, congenital anomalies, and necrotizing enterocolitis (NEC) were more likely to receive analgesia than those with other diagnoses or for whom further treatment was considered futile.[61,62] The provision of analgesics is frequently based on obvious risk factors for suffering as life support is withdrawn or for known painful conditions. Frequently, infant behaviors are not documented as the reason for administering pain medications.[63] As stated, the inability to demonstrate behavioral pain responses leaves the clinician without clear indices for assessment. Therefore, it may be necessary to consider that infants may have increasing pain intensity as death becomes imminent. Clearly, a population that receives little or no analgesia at a time when most older children and adults receive compassionate analgesia indicates a different standard of care and warrants a close examination of clinical practice.

In a study examining the educational needs of neonatal nurses regarding neonatal palliative care, 50% ranked "pain control" as an educational priority.[64] In some institutions, palliative care teams provide support for the patient and family and serve as consultants for pain management of the neonate during end of life.

INFANTS WITH DISEASES OR CONDITIONS WARRANTING PAIN ASSESSMENT

One way to think about pain related to particular conditions is to consider whether the condition would be painful in an older child or adult. If the answer is yes, then pain needs to be frequently assessed. Tests, procedures, or wound care may increase the level of pain and require careful observation to determine the need for pain medication. Some conditions, such as epidermolysis bullosa (EB), require varying amounts of analgesia, from ongoing baseline pain control to control of acute pain when dressings are changed. A study of 140 randomly selected children and 374 adults with various types and subtypes of EB found that only 12% to 13% reported no pain. Individuals with more extensive EB and EB subtypes reported pain levels greater than 5 on a 10-point pain scale.[65]

Another study of 35 children, ages 5 to 18, with osteogenesis imperfecta found they experienced moderate to severe pain related to fractures and less intense pain when no fractures were present. With nonverbal patients, it would be easy to assume that they have no pain when they have no fractures.[66] This would lead to undertreatment of pain in this population. These patients may experience acute pain during repositioning or other care not usually considered painful.

In a retrospective chart review of 25 infants with NEC, it was noted that infants averaged 13 to 19 painful events per day for 5 days. Although the unit standard was to document two PIPP scores per day, compliance was less than 8%. For 2 days after NEC was diagnosed, only 30% of infants had pain scores documented in the medical record. From day 3 to 5, PIPP scores were documented on 60% of the infants. On day 1 of diagnosis, 52% of infants were given analgesia, and on day 2, the percentage receiving analgesia rose to 76%. After day 2, the frequency of

analgesic treatment slowly declined.[67] In this study, the use of opioid analgesics for NEC was low, possibly in association with inadequate screening assessments for pain.

Neonatal transfers are increasing with the centralization of tertiary care. These infants are at risk for experiencing pain during transport. A prospective cohort study of 140 infants was conducted with the babies nested on a gel mattress in a covered transport incubator. For all gestational ages, pain peaked significantly during road transfer. Pain assessment during transfer and appropriate treatment are critical components of transport.[68]

Many conditions in the NICU result in prolonged pain that can affect the nature of biobehavioral responsiveness, and the onus of responsibility is on the clinician to appropriately determine whether the infant is in pain. When pain is questionable, a compassionate clinician can always err on the side that pain is present, initiate a trial of analgesic therapy, and evaluate infant response.

PARENTS' VIEWS ON PAIN

Core concepts central to family-centered care highlight the importance of information sharing and shared decision making between parents and the healthcare team. Over the past decade, parents have voiced an increasing desire for more information about their infant's pain and to have the opportunity to participate in their infant's pain relief.[69–71]

Surveys of parents regarding pain assessment and management for their infant in the NICU provide some guidance concerning their needs associated with neonatal pain. In one survey of 257 parents from nine NICUs in the United Kingdom and the United States, 64% of parents were dissatisfied because they received no information about infant pain. Thirty percent of parents were disappointed with the information they had received. Only 18% of parents reported being taught by NICU staff how to identify their infant's pain signals. Seventy percent of parents wrote comments on ways they identified their infant's pain signs, with crying (37%), movements (31%), or facial expression (21%) being the most commonly reported. Very few parents used skin color or cardiorespiratory monitor data for pain assessment.[72]

A study by Franck and colleagues in 2011 provided parents with written information on pain assessment and comfort techniques. Study findings demonstrated that while maternal satisfaction increased, stress was not reduced.[73] Timing of information received and increased involvement opportunities were important to NICU parents. Consideration of the parents' emotional states and quality of NICU staff communication and support are positively related to parents' desired level of participation.[74]

Interviews and focus groups with parents show that their infant's pain is a source of stress and that health team members have an important role in alleviating that stress.[75] The ability to participate in parenting their infant seems to provide a coping mechanism that relieved some of the distress associated with the pain their infant endured in the NICU. Parents felt there was a difference between their perceptions of their infant's pain and the perceptions of NICU staff members. This disparity increased their anxiety, as did the concern that staff might not intervene when presented with signs of pain by their infant after the parents left the NICU. These issues heightened parental stress, whereas support by NICU staff, parenting opportunities, and information or resources relieved it.

Parental participation in pain management activities are enhanced when parents are provided with adequate and ongoing information about their infant's medical procedures, knowledge of effective pain management strategies, and knowing how they can be an active partner in providing pain relief to their infant.[69] A family-friendly environment and close communication and collaboration between parents and the healthcare team may also foster parental participation. In contrast, work culture, unsupportive hospital policies, and paternalistic staff attitudes may hinder parental involvement during painful procedures.[69,70]

SUMMARY

Pain assessment is an essential prerequisite to optimal pain management. Pain is best assessed when a multidimensional approach is employed using a valid and reliable instrument. Balancing pain assessment data with the infant's risk factors for pain and contextual modifiers that can impact how the infant communicates pain will ensure that pain is recognized early and interventions are implemented in a timely manner.

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Assessment of the Newborn With Antenatal Exposure to Drugs

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15

Substance use disorders during pregnancy are a significant health issue. A thorough assessment of a newborn involves the evaluation for possible in utero exposure to tobacco, alcohol, marijuana, and other pharmacologic agents either prescribed or from illicit use. The National Survey on Drug Use and Health (NSDUH) conducted by the Substance Abuse and Mental Health Services Administration (SAMHSA) provides data on the use of tobacco, alcohol, marijuana, and illicit drugs, including the misuse of prescription medications, in the United States. This data have been collected for several years by asking mothers about their use of substances in the month prior to answering the survey questions. The surveys report this data as "past month substance use." In 2016, "past month use" among all females aged 15 to 44 increased from 7.7 million in 2015 to 8.3 million in 2016; although use in the past month among pregnant women aged 15 to 44 increased from 109,000 in 2015 to 143,000 in 2016 (Table 15.1).[1]

The most common illicit drug used was marijuana followed by opioids (heroin use or misuse of pain relievers), and misuse of psychotherapeutics.[1] The misuse of prescription pain relievers and the opioid epidemic in the United States has exponentially increased in recent years causing an increased number of infants experiencing neonatal abstinence syndrome (NAS) and requiring admission to a neonatal intensive care unit (NICU) for treatment. The rate of NICU admissions for NAS from 2004 to 2013 increased from seven cases per 1,000 admissions to 27 cases per 1,000 admissions, and the median length of stay increased from 13 to 19 days.[2] The rise in infants experiencing NAS is directly correlated with the increase in maternal use of opioids during pregnancy.[3] These trends support the need to adequately identify, assess, and manage the infant with perinatal substance exposure.

The use of alcohol during pregnancy also continues to be a significant health issue, yet alcohol use among pregnant females aged 15 to 44 decreased from 9.3% in 2015 to 8.3% in 2016 (Table 15.2).[1] Although it is difficult to obtain exact data on the number of infants exposed to alcohol in utero who subsequently develop fetal alcohol syndrome (FAS) or fetal alcohol spectrum disorders (FASD); it is estimated that 0.2 to 1.5 infants for every 1,000 live births have FAS and potentially two to five of every 100 school children exhibit characteristics of FASD.[4] In 2016 when compared in age, income, and insurance carrier, the population with the highest percentage of alcohol use during pregnancy was women aged 26 to 44, with incomes at 200% or more above the poverty level, who had private insurance coverage.[1]

In addition to illicit drug and alcohol use, there is concern for cigarette use during pregnancy. According to the survey, the percentage of all females aged 15 to 44 who used cigarettes in the past month decreased from 21.1 in 2015 to 19.5 in 2016; whereas the percentage of pregnant females aged 15 to 44 who used cigarettes decreased from 13.6 to 10.[1] Although this is an encouraging trend toward decreased cigarette use during pregnancy, there is still room for improvement.

TABLE 15.1: ILLICIT DRUG USE IN PAST MONTH AMONG PREGNANT FEMALES AGED 15 TO 44

DRUG	2015 (%)	2016 (%)
Illicit drugs (total)	4.7	6.3
Marijuana	3.4	4.9
Opioids (heroin use or pain reliever misuse)	0.8	1.2
Misuse of psychotherapeutics (pain relievers, tranquilizers, stimulants sedatives)	1	1.4
Cocaine	0.0	0.1
Hallucinogens (LSD, PCP, ecstasy)	0.1	0.0

LSD; lysergic acid diethylamide; PCP, phencyclidine.

Source: Center for Behavioral Health Statistics and Quality. 2017. 2016 National Survey on Drug Use and Health: Detailed tables. Rockville, MD: Substance Abuse and Mental Health Services Administration. Retrieved from https://www.samhsa.gov/data/sites/default/files/NSDUH-DetTabs-2016/NSDUH-DetTabs-2016.hem#tabs6-65B[5]

TABLE 15.2: PERCENTAGE OF ALCOHOL USE IN THE PAST MONTH AMONG FEMALES AGED 14 TO 55 BY PREGNANCY STATUS

2016	PREGNANT (%)	NONPREGNANT (%)
Total alcohol use in past month	8.3	53.5
Binge alcohol use in past month[a]	4.3	28.6
Heavy alcohol use in past month[a]	0.9	5.8

[a]"Binge alcohol use is defined as drinking five or more drinks (for males) or four or more drinks (for females) on the same occasion (i.e., at the same time or within a couple of hours of each other) on at least 1 day in the past 30 days. In 2015, the definition for females changed from five to four drinks. Heavy alcohol use is defined as binge drinking on the same occasion on each of 5 or more days in the past 30 days; all heavy alcohol users are also binge alcohol users."[1]

ASSESSMENT AND IDENTIFICATION

The first step in assessing an infant with perinatal substance exposure is accurate identification. Perinatal substance abuse is a complex issue and crosses socioeconomic and racial lines making an objective approach to identifying risk factors important.[6] The most comprehensive approach to identification of perinatal substance abuse includes an accurate review of maternal history for self-reported use, risk factors for maternal use, neonatal history and presentation, as well as biologic specimens supporting maternal use or neonatal exposure.[7]

MATERNAL RISK FACTORS

The combination of a structured interview and review of maternal history along with the evaluation of a biologic specimen increases the likelihood of identification.[8] Maternal history consistent with an increased risk of perinatal substance abuse includes limited, inadequate, or no prenatal care, exposure to violence and unresolved trauma, significant mental illness, perinatal depression, poor nutrition, presence of sexually transmitted maternal diseases, premature onset of labor, abruptio placentae, intrauterine growth restriction, and congenital malformations.[9,10] In one study, the presence of maternal perinatal depression was correlated with a 23% prevalence of a substance abuse issue.[10]

NEWBORN RISK FACTORS

Newborns born to women struggling with a substance use disorder during pregnancy have an increased risk of complications. Due to the prevalence of polysubstance use it is difficult to determine a direct cause between a specific substance use during pregnancy and a specific newborn complication. Prematurity, intrauterine growth restriction, jitteriness, hypertonicity, congenital malformations, and NAS have all been associated with perinatal substance abuse.[7]

BIOLOGIC SPECIMENS

Biologic specimens available to aide in the identification of in utero exposure to nicotine, alcohol, and illicit drugs include urine, hair, meconium, and umbilical cord tissue.

Each specimen has its own unique challenges with collection and accuracy of results.[7] Each specimen only identifies exposure within a limited time frame between exposure and collection of a sample. This variability in identification leads to the potential for a false negative test result. Biologic specimens should be considered only a part of a comprehensive evaluation for perinatal substance use. However, even with this combined approach of targeted interviews, a comprehensive review of both maternal and newborn histories, and the use of biologic specimens, some newborns with antenatal exposure may not be identified.[11]

IMPACT ON THE FETUS

An accurate assessment of the neonate with in utero drug exposure requires an understanding of the mechanism of the impact of fetal exposure to drugs. Impact on the fetus is determined by a multitude of factors. Exposure during the embryonic phase of gestation, when a women may not yet realize she is pregnant, can have teratogenic effects such as seen with FAS. During the next phase of development, the fetal period, major structural development has occurred. Drug exposure during this phase may be more subtle upon examination but can include abnormal growth patterns, as well as alterations in neurotransmitters, receptors, and brain organization.[12] In addition to exposure to substances in utero, maternal lifestyle challenges, including poor nutrition, limited or no access to healthcare, exposure to violence, as well as a history of maternal mental illness, can impact fetal growth and development.[9]

ALCOHOL

Teratogenic effects of maternal alcohol consumption during pregnancy have been well documented since 1973.[13] The impact on the fetus occurs throughout gestation and can include identifiable physical characteristics noted in the neonatal period as well as behavioral problems and learning deficits identified after the neonatal period. The wide range of potential findings are referred to as *FASDs*. The most impactful end of the FASD spectrum is FAS. A neonate with FAS may exhibit craniofacial abnormalities such as smooth philtrum, thin upper lip, short palpebral fissures, growth restriction, and central nervous system dysfunction.[9,12,13]

NICOTINE

Maternal smoking has been linked to intrauterine growth restriction and preterm birth. One study suggests a 5% reduction in relative fetal weight for each pack of cigarettes smoked per day.[13] A newborn exposed to in utero cigarette smoke may demonstrate jitteriness and hypertonicity from exposure to nicotine. Potential long-term morbidities related to fetal exposure to nicotine and toxins from cigarette smoke include decreased cognitive functioning, auditory processing deficits, impulsivity, anxiety, depression, attention deficit hyperactivity disorder, reduced lung function, and asthma in offspring.[14] Maternal nicotine use has limited data supporting an association with facial clefts in exposed newborns.[15,16]

COCAINE

Cocaine alters the norepinephrine, dopamine, and serotonin neurotransmitter pathways and readily crosses the placenta and is excreted into breast milk. Exposure to cocaine can result in tachycardia, arrhythmias, hypertension, vasoconstriction, diaphoresis, and mild tremors when in the bloodstream. Newborns may present with these symptoms when cocaine is present in the bloodstream as a result of maternal use. There is no specific withdrawal syndrome reported in newborns exposed to in utero cocaine. Antenatal exposure to cocaine has a significant impact on fetal growth due to restriction of placental blood flow resulting in decreased birth weight, length, and head circumference in term newborns.[11] The vasoconstriction associated with cocaine has also

been correlated with placental abruption, intestinal atresia, and necrotizing enterocolitis in a small series of term and preterm newborns prenatally exposed to cocaine.[11] However, no clear association with a teratogenic effect has been identified with maternal use of cocaine during pregnancy.[11]

AMPHETAMINES

Methamphetamine is a potent sympathomimetic stimulant causing vasoconstriction. Similar to the use of cocaine during pregnancy, the use of methamphetamine during pregnancy has been associated with premature birth, growth restriction, placental abruption, and fetal distress.[11] There are no clear associations with methamphetamine use during pregnancy and a withdrawal syndrome or congenital malformations, yet there is some concern for long-term cognitive deficits and physical dexterity.[11]

MARIJUANA

Marijuana is the most frequently used illicit drug during pregnancy. The frequency of use during pregnancy continues to rise in correlation with the legalization of marijuana for both medical and recreational use in some states.[1] The active psychogenic component of marijuana is cannabinoid delta-9-tetrahydrocannabinol (THC). Marijuana readily crosses the placenta and is in breast milk and may be associated with fetal growth restriction, prematurity, and stillbirth.[17] In addition, women who used marijuana during their pregnancy had a higher incidence of anemia and newborns had a lower birth weight and higher NICU admission rate.[18] Also, newborns exposed to marijuana prenatally have an association with decreased executive functioning skills, including poor impulse control, visual memory, and attention deficit.[19] No clear withdrawal syndrome has been associated with newborns exposed to antenatal marijuana.[17] Finally, it is important to note that the marijuana consumed currently has significantly higher THC levels than the marijuana consumed decades ago when the majority of research data regarding marijuana use during pregnancy and potential fetal/neonatal impacts were reported.[17]

OPIOIDS

Newborns may have been exposed to opioids in utero either from illicit maternal use, prescribed use, or for pain management in an NICU. Exposure can result in neurobehavioral findings consistent with NAS. No clear association of a teratogenic effect has been identified with maternal use of opioids.[13]

Maternal heroin abuse is more common in women leading a risky lifestyle, who are more likely to be unmarried; unemployed; uninsured; have unplanned pregnancies; receive limited or no prenatal care; and have multiple social, nutritional, physical, and mental health problems.[20] Newborns born to women abusing heroin during pregnancy have a higher incidence of prematurity, low birth weight, and growth restriction. These infants often develop symptoms of NAS immediately after birth.[20]

Women receiving treatment for opioid addiction may be treated with a synthetic medication to decrease their cravings for an opioid and to decrease their unhealthy lifestyle choices. Synthetic medications deemed safe for use during pregnancy include methadone, buprenorphine, and Suboxone (buprenorphine and naloxone). Due to the rise of illicit opioid use during pregnancy and the need for effective treatment, in 2015, the U.S. Congress passed the Protecting Our Infants Act requiring the U.S. Department of Health and Human Services to evaluate and make recommendations regarding available treatment options for women with opioid substance use disorders during pregnancy.[21]

Women treated within a methadone maintenance program have been shown to have improved prenatal care, decreased illicit drug use, and improved fetal outcomes. Buprenorphine and Suboxone have also been shown to be safe and effective for treatment of

opioid addiction during pregnancy. However, even a newborn born to a mother successfully enrolled in an opioid addiction treatment program is at significant risk for NAS. Maternal abuse of prescription pain medications during pregnancy also increases a newborn's risk of NAS.[20]

Women with perinatal depression are at increased risk for substance use disorders. In addition, antidepressants and benzodiazepines when used as prescribed have been associated with NAS. Selective serotonin reuptake inhibitors (SSRIs), selective norepinephrine reuptake inhibitors (SNRIs), and tricyclic antidepressants (TCAs) have all been associated with NAS. These increased causes of NAS: illicit heroin and prescription opioid use; opioid treatment programs with methadone or buprenorphine; antidepressants; and benzodiazepines have complicated the diagnosis and the management of NAS.[20]

NEONATAL ABSTINENCE SYNDROME

Abrupt discontinuation of prolonged opioid exposure to a fetus or newborn may result in NAS. The withdrawal experienced by the newborn is a physiologic withdrawal and should be distinguished from an addiction. An addiction includes drug-seeking behavior. Opioid withdrawal is complex and not well understood in adults; and even more complicated in newborns due to intricate maternal–fetal–placental pharmacokinetics and immature neurologic development of the fetus/newborn. Opioids primarily act through opioid receptors located in the central nervous system, peripheral nervous system, and gastrointestinal system.[22] The lack of opioids during withdrawal causes an increase in norepinephrine production triggering the majority of clinical symptoms present in NAS.[20]

CLINICAL PRESENTATION

The clinical presentation of NAS is the result of abrupt cessation of opioids experienced by the newborn at birth. Although NAS is complex, the presentation may vary from newborn to newborn depending on the type of opioid exposure, dose and length of exposure, length of time from last exposure to presentation of symptoms, and neurologic maturity of the newborn (Table 15.3). Premature infants tend to exhibit less signs of withdrawal, which may be due to decreased amount and length of exposure as well as neurologic immaturity limiting ability to demonstrate clinical signs of NAS. In general, heroin exposure results in earlier and shorter NAS symptoms when compared to methadone or buprenorphine.[20]

Clinical signs and symptoms of NAS include tremors, jitteriness, myoclonic jerks, irritability, poor sleep patterns, excessive crying, high-pitched crying, poor

TABLE 15.3: NAS PRESENTATION ONSET AND DURATION BASED ON DRUG EXPOSURE

DRUG	ONSET IN HOURS AFTER LAST EXPOSURE	DURATION OF NAS IN DAYS
Heroin	24–48	8–10
Methadone	48–72	Up to 30 or more
Buprenorphine	36–60	Up to 28 or more
Prescription opioid medications	36–72	10–30

NAS, neonatal abstinence syndrome.

Source: Kocherlakota P. 2014. Neonatal abstinence syndrome. *Journal of Pediatrics* 134(2): e547–e561.[20]

feeding, poor weight gain, hyperphagia (excessive desire to eat), emesis, diarrhea, sneezing, mottling, tachypnea, tachycardia, temperature instability, and skin excoriation. Perianal excoriation is secondary to gastrointestinal disturbances, whereas excoriation of face and body is secondary to excessive irritation and movements. In general, term newborns, with good birth weights, exposure to more than one substance in utero, and/or cigarette exposure, present with more severe clinical symptoms of NAS with a longer duration of symptoms.[20,23] Although rarely seen today, in the 1970s seizures were reported in 2% to 11% of neonates with severe NAS.[24] In 1988, a study by Pinto and associates reported abnormal EEG findings in 30% of infants with NAS.[25] Clinical and long-term implications of seizures and abnormal EEG findings are unknown. Withdrawal signs in the neonate can mimic other serious conditions such as infection, hypoglycemia, hypocalcemia, hyperthyroidism, intracranial hemorrhage, hypoxic ischemic encephalopathy, and hyperviscosity (Table 15.4).[11] Therefore, a complete assessment and evaluation are always necessary when an infant presents with any of the findings mentioned here.

ASSESSMENT TOOLS

When NAS was first recognized, a standardized, objective, and comprehensive approach to assessment became critical to guide treatment. Assessment should be ongoing and obtained when the infant is in a low-stimulation environment using a standardized and validated tool.[24] The predominant NAS scoring tool studied for validity and reliability and utilized in the United States is the Finnegan Neonatal Abstinence Scoring System.[26] Additional tools studied and utilized include the Neonatal Drug Withdrawal Scoring System,[27] Neonatal Withdrawal Inventory,[28] Neonatal Narcotic Withdrawal Index,[29] MOTHER NAS Scale,[30] and Neonatal Network Neurobehavioral Scale.[31] In addition, there are abbreviated scoring systems developed and utilized, including the Moro Scale,[32] Measurement of Movement,[33] Three-Sign Screening Index,[34] and Finnegan Neonatal Abstinence Scale—Short Form.[35,36]

Prenatal counseling and empowerment of parent involvement along with a simplified newborn functional assessment has shown a significant reduction in both the need for newborn pharmacologic treatment

TABLE 15.4: CAUSES OF THE CLINICAL PRESENTATION OF NAS

CENTRAL NERVOUS DISTURBANCES	METABOLIC/VASOMOTOR/ RESPIRATORY DISTURBANCES	GASTROINTESTINAL DYSFUNCTION
Excessive/continuous high-pitched cry	Sweating	Excessive sucking
Poor sleep patterns/excessive wakefulness	Temperature instability/fever	Poor feeding
Hyperactive Moro reflex	Mottling	Emesis
Tremors	Yawning	Loose/watery stools
Increased muscle tone	Nasal stuffiness	Excoriation of skin
Myoclonic jerks	Sneezing	
Seizures	Nasal flaring, Tachypnea	

NAS, neonatal abstinence syndrome.

Source: Jones HE., et al. 2012. Maternal opioid treatment: Human experimental research (MOTHER) approach, issues and lessons learned. *Addiction* 107: 28–35.[30]

and hospital length of stay. The simplified functional assessment includes assessing the newborn's ability to either breastfeed successfully or take greater than or equal to 1 ounce from a bottle per feed, sleep undisturbed for greater than or equal to 1 hour, and, if crying, be consoled within 10 minutes. Effective utilization of any scoring tool or functional assessment approach includes ongoing education and training for staff and providers regarding objective scoring and interpretation.[37] Effective management of NAS inc-ludes a team approach to family-centered care, objective assessments, a supportive and low-stimulation environment, and pharmacologic treatment when indicated.

SUMMARY

Substance use disorders during pregnancy continue to be a significant health issue impacting the fetus and newborn. A thorough assessment of a newborn involves the systematic, objective evaluation for possible in utero exposure to tobacco, alcohol, marijuana, opioids, and other illicit drugs. Recognizing varying risks associated with specific substances used in utero will aid the provider in offering appropriate support and care for each newborn and family impacted by substance use disorders.

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Antepartum Tests and Intrapartum Monitoring

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A

COMMON ANTEPARTUM TESTS

Alpha-Fetoprotein Level

Alpha-fetoprotein is evaluated at 15 to 18 weeks of gestation to screen for neural tube defects, which are associated with elevated levels of this protein. Maternal serum or amniotic fluid (AF) can be used to determine levels. High levels are also associated with multiple gestation, congenital nephritis, exstrophy of the bladder, omphalocele, gastroschisis, intrauterine growth restriction, and fetal death. A lower than normal level is associated with trisomies 21, 18, and 13.

Amniocentesis

Amniocentesis is advised for mothers with known hereditary disorders or carrier states (e.g., cystic fibrosis) to identify a potential genetic/chromosomal disorder in the fetus and for those over 35 years of age to screen for chromosomal abnormalities. Pregnancies affected by Rh disease are monitored by examining the AF for optical density. A low serum alpha-fetoprotein level or positive triple screen in the mother may indicate that trisomy 21 (Down syndrome) is present in the fetus and may suggest the need for amniocentesis for chromosomal analysis. Other indications for amniocentesis for chromosomal analysis are known consanguinity in the parents and a previous child with a chromosomal or hereditary disease. Amniocentesis traditionally is performed at 18 to 20 weeks gestation, but some practitioners may offer it at 13 to 14 weeks gestation.[1,2] There is an increased risk for rupture of membranes if the test is performed before 13 weeks gestation.[2] For chromosomal analysis, the cells acquired from the AF can take from 10 to 14 days for karyotyping. At later gestational ages, amniocentesis can be used to evaluate fetal lung maturity.

Assessment of Fetal Activity

Assessment of fetal activity, or kick counts by the mother, is a noninvasive technique for monitoring fetal well-being. This is thought to be an effective method of reducing fetal stillbirth.[1] Usually, monitoring begins at 28 weeks gestation.[2,3] For this evaluation, the mother rests for 1 hour daily at the same time each day in a quiet room. During that rest period, she records the fetal movements on an activity chart. Most practitioners consider a baseline of four fetal movements per hour acceptable.[2] If fewer than four fetal movements are detected, further testing is indicated.

Biophysical Profile

The biophysical profile is a more extensive evaluation of the fetus using ultrasound and the nonstress test (NST). Scoring and recommended management approaches are explained in Tables A.1 and A.2.[4] In addition to NST, ultrasound evaluations of the AF index, fetal breathing, gross and fine movement, and tone are obtained.[1,5]

TABLE A.1: BIOPHYSICAL PROFILE SCORING: VARIABLES AND SCORING CRITERIA

BIOPHYSICAL VARIABLE	NORMAL (SCORE = 2)	ABNORMAL (SCORE = 0)
FBM	At least one episode of FBM of at least 20 seconds' duration in 30 minutes of observation	Absent FBM or no episodes of >20 seconds in 30 minutes
Gross body movements	At least two discrete body/limb movements in 30 minutes (episodes of active continuous movement are considered to be a single movement)	Less than two episodes of body/limb movements in 30 minutes
Fetal tone	At least one episode of active extension with return to flexion of fetal limb(s) or trunk; opening and closing of hand considered normal tone	Either slow extension with return to partial flexion or movement of limb in full extension or absent fetal movement or partially open fetal hand
Qualitative pockets of AF volume	At least one pocket of AF that measures at least 2 cm in vertical axis	Either no pockets or a pocket of <2 cm in vertical axis
Reactive FHR episodes	At least two episodes of FHR accelerations of at least 15 bpm and of >15 seconds' duration associated with fetal movement in 20 minutes	Fewer than two accelerations of FHR or accelerations of <15 bpm in 20 minutes

AF, amniotic fluid; bpm, beats per minute; FBM, fetal breathing movements; FHR, fetal heart rate.

Source: Adapted from Manning FA. 1999. The fetal biophysical profile. *Obstetrics and Gynecology Clinics of North America* 26(4): 560. Reprinted by permission.[4]

Chorionic Villus Sampling

Chorionic villus sampling is indicated for suspected chromosomal or biochemical defects in the fetus. It has the advantage of earlier diagnostic analysis than when amniocentesis is used. Sampling can be by the transabdominal or transcervical approach.[1,2] Chorionic tissue has the same genotype as the fetus, and testing can be performed at 10 to 12 weeks. Results are available within 24 hours because living tissue is analyzed.

Contraction Stress Test

A contraction stress test is a method of determining fetal well-being by evaluating the response of fetal heart rate (FHR) to uterine contractions. It is a nonspecific test of placental reserve. Uterine contractions normally increase FHR variability and can result in increased fetal movement. Uterine contractions are induced by oxytocin (Pitocin) or by nipple stimulation. Three contractions in a 10-minute period are evaluated. A negative test indicates the absence of late deceleration in FHR with contractions. A positive test shows late deceleration in FHR with contractions. In cases of positive stress tests and suspicious or technically poor tests, a biophysical profile may be ordered.[1,2,6,7]

Doppler Velocimetry

Doppler velocimetry evaluates blood flow in the umbilical arteries during diastole. The flow study is performed on the fetal umbilical arteries to evaluate the placental perfusion and vascular resistance of flow. Fetal deterioration and sudden critical fetal and neonatal outcome characteristics, such as growth restriction and progressively worsening placental perfusion, can be correlated with the overall measure. A decrease in diastolic flow indicates an increase in downstream placental resistance. Absent or reversed flow is an ominous sign and may indicate uteroplacental insufficiency.[1,8]

TABLE A.2: INTERPRETATION OF FETAL BIOPHYSICAL PROFILE SCORE RESULTS AND RECOMMENDED CLINICAL MANAGEMENT

SCORE	INTERPRETATION	RISK OF FETAL DEATH/1,000/WK UNDELIVERED	MANAGEMENT
10 of 10	Nonasphyxiated	0.565	Conservative
8 of 10; normal AFV	Nonasphyxiated	0.565	Conservative
8 of 8 (NST not done)	Nonasphyxiated	0.565	Conservative
8 of 10; decreased AFV	Chronic compensated asphyxia	20–30	If mature (≥37 weeks): deliver
			If immature: serial testing twice a week
6 of 10; normal AFV	Possible acute fetal asphyxia	50	If mature (≥37 weeks): deliver
			If immature: repeat test in 24 hours and deliver if score remains ≤6 of 10
6 of 10; decreased AFV	Chronic asphyxia, with possible acute asphyxia	>50	If ≥32 weeks: deliver If <32 weeks: test daily
4 of 10; normal AFV	Acute asphyxia likely	115	If ≥32 weeks: deliver If ≤32 weeks: test daily
4 of 10; decreased AFV	Chronic asphyxia, with acute likely	>115	If ≥26 weeks: deliver
2 of 10; normal AFV	Acute asphyxia almost certain	220	If ≥26 weeks: deliver
0 of 10	Gross severe asphyxia	550	If ≥26 weeks: deliver

AFV, amniotic fluid volume; NST, nonstress test.

Source: Adapted from Manning FA. 1999. Fetal biophysical profile. *Obstetrics and Gynecology Clinics of North America* 26(4): 564. Reprinted by permission.[4]

It has been estimated that the use of Doppler flow velocimetry has been associated with a 29% decrease in perinatal mortality.[1] This test is used to detect a compromised fetus in diabetic, chronic hypertensive, and preeclamptic pregnancies. Doppler velocimetry is also used to detect twin-to-twin transfusion in utero.

Fetal Fibronectin Levels

Fetal fibronectin is the "glue" that helps to attach the membranes surrounding the infant to the lower part of the uterus. Shortly before labor (or sometimes during labor), this lower part of the uterus begins to change shape, causing the membranes to separate from the uterine wall. When this happens, fibronectin is released and mingles with a woman's cervical and vaginal secretions. The fetal fibronectin test measures the absence or presence of fibronectin levels in these secretions. When a positive fetal fibronectin value is found and added to the clinical sign of cervical shortening, the combined findings are a powerful positive predictor of preterm labor and impending birth.[9]

Hemoglobin A_{1c} Levels

Hemoglobin A_{1c} ($HgbA_{1c}$) peptide levels are used for monitoring diabetic pregnancies and reflect net hyperglycemia over previous weeks. Higher first-trimester levels are associated with a greater incidence of congenital anomalies and cardiac defects and reflect poor metabolic control.[1] Tight control of blood sugar before pregnancy and in the first trimester is recommended to avoid hyperglycemic exposure of the fetus. $HgbA_{1c}$ levels should be less than 8%, and blood glucose levels should be below 150 mg/dL.[1]

Human Placental Lactogen Levels

Human placental lactogen is a hormone released by cytotrophoblasts into the maternal circulation. Production increases until approximately 37 weeks gestation and then may stabilize or decrease slightly. Low levels can indicate uteroplacental insufficiency and are seen with maternal hypertension. Serial levels are recommended. Because high placental lactogen levels are seen in multiple gestation, erythroblastosis fetalis, and poorly controlled diabetes, however, levels are not helpful indicators in women with these conditions. The normal range for human placental lactogen levels near term is 5.4 to 7 mcg/mL.[6,7]

Lecithin–Sphingomyelin Ratio

The lecithin–sphingomyelin ratio (L/S ratio) is used to determine fetal lung maturity and is done on AF. Levels of 2 or higher indicate fetal lung maturity. Falsely high levels are seen if blood, meconium, or vaginal secretions contaminate the AF. Mature L/S ratios are not always accurate in the diabetic pregnancy and in those affected by erythroblastosis fetalis. Use of a lung profile with a phosphatidylglycerol (PG) level (see next section) is more accurate in determining fetal lung maturity in these pregnancies.[1,6,7]

Lung Profile

The lung profile evaluates AF for the L/S ratio and for PG and phosphatidylinositol levels. PG appears in the AF as phosphatidylinositol begins to fall. The presence of PG indicates mature lungs and is not affected by a diabetic or erythroblastotic pregnancy. Contamination of the AF does not alter test accuracy. Some institutions use the lung profile for the L/S ratio and PG measurement only.[1,4]

A commercial test, the **AmnioStat-FLM-PG** (Irvine Scientific Products, Santa Ana, California) is available and detects PG in a sample of AF. The test takes approximately 15 minutes and is not affected by contamination with blood or meconium. A positive test indicates pulmonary maturity.

The **TDx FLx FLM II assay** (Abbott Laboratories, Abbott Park, Illinois) is an automated fetal lung maturity test that also can be performed in less than an hour. This test determines the relative concentrations of surfactant and albumin in a sample of AF. The manufacturer's recommended interpretation of the surfactant/albumin values is as follows[10,11]:

- A value less than 30 mg/g is considered definitely immature
- A value of 30 to 50 mg/g is considered transitionally immature and risky
- A value of 50 to 70 mg/g is considered transitionally mature and should be treated with caution
- A value greater than 70 mg/g indicates lung maturity

Maternal Serum Estriol Levels

Maternal serum estriol levels increase as pregnancy advances. Estriol rises rapidly until 24 weeks gestation, more slowly between 24 and 32 weeks, and then resumes a rapid rise until term. A sharp or progressive decrease indicates fetal compromise. In the past, serial assays were used to determine whether the fetus was compromised; currently, this method is seldom used. Falsely

low levels are seen with maternal corticosteroid use and with impaired maternal renal and hepatic function.

Noninvasive Prenatal Screening

Noninvasive prenatal screening (NIPS) utilizes a cell-free DNA analysis to screen for aneuploidies and specific genetic copy number variations (CNV) also known as *microdeletions*. Examples of these genetic disorders include Patau (Trisomy 13), Edwards (Trisomy 18), and Down (Trisomy 21) syndromes; Turner syndrome (XO syndrome); Klinfelter syndrome (46, XXY); Microdeletions, 22q 11.2 deletion (DiGeorge syndrome), 1p36 deletion, and 15q11.2-13 deletion.[12] This noninvasive test needs only 1 mL of the mother's blood to detect the fetal DNA, DNA fractions of greater than 4% of fetal DNA found in the mother's blood are accurate for aneuploidy detection by NIPS. The NIPS is highly sensitive and specific toward the end of the first trimester.[13]

Nonstress Test

The NST is an indirect test of placental function that assesses FHR acceleration and variability. External fetal monitoring is done, and uterine activity is monitored. Observations of the baseline FHR, accelerations, and decelerations are made. Fetal movements and changes in FHR with movement are assessed. Generally, patients are monitored for 20 to 60 minutes.[2,6] Fetal movements can be spontaneous, or they can be induced by manipulation or vibroacoustic stimulation to the mother's abdomen.[2] Accelerations in the FHR of at least 15 bpm correlate positively with fetal well-being. The results are classified as reactive, which indicates that two accelerations in FHR of at least 15 bpm occurred with fetal movement in a 20-minute period, or nonreactive, which indicates that no or fewer than two periods of FHR acceleration occurred. If test results are nonreactive, testing time may be increased, or the test may be repeated later the same day.[2] In some cases, a contraction stress test or a biophysical profile will be the next step.

Shake Test

The shake test, done on AF, is a rapid and inexpensive bedside screening test for lung maturity. Lung maturity is indicated by a complete ring of bubbles in a dilution of AF and ethanol. Persistence of an intact ring of bubbles at the air–liquid interface after 15 minutes is considered positive, indicating pulmonary maturity. Contamination of the fluid with blood or meconium results in false positives.[1]

Ultrasound

Ultrasound examination is useful in helping to determine the gestational age, for detecting altered or abnormal growth, and for identifying the presence of malformations. In the first trimester, it can be used to locate the gestational sac and the embryo or embryos and to determine crown-to-rump length. Pregnancy can be detected by ultrasound as early as 5 weeks from the last menstrual period. In the second and third trimesters, ultrasound can be used to evaluate for fetal loss; fetal age; a four-chamber heart; and abnormalities of the bladder, kidneys, brain, spine, and extremities. Placental maturity can be determined by grading the placenta from Grade 0 to Grade III. Changes in the chorionic plate, placental surface, and basal layers of the placenta can be evaluated to provide supporting information to help date the pregnancy or when deciding on the timing for cesarean section. Biparietal diameters of greater than or equal to 9.3 cm with a Grade III placenta have been consistent with a mature fetus in the absence of maternal diabetes.[14] The status of amniotic fluid volume (AFV) can be evaluated, and the umbilical cord can be inspected.[1] Hydrops fetalis can also be detected and monitored.

Increasingly, ultrasound is used in the first trimester for early detection of trisomy 21. Fetal **nuchal fold** (the space between

the back of the neck and the overlying skin) **translucency** is a test done to measure nuchal fold thickness in the fetus. A measurement over 3 mm is related to an abnormal collection of fluid in this area and is associated with trisomy 21. The combination of advanced maternal age, increased nuchal fold thickness, and elevated maternal serum levels of free beta-human chorionic gonadotropin and pregnancy-associated plasma protein are highly predictive of trisomy 21. Risk is calculated using maternal age, the crown-to-rump measurement of the baby, and the thickness of the nuchal fold. The overall Down syndrome detection rate is 71%, and the false positive rate is 2.4%.[1]

Another ultrasound finding of Down syndrome is short long bones (femur and humerus). The nuchal fold thickness is also increased in cases of Turner syndrome (karyotype XO) due to webbing of the neck, when a cystic hygroma is present in this area, and with congenital heart disorders of the great arteries.[15]

Unconjugated Estriol and Human Chorionic Gonadotropin Levels

Unconjugated estriol and human chorionic gonadotropin levels may be evaluated with the alpha-fetoprotein level. A lower than normal unconjugated estriol level with an elevated human chorionic gonadotropin level in the second trimester correlates with trisomies 21 and 18 in the fetus.[1,4] These tests, along with an ultrasound positive for increased nuchal fold translucency, are sometimes referred to as *triple markers* for trisomies 21 and 18. Use of the triple-marker tests along with the indicator of maternal age has led to a prediction rate accuracy of 60% to 90% for trisomy 21.[1,4]

INTRAPARTAL MONITORING

Cord Blood Gases

Cord blood gases are often obtained following delivery of depressed neonates and those with abnormal monitoring results during the intrapartum period. Information from the cord blood gases often aids understanding of the events surrounding the birth of a neonate with low Apgar scores. Cord blood gases are routinely obtained in many institutions.[1,15,16] Normal values are listed in Table A.3.

FHR Monitoring

FHR monitoring is commonly used during labor to identify fetal distress. The beat-to-beat FHR is recorded, with simultaneous recording of uterine activity. The normal baseline FHR is 120 to 160 bpm and should be accompanied by good baseline FHR variability. The following FHR abnormalities may occur.

Decreased or lost beat-to-beat variability can result from fetal hypoxia. It is also seen when narcotics, sedatives, or analgesic drugs have been administered to the mother. A decrease in variability can be seen with fetal sleep. Other factors that can lead to decreased

TABLE A.3: NORMAL ARTERIAL UMBILICAL BLOOD GAS VALUES FOR TERM AND PREMATURE INFANTS

VALUE	TERM (*N* = 3,520)	PREMATURE (*N* = 1,015)
pH	7.25 ± 0.07	7.28 ± 0.089
PCO_2 (mmHg)	50.30 ± 11.1	50.20 ± 12.3
HCO_3^- (mmol/L)	22.00 ± 3.6	22.40 ± 3.5
BE (mmol/L)	−2.70 ± 2.8	−2.50 ± 3.0

BE, base excess.

Source: Adapted from Riley RJ, and Johnson JWC. 1993. Collecting and analyzing cord blood gases. *Clinical Obstetrics and Gynecology* 36(1): 13. Reprinted by permission.[17]

or lost variability are administration of magnesium sulfate to the mother, fetal immaturity, and fetal tachycardia.[1,5,18]

Fetal tachycardia, indicated by a FHR greater than 160 bpm, can be associated with maternal fever, chorioamnionitis, and administration of certain medications to the mother, such as atropine, terbutaline, and others that may cause a decrease in uterine blood flow. A small percentage of fetuses whose mothers are hyperthyroid demonstrate tachycardia. Rates of 200 to 300 bpm are seen with fetal supraventricular tachycardia. Tachycardia is frequently seen in the recovery phase following fetal bradycardia related to a hypoxic episode, but can also be seen with the gradual onset of fetal hypoxemia.

Fetal bradycardia is indicated by a heart rate less than 120 bpm. It is often referred to as physiologic, not related to hypoxemia, if accompanied by good baseline variability. Some practitioners think that a baseline FHR of 90 to 120 bpm with good variability represents a normal heart rate.[1,2] Sustained low heart rates with normal variability can also be seen with fetal heart block, fetal central nervous system anomalies, and administration of beta-adrenergic receptor blocking drugs, such as propranolol, to the mother. Loss of variability with bradycardia can be associated with fetal hypoxia.

Early FHR decelerations are a benign response to uterine pressure on the fetal head. Baseline FHR and variability are normal.

Variable decelerations of FHR are caused by umbilical cord compression as uterine contraction activity peaks. Transient fetal hypoxemia and acidosis occur with compression of the arteries.

Late decelerations reflect fetal hypoxemia. Initially, the late deceleration pattern may be mediated by the vagus nerve, but with continued, prolonged hypoxia, the heart rate is low because of myocardial depression. Other factors leading to this pattern are maternal hypotension; hypercontractility of the uterus due to oxytocin; regional anesthesia; and major maternal emergencies such as hemorrhage, seizures, or respiratory arrest.[1,18]

Sinusoidal patterns of FHR appear as baseline oscillations of FHR similar to a sine wave, with fixed periodicity and loss of variability. They are associated with severe fetal anemia and are seen with Rh-sensitized fetuses and with large fetofetal transfusion; fetomaternal transfusion; and fetal hemorrhage, as into a cavernous hemangioma.[1,18]

pH of Umbilical and/or Fetal Scalp Blood

Umbilical and fetal scalp pH has been a confusing and difficult predictor of poor Apgar scores, fetal distress, and neonatal outcome. It is now known that the former cutoff for pH less than 7.2 is not an appropriate predictor.[1] Even an umbilical cord blood pH of 7.0 can have equivocal correlations with neurologic outcomes and Apgar scores. Normal mean umbilical pH at the end of labor is 7.27 to 7.28.[1]

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Glossary of Terms

Abduction: Drawing a limb or limbs away from the midline of the body or the digits away from the axial line of the limb. The lower the neonate's gestational age, the greater the amount of hip abduction seen.

Acetabulum: The cup-shaped cavity in which the femoral head articulates.

Achondroplasia: Congenitally shortened lower extremities, seen when the ratio of upper body length to lower body length exceeds 1.7:1.

Acrocyanosis: Bluish discoloration of the palms of the hands and the soles of the feet. This condition is normal in otherwise normal neonates immediately after birth, but should not persist longer than 48 hours. Also sometimes called *peripheral cyanosis.*

Active alert state: A wakeful state of infant consciousness characterized by increased motor activity and sensitivity to stimuli. The eyes are less attentive than in the quiet alert state, and periods of fussiness may be seen. In this state, preterm infants may become distressed and unable to focus.

Active sleep: A sleeping state of infant consciousness characterized by low activity but with some response to external stimuli (startle, brief crying noises). Rapid eye movements may be seen.

Adduction: Drawing a limb or limbs toward the midline of the body or the digits toward the axial line of the limb. In the neonate, hip adduction increases with gestational age.

Adventitious breath sounds: Abnormal breath sounds or sounds not normally found within the lungs.

Agenesis: Absence or failure of development of any organ or part.

Allis sign: A procedure for observing femoral length. With the infant's feet flat on the bed and the big toes and femurs aligned, flex the infant's knees. Face the feet and observe the height of the knees. A positive Allis sign is present if one knee is higher than the other.

Anal wink: Reflexive contraction of the anal sphincter in response to gentle stroking of the anal area. This reflex aids inspection of sphincter placement and tone in the newborn. Also called *anocutaneous reflex.*

Anencephaly: A defect in the newborn's skull (beginning at the vertex and extending, in some cases, to the foramen magnum) resulting from defective closure of the anterior neural tube. Hemorrhagic and fibrotic cerebral tissue protrudes, uncovered, through the defect. Generally, anencephaly involves the forebrain and some amount of the upper brainstem. The infant displays an underdeveloped cranial vault, shallow orbits, and protruding eyes. Abnormalities of other systems are also seen.

Anomaly: A deviation from normal, especially a defect of congenital or hereditary origin. A **minor anomaly** is one that is of little cosmetic significance and requires minimal or no intervention. Many deformations fall into this category. A **major anomaly** is medically or cosmetically significant and requires medical intervention. Most malformations and disruptions are categorized as major anomalies. The greater the number of minor anomalies seen in an infant, the higher the risk of a major anomaly also being present.

Anterior axillary line: A reference line (used to describe the location of physical findings)

that passes vertically through the anterior axillary fold.

Anterior vascular capsule of the lens: Within the first 24 to 48 hours of life, transient embryologic vascular systems that nourish the eye during active intrauterine growth can be seen on the anterior vascular capsule of the preterm infant's lens using an ophthalmoscope. Because these systems appear at week 27 and disappear by the end of week 34 of gestation, the degree of their presence in the neonate's lens can be used to determine gestational age.

Aphonia: Loss of the voice.

Apical impulse: The forward thrust of the left ventricle during systole, usually seen in the newborn in the fourth intercostal space at or left of the midclavicular line. An apical impulse downward and to the left suggests left ventricular dilatation. A very sharp apical impulse indicates high cardiac output or left ventricular hypertrophy.

Aplasia cutis congenita (ACC): A congenital abnormality of unknown cause, ACC most often appears on the scalp (over the parietal bones or near the sagittal suture) and is characterized by the absence of some or all layers of the skin. It may be an isolated defect or may be associated with chromosomal disorders such as trisomy 13.

Apnea: A lapse of 20 seconds or more between respiratory cycles, with bradycardia or color changes. Apnea is commonly seen in premature neonates, and is outgrown as the infant approaches term. It is abnormal in near-term or term infants.

Appropriate for gestational age (AGA): An infant whose weight, length, and/or occipital–frontal circumference (OFC) falls between the 10th and the 90th percentiles for gestational age when plotted on a standard growth chart.

Areola: The darkened area surrounding the nipple. In a full-term infant, it is raised and stippled, with 0.75 to 1 cm of palpable breast tissue. The distance between the outside of the two areolae should be less than 25% of the chest circumference.

Arnold–Chiari malformation: A congenital anomaly marked by inferior displacement of the medulla and fourth ventricle into the upper cervical canal and elongation and thinning of the upper medulla and lower pons, along with interior displacement of the lower cerebellum through the foramen magnum into the lower cervical canal. It is generally associated with hydrocephalus and myelomeningocele.

Arrhythmia: An irregularity in the heart rhythm. Arrhythmias are common—and usually benign—in the newborn. **Sinus arrhythmia** is characterized by irregularity of the R-R interval with an otherwise normal cardiac cycle. **Premature atrial beat**, seen in perhaps a third of healthy term and premature infants, is an early beat arising from a supraventricular focus, with normal ventricular conduction. **Premature ventricular beat** is an early beat arising from an irritable ventricular focus, with abnormal ventricular conduction producing a wide QRS complex.

Arthrogryposis: Congenital limitation of movement of limbs due to nonprogressive contracture of the joint.

Ascites: Accumulation of serous fluid in the peritoneal cavity.

Asphyxia, perinatal: Acidosis, hypoxia, and hypercapnia caused by lack of oxygen and carbon dioxide exchange and decreased perfusion in the fetus or the newborn. It may result in hypoxic–ischemic encephalopathy.

Association: A combination of defects that occur together more frequently than would be normal by chance, but that is less fixed than the pattern seen in a syndrome. Associations are generally named by acronyms formed from the initial letter of each defect in them.

Atelectasis: Incomplete expansion of all or part of a lung.

Atresia, anal: Congenital absence of the anal canal and/or orifice, indicated by lack of stool passage, progressive abdominal distention, and finally vomiting. Also called *imperforate anus.*

Atresia, bilateral choanal: Obstruction of the posterior nasal passages. Infants affected are cyanotic at rest and pink when crying because they are breathing through an open mouth.

Atresia, esophageal: Congenital lack of continuity of the esophagus, characterized by excessive salivation.

Auscultation: A physical assessment technique involving listening for body sounds—chiefly those of the heart and lungs, but also those of the pleura and abdomen. *Direct* auscultation is performed without the aid of a stethoscope; *indirect* (*mediate*) auscultation involves use of the stethoscope.

Avulsion: A tearing away.

Ballotable: Use of palpation to detect movement of objects suspended in fluid, such as a fetus in amniotic fluid.

Barrel chest: An abnormality of the bony structure of the chest in which the anterior–posterior diameter of the chest is greater than normal. It is seen with transient tachypnea of the newborn (TTN), meconium aspiration, and hyperinflation.

Behavioral assessment: Evaluation of the infant's ability to interact with the environment; includes assessment of organization, changes in state of consciousness, response to stimuli, and evaluation of motor maturity, and muscle tone, strength, and coordination.

Behavioral cues: Signs infants give to indicate their physical, psychological, and social needs. **Signs of attention** (also called *signs of approach*) indicate a readiness to interact. These cues include an alert, focused gaze, and regular breathing; they may also include grasping, sucking, or hand-to-mouth movements. **Time-out signals** (also called *avoidance behaviors*) indicate the need for a respite from interaction. These cues include an averted gaze, frowning, sneezing, yawning, vomiting, and hiccups, sometimes accompanied by finger splaying, arching, and stiffening. Color changes, apnea, irregular breathing, and decreased oxygen saturation levels may also be seen.

Bell palsy: Paralysis of the facial nerve with a resultant characteristic distortion of the face—unilateral.

Bones, cranial: There are four cranial bones: *frontal, occipital, parietal*, and *temporal*. See Figure 5.2 for locations.

Bowel sounds: Metallic, tinkling sounds heard every 15 to 20 seconds when the four abdominal quadrants are auscultated beginning within 15 minutes after birth. Hyperactive sounds immediately after feeding, with the intensity of the sounds diminishing as the next feeding approaches, are normal. Auscultation of *friction rubs* (resembling the sound of rubbing a finger over a cupped hand held near the ear) may indicate peritoneal irritation. When heard in the newborn over the lung fields, bowel sounds are likely referred sounds from the abdomen. If they persist, however, they could indicate diaphragmatic hernia.

Brachial plexus palsy: A functional paralysis of part or all of the arm, resulting from birth trauma and more common in large babies. It is caused by stretching injuries to the components of the brachial plexus during delivery. The upper arm is more commonly affected than the lower or the entire arm. The Moro reflex is absent on the affected side, but the grasp reflex remains. If the nerve roots are not injured, neurologic function returns within several days of birth as hemorrhage and edema resolve.

Brachycephaly: Broad skull shape caused by craniosynostosis (premature fusion) of the coronal suture, limiting forward growth of the skull.

Bradycardia, sinus: A heart rate less than 80 beats per minute in a newborn. Transient bradycardia, with origin in the sinus node, is commonly seen in both full-term and premature newborns, generally in response to vagal stimulation.

Bradypnea: Excessively slow respirations. In the neonate, persistent respirations of less than 40 per minute are associated with central nervous system depression from such factors as maternal drug ingestion, asphyxia, or birth injury.

Branchial sinus: An abnormal opening seen anywhere along the sternocleidomastoid muscle. It may communicate with deeper structures, with potential for infection.

Bronchial breath sounds: Seldom heard in the neonate, bronchial sounds are the loudest of the normal breath sounds. They are found over the trachea and are marked by a short inspiration and a longer expiration.

Bronchopulmonary dysplasia (BPD): *See* Chronic lung disease (CLD).

Bronchovesicular breath sounds: Normally found over the manubrium and the intrascapular regions, bronchovesicular sounds are intermediate in intensity between bronchial and vesicular breath sounds. In these sounds, inspiration and expiration are equal in quality, intensity, pitch, and duration.

Bruit: (a) A murmur-like sound auscultated over the fontanels or the lateral skull. It may signify intracranial arteriovenous malformation in an infant with congestive heart failure. (b) A sound, resembling a systolic murmur, heard with dilated, tortuous, or constricted vessels during auscultation of the skull. It is produced by the vibration of vessel walls within the skull, caused by flow disturbances that interfere with the normal laminar flow through the vessels. Bruits may be heard with Sturge–Weber syndrome. (c) If heard over the newborn's abdomen and not eliminated by changing the infant's position, bruits may indicate umbilical vein or hepatic vascular system abnormalities, renal artery stenosis, or hepatic hemangiomas.

Brushfield spots: White specks scattered linearly around the circumference of the iris. These spots are associated with Down syndrome, but may also be seen as a normal variant.

Bulla: An elevation of the skin filled with serous fluid (a vesicle) greater than 1 cm in diameter.

Café au lait spot (patch): A tan or light-brown macule or patch with well-defined borders. One patch is found in up to 19% of normal children, and these patches are of no pathologic significance when they are less than 3 cm in length and less than six in number. A greater number of patches or patches larger than 3 cm may indicate a cutaneous neurofibromatosis, an autosomal dominant genetic disorder.

Candida diaper dermatitis: A moist, erythematous rash caused by or associated with *Candida albicans* and consisting of small white or yellow pustules over the buttocks and perianal region (and occasionally the thighs).

Canthus: The angle at either end of the opening between the eyelids. The eye has two canthi, the inner (adjacent to the nose) and the outer.

Capillary filling time: A measurement used to assess cardiac perfusion in the newborn. Press the infant's skin with a finger until it blanches; then count the seconds required for the color to return. This should take no more than 3 to 4 seconds in a normal infant. Check filling time in both a central and a peripheral area.

Caput: Referring to the head.

Caput succedaneum: Edema occurring in and under the presenting part of the fetal scalp during delivery, generally due to pressure restricting venous and lymph flow. The edema has poorly defined edges, pits on pressure, and usually crosses suture lines. It resolves within a few days. Caput succedaneum is the most common form of birth trauma to the head.

Cephalhematoma: Collection of blood (hemorrhage) between the periosteum and the skull, generally resulting from birth trauma. The edges are clearly demarcated and bounded by suture lines. Cephalhematomas are most commonly seen over the parietal and occipital bones. Over time, a cephalhematoma may liquefy and become fluctuant on palpation; resolution can take weeks to months.

Chordee: Ventral bowing of the penis, which produces a downward curvature on erection. It is often seen with hypospadias.

Chromosomal aneuploidy: Too many or too few chromosomes, resulting in a genetic malformation. Normal infants have 46 chromosomes (22 pairs of autosomes and two sex chromosomes). Forms of aneuploidy include monosomy and trisomy.

Chronic lung disease (CLD): A chronic respiratory problem characterized by bronchiolar metaplasia and interstitial fibrosis. It affects neonates born before 30 weeks gestation who have previously received mechanical ventilation and/or supplemental oxygen. Also known as bronchopulmonary dysplasia (BPD).

Clavicle: One of the pair of bones linking the sternum with a scapula. Commonly called the *collarbone.*

Cleft lip: A congenital defect in the upper lip.

Cleidocranial dysostosis: Syndrome characterized by congenital complete or partial absence of the clavicles. Defective cranial ossification, large fontanels, and delayed suture closure are often seen with complete absence of clavicles. This anomaly is characterized by excessive scapulothoracic motion.

Clonic: Relating to or characterized by clonus.

Clonus: Involuntary repetitive muscle contraction and relaxation.

Clubfoot (*talipes equinovarus*): One of the common congenital deformities of the foot, seen more often in male infants than in female, and varying in severity. The forefoot is adducted and points medially, there is pronounced varus of the heel, and the foot and toes point downward. In unilateral presentation, the affected foot is smaller, with thickened joints.

Coloboma: A malformation characterized by absence of or a defect in the tissue of the eye. It is sometimes seen as a keyhole-shaped pupil and associated with other anomalies.

Congenital absence of the radius: A bilateral anomaly in half the infants who exhibit it, radial absence presents with a deviated hand and wrist, a shortened forearm with a bowed ulna, an absent or hypoplastic thumb, and limited elbow movement. The condition may require surgery.

Congenital absence of the tibia or fibula: This unusual anomaly more frequently affects the fibula than the tibia and is more often unilateral than bilateral. There may be partial or complete absence of the bone. Foot deformity and leg length discrepancies are generally seen in the absence of either long bone.

Congenital constricting bands (Streeter dysplasia): Bands of tissue (thought to be amniotic in origin) encircling one or more extremities of the newborn. These bands can produce shallow indentations of the soft tissue or constrictions severe enough to cause partial or complete amputation of the extremity. Treatment depends on severity; a surgical consultation may be required.

Congenital heart disease (CHD): A spectrum of cardiac anomalies present at birth. There is increased risk of CHD in infants of mothers with CHD, in infants with an older sibling who has CHD, and in low-birth-weight infants. An increased incidence of CHD is also associated with extracardiac anomalies, including those of the gastrointestinal, renal, and urogenital systems; tracheoesophageal fistulas; and diaphragmatic hernias.

Consolability: An infant's ability to quiet himself when in the crying state or to respond to attempts by a caregiver to quiet him. An infant with well-developed organization generally exhibits self-consoling behaviors, including bringing the hands to the mouth, sucking on the fist or tongue, or using environmental stimuli as a distraction.

Consolidation: An area of increased lung tissue density.

Cost of attention: A term used by T. B. Brazelton to describe the amount of energy an infant must expend to maintain an alert state of consciousness. The cost of attention depends on both the health and maturity of the infant.

Crackles: Adventitious breath sounds involving a series of brief crackling or bubbling sounds. (Crackles were previously termed *rales*.) **Fine crackles**, usually heard at the end of inspiration, resemble the sound made by rubbing a lock of hair. They may be associated with respiratory distress syndrome (RDS) or BPD. **Medium crackles** sound like carbonated fizz. They are believed to reflect the passage of air past sticky surfaces, as found with pneumonia or TTN. **Coarse crackles** (also called *rhonchi*) are loud, bubbly sounds associated with mucus or fluid accumulation in the large airways.

Craniosynostosis: Premature fusion of one or more sutures, stopping bone growth perpendicular to the suture, but permitting it parallel to the suture.

Craniotabes: Areas of soft (demineralized) bone in the skull, usually in the occipital and parietal bones along the lambdoidal suture. Craniotabes is sometimes seen with hydrocephalus.

Cremasteric reflex: Contraction of the cremaster muscle resulting in a drawing up of

the ipsilateral testis when the inner thigh is stroked or stretched longitudinally.

Cri du chat syndrome: A chromosomal disorder in which the short arm of the fifth chromosome is missing. It presents with a catlike cry in the newborn and other associated anomalies.

Crust: Dried serous exudate, blood, or pus.

Crying: A wakeful state of infant consciousness characterized by crying (in term infants), increased motor activity, color changes, and exaggerated response to unpleasant stimuli. Some term infants console themselves by returning to a lower state of consciousness. Preterm infants may have a very weak (or even absent) cry, but exhibit the other signs of stress indicative of this state.

Cryptorchidism: Failure of one or both testes to descend into the scrotum. It can be unilateral (most often occurring on the right side) or bilateral. In full-term infants who present with undescended testes at birth, about half show descent by 6 weeks of age; in 75% of preterm infants, the testes descend by 3 months of age. Testes do not usually descend spontaneously after 9 months of age. Inguinal hernia is commonly seen with cryptorchidism.

Cutis marmorata: Bluish mottling or marbling of the skin in response to chilling, stress, or overstimulation, usually disappearing when the infant is warmed.

Cyanosis, central: Bluish discoloration of the skin and mucous membranes due to significant arterial oxygen desaturation. It presents when at least 5 g of unbound (to oxygen) hemoglobin are present per 100 mL of blood. An important indicator of CHD, cyanosis due to cardiac causes does not improve on administration of 100% oxygen and also increases with crying. In cyanotic infants without increased respiratory effort, the cause of the cyanosis is most likely CHD. Central cyanosis may also occur in the presence of persistent pulmonary hypertension, lung disease, sepsis, or neurologic disease. Central cyanosis must be differentiated from acrocyanosis (peripheral cyanosis) and circumoral cyanosis, both of which are normally seen in the first few days after birth.

Cyanosis, circumoral: Bluish discoloration of the lips and area surrounding the mouth.

Darwinian tubercle: A normal variant appearing as a small nodule on the upper helix of the ear.

Deep sleep state: A sleeping state of infant consciousness characterized by closed eyes, regular breathing, and lack of spontaneous activity. Response to external stimuli is delayed.

Defect, single-gene: A genetically caused malformation. The form of inheritance may be dominant (carried by one gene from one parent), recessive (requiring one gene from each parent), or X-linked (carried on the X chromosome and expressed in male offspring, with female offspring being carriers).

Deformation: A congenital abnormality caused by pressure exerted by mechanical forces on the fetus generally late in gestation after the major organ systems are formed. The source of the pressure is generally uterine constraint (maternal or fetal). Most deformations occur to the musculoskeletal system.

Depigmentation lesion: A white, macular lesion with an irregular, leaflike border. These lesions may be early indicators of tuberous sclerosis, a progressive degenerative neurologic disease.

Dermis: Inner, vascular layer of the skin containing fibrous and elastic tissues, sweat and sebaceous glands and ducts, hair follicles and shafts, blood vessels, and nerves.

Developmental dysplasia of the hip: An abnormality in which the hip joint develops abnormally, causing instability of the hip ranging from minimal to dislocation. The causes of congenital hip dysplasia may include lack of acetabular depth, laxity of the ligaments, and/or intrauterine breech position. Congenital hip dysplasia is more often seen in Caucasian than in African American or Asian infants; it is more common in females than in males, and the left hip is affected more often than the right. In multiple births, it is more common in the first born.

A positive Allis sign may indicate congenital hip dysplasia. It is associated with other orthopedic anomalies and deformities.

Diaphragmatic hernia: Protrusion of abdominal contents through an opening in the diaphragm.

Diastasis recti: A palpable midline gap between the rectus muscles of the abdominal wall. Bulging may be seen when the infant cries. Diastasis recti is often seen in otherwise normal infants and is benign unless a hernia is present.

Diastole: Dilation of the heart (particularly of the ventricles), coincident with the interval between the second heart sound (S2) and the first heart sound (S1).

Disruption: A congenital abnormality caused by the breakdown of normal tissue in utero. Most disruptions result from amniotic bands wrapping around fetal extremities or entering the fetus' mouth.

Doll's eye maneuver: Used to evaluate movement of the eyes as the head is turned. The infant's head is gently rotated from side to side; the eyes should deviate away from the direction of rotation, that is, turning the head to the right, the eyes should deviate to the left.

Dorsiflexion: Flexion toward the back, as in flexion of the foot so that the forefoot is higher than the ankle.

Drowsiness: The transitional state of infant consciousness between sleep and wakefulness. The infant exhibits a variable activity level, with dull, heavy-lidded eyes that open and close periodically. Movements are smooth, and there is response to stimuli, but not immediately.

Dysmorphogenesis: Abnormal development of form or structure, as seen in congenital malformations. Dysmorphic features are also called anomalies.

Ecchymosis (plural: ecchymoses): A nonblanching area of subepidermal hemorrhage, initially bluish black in color, then changing to greenish brown or yellow. Ecchymoses may be seen on the labia of female infants and the scrotal area of male infants after breech deliveries.

Edema: Abnormally large amounts of fluid in the intercellular tissue spaces of the body, usually the subcutaneous tissues. Most newborns have some edema around the face and eyes due to excess fluid volume after delivery. After difficult deliveries, edema of the scalp or in dependent areas is often seen. In *pitting edema*, indentations produced by pressure remain for prolonged periods.

Ejection click: A snappy, high-frequency heart sound normal during the first 24 hours of life; after that, it is considered abnormal. If present, it is best heard just after S1 and resembles in timing, but not in quality, a widely split S1.

Embryonic period: The third to the eighth week of pregnancy, the period during which differentiation and major organ system development take place. Insults to the embryo during this period can result in major anomalies in the newborn.

Encephalocele: A restricted disorder of anterior neural tube closure producing a defect in the skull called *cranium bifidum*. In some cases, a skin-covered protrusion (sac) containing meninges and cerebrospinal fluid (and sometimes cerebral tissue) is present. In other cases, the defect appears as a small tissue-covered opening (often surrounded by tufts of abnormal hair) on the skull, with no protrusion of skull contents. The most common location for encephaloceles is the midline of the occipital bone, but they are also seen in the parietal, temporal, or frontal nasopharyngeal areas. Size varies from tiny to massive; severity depends on location, size, and the contents of the protrusion. Skull x-rays can identify the cranium bifidum, and transillumination and ultrasound help identify the contents of the protrusion.

Encephalopathy, hypoxic–ischemic: A syndrome resulting from perinatal asphyxia. It can be categorized as **mild** (associated with irritability, jitteriness, and hyperalertness), **moderate** (characterized by lethargy, hypotonia, decreased spontaneous movement, and seizures), or **severe** (characterized by coma, flaccidity, disturbed brainstem function, and seizures). The pattern of limb weakness displayed by an affected infant reflects the area of the brain injured by the asphyxia. In term neonates, injury to the parasagittal areas (resulting in weakness

of the shoulder girdle and proximal upper extremities) or the middle cerebral artery (presenting as hemiparesis) predominates. In the premature newborn, injury is often to the periventricular area, resulting in weakness in the lower extremities.

Epicanthal fold: A vertical fold of skin on either side of the nose that covers the lacrimal caruncle. These folds are normal in Asian infants but suggestive of Down syndrome in neonates of other races.

Epidermis: Outermost layer of the skin, itself composed of five layers. The top layer is the *stratum corneum* (dead cells that are constantly being sloughed off and replaced); lower layers contain keratin-forming cells and melanocytes.

Epispadias: Location of the urethral meatus on the *dorsal* surface of the penis. It is a less common abnormality than hypospadias. In **balanic epispadias**, the meatus is found at the base of the balanus (glans); in **penile epispadias**, it is on the penile shaft; and in **penopubic epispadias**, it is found, not on the penis at all, but directly below the symphysis pubis.

Epstein pearls: Epidermal inclusion cysts seen in the newborn at the junction of the hard and soft palates of the mouth and on the gums. They usually disappear by a few weeks of age.

Erb palsy: Damage (generally due to birth trauma) to the upper spinal roots C5 and C6, resulting in paralysis of an arm (but usually without involvement of the hand). *See also* **Brachial plexus palsy**.

Erythema toxicum neonatorum: A benign rash consisting of small white or yellow papules or vesicles with an erythematous base. Seen in up to 70% of term infants between 24 and 48 hours of age, the rash occurs most often on the face, trunk, or extremities.

Everted: Turned out and away from the midline of the body.

Expected date of confinement (EDC): Delivery due date. Generally 280 days (40 weeks) from the onset of the mother's last menstrual period.

Exophthalmos: synonym Proptosis. Abnormal anterior displacement (protrusion) of the eye. It may be associated with hyperthyroidism.

Exstrophy of the bladder: A visible fissure between the anterior abdominal wall and the urinary bladder. In male infants, it is commonly accompanied by epispadias.

Extension: The straightening of a limb at a joint.

Familial traits: Unusual features (subtle or significant) identified by a family as characteristic of its phenotype.

Fasciculation: The spontaneous localized contraction of a group of fibers in a motor unit, visible through the skin. In Werdnig–Hoffmann disease, these continuous and rapid twitching movements can be seen by observing the tongue.

Fetal period: The ninth week of pregnancy through birth. The fetus becomes more and more resistant to teratogens as this period progresses.

Fibrils: Tiny fibers or filaments connecting the dermis and the epidermis.

Fistula, rectourethral: An abnormal connection between the rectum and the urethra, indicated by the presence of meconium in the urethral orifice.

Fistula, rectovaginal: An abnormal connection between the rectum and the vagina, indicated by the presence of meconium in the vaginal orifice.

Flexion: The act of bending or the condition of being bent. The bending of a limb at a joint. In the neonate, the degree of arm, knee, and hip flexion is indicative of gestational age.

Flexion contracture: Resistance of a muscle to passive flexion. The newborn's hips generally have a flexion contracture; when the right hip is flexed to stabilize the pelvis and flatten the lumbar spine, a flexion contracture of the left hip may be seen. The infant's elbow also shows a flexion contracture for the first few weeks of life, making it difficult to examine.

Fontanel: A membrane-covered space (soft spot) in the infant's skull reflecting incomplete ossification of the skull. Fontanels occur where two sutures meet. The **anterior fontanel** occurs at the intersection of the metopic, coronal, and sagittal sutures. The **posterior fontanel** occurs where the sagittal and lambdoidal sutures meet. The **sphenoid fontanel** is at the juncture of the coronal and squamosal sutures. The **mastoid fontanel** occurs at the intersection

of the squamosal and lambdoidal sutures. A defect of the parietal bone along the sagittal suture may appear to be—but is not—a true fontanel (third fontanel). It may be a normal variant or may be associated with Down syndrome or congenital hypothyroidism.

Forcep marks: Red, bruised, or abraded areas on the cheeks, scalp, and face of infants born after application of forceps. Observation of forcep marks calls for examination for facial palsy, fractured clavicles, skull fractures, or other complications of birth trauma.

Frog leg position: A resting posture, normal in premature neonates of 36 or fewer weeks gestation, in which the legs are abducted and the lateral thigh rests against the bed surface. This position is abnormal if seen in newborns older than 36 weeks gestational age.

Funnel chest (pectus excavatum): An abnormality of the bony structure of the chest in which the sternum is indented. It is seen in infants with Marfan syndrome and rickets.

Gastroschisis: Protrusion of abdominal contents and other organs through an abdominal wall defect lateral to the midline; the protrusion is not covered by a membrane. A gastroschisis is associated with fewer congenital malformations than is an omphalocele.

Gestational age: The length of time between fertilization of the egg and birth of the infant.

Glabella: The area above the nose and between the eyebrows (bridge of the nose).

Growth parameters: Measurements used to determine whether an infant is small, appropriate, or large for gestational age (LGA). The three parameters are weight, length, and occipital–frontal head circumference.

Habituation: A newborn's ability to alter his response to a repeated stimulus, decreasing and finally eliminating the response on repetition of the stimulus. Habituation is a defense mechanism.

Hamstring: In human anatomy, the **hamstring** is any one of a group of six tendons contracted by three posterior thigh muscles (semitendinosus, semimembranosus, and biceps femoris). The term is sometimes used to refer to the muscles themselves.

Harlequin sign: A sharply demarcated red color in the dependent half of the body, with the superior half of the body appearing pale, when the infant is placed on his side. Rotating the infant to the other side reverses the coloring. Harlequin color change occurs in both healthy and sick infants but is more common in low-birth-weight infants. It has no pathologic significance.

Heart rate: The normal heart rate for a full-term neonate is 80 to 160 beats per minute.

Heart sounds: **S1**, the first heart sound, is best heard at the mitral or tricuspid area. It is usually loud at birth, but decreases in intensity by 2 days of life. **S2**, the second heart sound, is best heard at the aortic or pulmonic area. It is usually single at birth but is split in two thirds of infants by 16 hours of age and in 80% by 48 hours of age. Wide splitting of S2 is abnormal. **S3**, commonly heard in premature infants with a patent ductus arteriosus (PDA), is best heard at the apex of the heart during early diastole. **S4**, a pathologic sound, is heard at the apex of the heart in infants with conditions characterized by decreased compliance.

Heave: A point of maximum impulse that is slow rising and diffuse, generally associated with ventricular dilation or volume overload. Also called a *lift*.

Helix: The upper and outer margin of the pinna of the ear.

Hemangioma, cavernous: A raised, lobulated, soft and compressible bluish-red tumor, with poorly defined margins. The cavernous hemangioma consists of large, mature vascular elements lined with endothelial cells and involves the dermis and subcutaneous tissues of the skin. It increases in size during the first 6 to 12 months of life, and then involutes spontaneously. Two syndromes—Kasabach–Merritt and Klippel–Trenaunay–Weber—may be associated with cavernous hemangiomas.

Hemangioma, infantile: A raised, lobulated, soft and compressible bright-red tumor, with sharply demarcated margins, on the head, neck, trunk, or extremities. (Tumors occurring in the throat can obstruct the airway.) Caused by dilated capillaries, with associated endothelial proliferation in the dermal and subdermal layers of the skin, the

infantile hemangioma occurs in up to 10% of newborns. It gradually increases in size for about 6 months and then spontaneously regresses over a period of up to several years.

Hemiparesis: Muscle weakness or partial paralysis affecting one half of the body.

Hepatomegaly: Enlargement of the liver.

Hermaphroditism: Anomalous differentiation of the gonads, with presence of both ovarian and testicular tissue and ambiguous morphologic sex criteria. Hermaphroditism is suspected when only one testis is confirmed in a male-appearing infant.

Hernia, epigastric: Protrusion of fat through a defect on the midline above the umbilicus and below the sternum, presenting as a firm palpable mass. Palpation may elicit a pain response. Surgical intervention is necessary.

Hernia, femoral: A small bulge adjacent and medial to the femoral artery. It is more common in females than in males.

Hernia, inguinal: Muscle wall defect in the inguinal area through which bowel loops or gonads enter the scrotal sac (in males) or the soft tissues (in females). The hernia usually presents as bulging in the groin area and is more common in male than in female infants.

Hernia, umbilical: Skin- and subcutaneous-tissue-covered protrusion of part of the intestine at the umbilicus. It is seen with some frequency in low-birth-weight and African American male infants and in infants of other races with hypothyroidism. The hernia usually reduces by 2 years of age, but intervention is required with strangulation of abdominal contents or large size.

Holosystolic: Occurring throughout systole. Also called *pansystolic.*

Hydranencephaly: Partial or complete absence of the cerebral hemispheres, with the space being filled with cerebrospinal fluid.

Hydrocele: A circumscribed collection of clear fluid in the scrotum, which may resolve spontaneously by 6 months of age. A hydrocele may communicate with the inguinal canal, however, and may be associated with a hernia. **Hydrocele of the cord** presents as a palpable sausage-shaped, smooth bulge above the testes.

Hydrocephalus: Accumulation of cerebrospinal fluid (often under increased pressure) within the skull, due to obstruction of the cerebrospinal fluid pathways causing dilation of the cerebral ventricles. It may be congenital or may develop after birth and is characterized by head enlargement, forehead prominence, brain atrophy, mental deterioration, and convulsions. The skull is often globular in shape.

Hydrometrocolpos: Collection of secretions in the vagina and uterus due to an imperforate hymen. It may be palpated as a small midline lower abdominal mass or a cystic movable mass as the quantity of secretions increases.

Hygroma, cystic: The most commonly seen neck mass in newborns, a cystic hygroma is soft, fluctuant, and can range from only a few centimeters in size to massive. It is usually seen laterally or over the clavicle and transilluminates well.

Hymenal tag: A small appendage or flap on the hymen, normal in female infants. It disappears in several weeks.

Hyperplasia, sebaceous gland: Numerous white or yellow papules (<0.5 mm in size) on the nose and upper lips. These enlarged sebaceous glands spontaneously decrease in size with age and require no treatment.

Hypertelorism: Widely spaced eyes—those with greater than a palpebral fissure length between them.

Hypertonia: Increased resistance of the skeletal muscles to passive stretching. In the presence of hypertonia, passive manipulation of the limbs often increases the tone.

Hypertrophy: Enlargement or overgrowth of an organ or part.

Hypertrophy of the clitoris: The most common genitourinary abnormality in female infants, suggesting pseudohermaphroditism.

Hypoplastic nails: Nails that are incompletely developed. Edema of the hands can give the appearance of hypoplastic nails.

Hypospadias: Location of the urethral meatus on the **ventral** surface of the penis. In **balanic (glanular) hypospadias**, the meatus is located at the base of the balanus (glans). In **penile hypospadias**, the meatus is found between the glans and the scrotum. It is often accompanied by chordee, flattening of the glans, and an absent

ventral foreskin. In **penoscrotal (perineal) hypospadias**, the meatus is found at the penoscrotal junction. Gender ambiguity is associated with penoscrotal hypospadias, as is bifid scrotum, small penis with a large meatus, and undescended testes.

Hypotelorism: Closely spaced eyes—those with less than a palpebral fissure length between them.

Hypotonia: Depressed postural muscle tone.

Inner canthal distance: The measurement between the inner canthi of the two eyes. If the eyes are normally spaced, this distance should be equal to the length of the palpebral fissure.

Inspection: A physical assessment technique involving careful visual attention to, measurement of, and notation of the status of external body parts and systems, for the purpose of forming a judgment.

Intrauterine growth restricted (IUGR) neonate: A newborn who did not grow in utero at the expected rate for weight, length, or occipital–frontal head circumference. Although the term is often used synonymously with *small for gestational age (SGA)*, the two terms do not necessarily mean the same thing. A neonate may be growth restricted but may not fall below the 10th percentile for gestational age.

Inverted: Turned inward toward the midline of the body.

Jaundice: Yellow coloring of the skin and whites of the eyes, generally appearing first on the head and face and then progressing toward the feet. Jaundice is due to excess bilirubin in the blood (hyperbilirubinemia).

Jitteriness: A series of movements seen at times in normal preterm neonates and in normal term neonates with vigorous crying. (Persistent jitteriness may indicate a disorder.) Jitteriness is characterized by rapid movements of equal amplitude in alternating directions, generally occurs in response to a stimulus, can be stopped by flexing and holding the involved extremity, and is not associated with abnormal eye movements or other signs of seizure. Jitteriness must be distinguished from seizure activity. *See also* **Seizure**.

Karyotype: Representation by diagram of the chromosomal characteristics of an individual or species.

Karyotyping: Chromosomal evaluation.

Keratin: An insoluble, fibrous protein that is the main component of certain protective and/or supportive elements of the body, including the nails, hair, and epidermis.

Klippel–Feil syndrome: Defects of the cervical vertebrae, resulting in a shorter-than-normal neck with limited motion and asymmetry in rotation and lateral flexion.

Klumpke palsy: Damage (generally due to birth trauma) resulting in paralysis of the forearm. *See also* **Brachial plexus palsy**.

Kyphosis: A spinal abnormality in which the curvature of the thoracic spine is excessively convex. Commonly called *hunchback.*

Lacrimal caruncle: The red area at the inner canthus of the eye.

Lanugo: A fine, soft, downy hair covering the fetus in utero and sometimes the neonate. It first appears at about 20 weeks gestation, covering most of the body including the face. Most of it disappears by 40 weeks gestation.

Large for gestational age (LGA): An infant whose weight falls above the 90th percentile for gestational age when plotted on a standard growth chart.

Lesion: Any discontinuity of tissue or change in the structure or function of an organ or part due to disease or trauma.

Leukoplakia: A white spot or patch on the tongue or mucous membranes in the mouth, which cannot be wiped off and that does not clinically represent any other condition.

Light sleep state: See **Active sleep.**

Lipoma: A benign tumor composed of fat cells.

Lordosis: A spinal abnormality in which the curvature of the lumbar and cervical spine is excessively concave. Commonly called *swayback.*

Low birth weight: An infant who weighs less than 2,500 g at birth.

Macrocephaly: Excessive head size (an occipital–frontal circumference above the 90th percentile for gestational age, with otherwise normal weight and length percentiles). Macrocephaly can be familial or it may be due to hydrocephalus or

associated with dwarfism or osteogenesis imperfecta.

Macrodactyly: An enlarged finger or toe. It is seen in otherwise normal infants, or it may indicate neurofibromatosis.

Macroglossia: An abnormally large tongue.

Macrostomia: An abnormally large oral opening.

Macule: A discolored, flat, nonpalpable spot or patch less than 1 cm in diameter.

Malacia: Abnormal softening of tissues.

Malformation: A congenital defect arising from abnormal tissue. The cause of the tissue abnormality may be intrinsic (genetic) or extrinsic (environmental).

Manubrium: Uppermost portion of the sternum.

Mediastinum: A space within the chest cavity containing the heart, esophagus, trachea, mainstem bronchi, thymus, and major blood vessels.

Melanosis, transient neonatal pustular: Small pigmented macules (often surrounded by very fine white scales) caused by the rupture of superficial vesiculopustular lesions 12 to 48 hours after birth. The condition—seen in up to 5% of African American and about 0.2% of Caucasian infants—is benign and generally resolves by 3 months of age.

Meningocele: A lesion associated with spina bifida in which more than one vertebra is involved. The meninges protrude through the bony defect and are covered by a thin atrophic skin. The spinal roots and nerves are normal.

Metatarsus adductus: A common congenital foot anomaly caused by intrauterine positioning. It may be positional (flexible) or structural (fixed). In the structural type (with bony abnormalities), there is limited abduction of the forefoot, and the heel is in a valgus position. In the positional type, the forefoot abducts easily, and the heel is in a varus or neutral position.

Microcephaly: Abnormal smallness of the head (an OFC below the 10th percentile for gestational age), generally due to poor brain growth.

Micrognathia: An abnormally small lower jaw, seen in Robin sequence and other syndromes.

Micropenis: Abnormally small penis. Micropenis suggests congenital hypopituitarism when seen with certain other anomalies or conditions. Also called *penile hypoplasia*.

Microstomia: An abnormally small oral opening. It may be seen with some trisomies.

Midclavicular line: A reference line (used to describe the location of physical findings) passing vertically through the clavicle.

Midsternal line: A reference line (used to describe the location of physical findings) bisecting the suprasternal notch.

Milia: Multiple yellow or pearly white papules about 1 mm in size. These epidermal cysts, found on the brow, cheeks, and nose of up to 40% of newborns, are caused by accumulation of sebaceous gland secretions and spontaneously resolve during the first few weeks of life.

Miliaria: Changes in the skin caused by obstruction of the sweat ducts. Miliaria is generally associated with excessive warmth and/or humidity and is classified into four types, based on severity. **Miliaria crystallina** occurs when sweat escapes into the epidermal stratum corneum, causing formation of clear, thin vesicles 1 to 2 mm in diameter. **Miliaria rubra**, commonly called *prickly heat*, presents as small erythematous papules and occurs when continued obstruction of the sweat ducts causes release of sweat into adjacent tissues of the epidermis. **Miliaria pustulosa** occurs with continued occlusion of the sweat ducts, as leukocytes infiltrate the vesicles. Unresolved miliaria pustulosa can lead to **miliaria profunda**, secondary infection of the deeper dermal portions of the sweat glands.

Mongolian spot: A large gray or blue-green macule or patch caused by melanocyte infiltration of the dermis. This pigmented lesion, generally found on the buttocks, flanks, or shoulders, is seen in up to 90% of African American, Asian, and Hispanic and up to 10% of Caucasian infants.

Morphogenesis: Differentiation of cells and tissues in the early embryo that establishes

the form and structure of the various organs and parts of the body.

Mottling: Blotchy skin showing areas of different color. It may indicate cardiogenic shock in the neonate.

Mucocele: A mucous retention cyst (distended cavity containing mucus) presenting as a translucent or bluish swelling under the tongue.

Murmur: A prolonged heart sound caused by turbulent blood flow. Documenting the timing, location, intensity, radiation, quality, and pitch are all important to the evaluation of the origin of a murmur, as is the age of the newborn. There are two types of murmurs: *innocent* and *pathologic*; see **Murmurs, innocent** and **Murmurs, pathologic**.

Murmur, continuous: A pathologic murmur, heard through both systole and diastole, present in about a third of premature infants with a PDA and also in full-term or premature infants with arteriovenous fistulas.

Murmur, continuous systolic (crescendo systolic): An innocent murmur heard in up to 15% of newborns, beginning within the first 8 hours of life. It is best heard in the upper left sternal border.

Murmur, early soft midsystolic ejection: An innocent murmur often heard in premature infants, beginning within the first or second week of life and disappearing by the eighth week. Medium to high pitched, it is best heard in the upper left sternal border, with wide radiation.

Murmur, loud systolic ejection: A pathologic murmur that presents within hours of birth, generally due to aortic or pulmonary stenosis or coarctation of the aorta.

Murmur, systolic ejection: An innocent murmur heard in more than half of all newborns, beginning within the first day of life and lasting up to a week. It can be described as "vibratory" and is best heard along the mid- and upper left sternal border.

Murmurs, innocent: Innocent murmurs have no pathologic cause within the heart or great vessels. They are generally systolic, symptomless, and common in normal newborns during the first 48 hours of life when they are associated with decreasing pulmonary vascular resistance and with closure of the PDA. They include **systolic ejection murmurs, continuous systolic (crescendo systolic) murmurs**, and **early soft midsystolic ejection murmurs**.

Murmurs, pathologic: Pathologic murmurs are due to cardiovascular disease. When they present is dependent on their cause and on normal changes associated with transitional circulation. Many do not appear until pulmonary vascular resistance falls. Absence of a murmur does not rule out CHD; as many as a fifth of the infants who die from CHD by 1 month of age do not have murmurs. Pathologic murmurs include **loud systolic ejection murmurs** and **continuous murmurs**.

Muscle tone, phasic: The brief, forceful contraction of a muscle in response to a short duration, high-amplitude stretch. In the newborn, phasic tone is evaluated by testing the resistance of the extremities to movement (scarf sign and arm and leg recoil) and by assessing the activity of the deep tendon reflexes (biceps and patellar reflexes).

Muscle tone, postural: The long-duration, low-amplitude stretch of a muscle in response to gravity. In the newborn, the traction response (pull-to-sit maneuver) is used to evaluate postural tone. Depressed postural tone is called *hypotonia.*

Myelomeningocele: A lesion associated with spina bifida in which bilateral broadening of the vertebrae or absence of the vertebral arches is seen. The lesion is most often seen in the lumbar spine and is characterized by protrusion of meninges and spinal roots and nerves, along with fusion of remnants of the spinal cord and an exposed neural tube. The higher the defect on the spine, the greater the degree of paralysis. Hydrocephalus is frequently seen with a myelomeningocele, especially in the Arnold–Chiari malformation.

Necrosis, subcutaneous fat: A hard, nonpitting, sharply circumscribed subcutaneous nodule that appears during the first weeks of life, grows larger over several days, and then resolves spontaneously

over several weeks. Color will vary. Possible causes include trauma, cold, or asphyxia. Intervention may be necessary if the condition is associated with hypercalcemia.

Neonatal Behavioral Assessment Scale (NBAS): An examination tool developed by T. B. Brazelton to assess behavior in the term infant.

Neonatal torsion of the testis: A condition in which a testis and its tunica vaginalis rotate in the scrotal sac, inguinal canal, or abdomen. Torsion of a descended testis presents with reddish to bluish coloring of the scrotal skin and a nontender or mildly tender mass. Torsion is difficult to identify if the testis has not descended.

Nevus, pigmented: A dark-brown or black macule commonly seen on the lower back or buttocks. Although pigmented nevi are generally benign, malignant changes occur in up to 10% of these lesions.

Nevus, port wine: A flat, nonblanching pink or reddish-purple lesion with sharply delineated edges directly beneath the epidermis. The port wine nevus generally appears on the face, consists of dilated capillaries, and varies in size from small to covering almost half the body. Although usually unilateral, on occasion it crosses the midline. It neither grows in size nor resolves spontaneously. Port wine nevi located over the branches of the trigeminal nerve may be associated with Sturge–Weber syndrome.

Nevus, sebaceous: A small yellow or yellowish-orange papule or plaque consisting of immature hair follicles and sebaceous glands. It is commonly found on the scalp or face.

Nevus simplex: An irregularly bordered pink macule that blanches with pressure and becomes more prominent with crying. Found most often on the nape of the neck, the upper eyelids, the bridge of the nose, or the upper lip, the nevus simplex is composed of dilated capillaries and is seen in up to 50% of newborns. It is commonly called *stork bite.*

Nipple line: A reference line (used to describe the location of physical findings) passing horizontally through the nipples.

Nipples, supernumerary: Accessory nipples—raised or pigmented areas 5 to 6 cm below the normal nipples. In Caucasian infants, they may be associated with congenital anomalies.

Nodule: An elevated palpable lesion, solid and circumscribed (a papule), greater than 1 cm in diameter.

Normal variants: Minor differences in the appearance of the features between races, but that are normal for the race. Also called *minor variants*.

Nuchal cord: An umbilical cord that has become wrapped around the fetus's neck.

Nystagmus: A rapid, searching movement of the eyeballs seen in some newborns until 3 to 4 months of age. Occasional nystagmus is normal in the otherwise normal neonate; persistent is abnormal and may indicate loss of vision.

Observation: A physical assessment technique involving careful visual attention to, and notation of the status of external body parts and systems, for the purpose of forming a judgment. Also called *inspection.*

Occipital–frontal circumference (OFC): Measurement of the neonate's head taken over the occipital, parietal, and frontal prominences, avoiding the ears. This growth parameter is used, with the infant's gestational age, to determine whether the infant's head is small, appropriate, or LGA. In the term infant, the normal OFC is 31 to 38 cm.

Oligohydramnios: The presence of less than 500 mL of amniotic fluid at term or 50% of normal volume at any time during pregnancy. The intrauterine compression caused by the lack of fluid may produce unusual flattening of the facial features in the newborn.

Omphalocele: Protrusion of abdominal contents through a defect at the umbilicus. A thin, translucent membrane covers the protrusion, and may rupture at delivery. Omphalocele is seen more often in male infants, especially premature males, and is usually associated with trisomies 13, 18, and 21; Beckwith–Wiedemann syndrome; or CHD.

Omphalomesenteric duct: Persistence in the newborn of this embryological duct between the ileum and the umbilicus, with seepage of ileal liquid from the opening.

Ophthalmoscope: A hand-held device, containing lenses and a mirror, that produces a beam of light for inspection of the interior of the eye. In physical examination of the newborn, it is used to assess pupillary constriction and the red reflex. It can also be used to inspect the anterior vascular capsule of the lens to determine gestational age.

Opisthotonus: Marked extensor hypertonia, with arching of the back. It may be seen with bacterial meningitis, severe hypoxic-ischemic brain damage, and massive intraventricular hemorrhage.

Orchiopexy: Surgical fixation of an undescended testis in the scrotum. Generally done between 9 and 18 months of age.

Organization: An infant's ability to integrate physiologic (heart rate, respiratory rate, oxygen consumption, digestion) and behavioral (state, motor activity) systems in response to the environment. Preterm infants show decreased organizational abilities.

Ortolani maneuver: A procedure for evaluating hip stability in the newborn. After flexing the infant's knee and hip, grasp the thigh and first abduct with a lifting motion and then adduct the leg. If a clunk is felt as the femoral head passes over the acetabulum, the infant may have congenital hip dysplasia.

Osteoblast: A cell that arises from a mesenchymal stem cell and works with other osteoblasts to synthesize bone.

Otoscope: A device used to inspect the ears. Due to the presence of vernix in the neonate's ear canal, the device is normally not used in newborn examinations.

Outer canthal distance: The measurement between the outer canthi of the two eyes.

Pallor: Paleness of the skin in the newborn may indicate compromised cardiac status. The paleness may be due to vasoconstriction and to shunting of blood away from the skin to vital organs or anemia.

Palpation: A physical assessment technique involving application of pressure to the skin with the fingertips or part of the palm to assess the condition of underlying body parts.

Palpebral fissure: The eye opening.

Papule: An elevated palpable lesion, solid and circumscribed, less than 1 cm in diameter.

Patch: A discolored, flat, nonpalpable spot (a macule) greater than 1 cm in diameter.

Patent ductus arteriosus (PDA): A cardiac abnormality marked by failure of the ductus arteriosus to close after birth. In a left-to-right shunt, blood flows from the aorta to the pulmonary artery, resulting in recirculation of arterial blood through the lungs.

Patent urachus: Persistence of an embryological communication between the urinary bladder and the umbilicus, with passage of urine through the umbilicus.

Percussion, direct: A physical assessment technique involving striking the body surface sharply and listening to the sound produced, to determine the condition of an underlying body part. In *indirect* (*mediate*) percussion, the examiner places the middle finger of the left hand on the area to be assessed and then strikes the finger with the middle finger of the other hand. Although not generally employed in examination of the abdomen, percussion can distinguish areas of tympanic (resonant or bell-like) sound from areas of dull sound. Tympanic sounds are present over the organs that contain air; increased tympany suggests abnormal amounts of air. Dull sounds are percussed over the liver, spleen, and bladder. Extended areas of dullness suggest organ enlargement; dullness in unusual areas may indicate masses. Shifting dullness is heard with ascites. It is of limited value in examination of the neonate's chest due to its small size, but can be useful in distinguishing among air, fluid, and solid tissue in some situations. Changes in resonance indicate changes in the consistency of the underlying tissue.

Perfusion, peripheral: Cardiac perfusion to the skin.

Perinatal history: Record of occurrences in the life of the mother, the fetus, and the newborn.

Perinatal period: Definitions of the length of the perinatal period vary; it extends from about week 20 or 28 of gestation until 1 week to 1 month after birth.

Perineum: Area between the scrotum and anus in the male and the vulva and anus in the female. It should be smooth in newborns. Dimpling suggests genetic anomalies or fistulas.

Periodic breathing: A series of respirations followed by a pause of up to 20 seconds.

Periosteum: Fibrous membrane covering the bones of the skull.

PERL: A notation that means "pupils equal and reactive to light."

Petechia (plural: petechiae): A pinpoint-sized hemorrhagic spot on the skin.

Phimosis: An irretractable foreskin. It may not be diagnosed until the infant reaches 3 months of age.

Pigeon chest: An abnormality of the bony structure of the chest in which the sternum protrudes. It is seen in infants with Marfan syndrome and rickets.

Pinna: The part of the ear that projects from the head. Formation, amount of cartilage present, and recoil when folded and released are considered indicators of gestational age.

Pit, ear: A slight depression anterior to the tragus. It may lead to a congenital preauricular sinus or fistula. Ear pits may be familial or associated with other anomalies.

Plagiocephaly: Asymmetrical skull shape caused by craniosynostosis (premature fusion) of the sutures on one side of the skull.

Plantar flexion: Flexing the foot toward the plantar surface.

Plantar surface: The sole of the foot. Creases appear on the plantar surface of the foot between 28 and 30 weeks gestation and cover the surface at or near term. Therefore, plantar surface creasing is one indicator of gestational age.

Plaque: A fusion or coalescence of several papules (solid, circumscribed, elevated palpable lesions <1 cm in diameter).

Plethora: Ruddy appearance of the skin in a newborn. Plethora may indicate a high level of red blood cells to blood volume.

Pleural cavities: Two potential spaces (left and right) within the chest cavity enclosed by a serous membrane called the pleura. A portion of the pleura called the **parietal pleura** lines the walls of the thoracic cavity; the portion called the **visceral pleura** envelops the lungs.

Pneumothorax: Accumulation of air or gas in the pleural space.

Polycythemia: A condition in which the infant's central hematocrit is greater than 65. A polycythemic infant is pink at rest but plethoric to purplish when crying. The ruddy coloring can sometimes be mistaken for cyanosis (because the unsaturated hemoglobin in the blood of the infant may mask the saturated hemoglobin, making the infant appear purplish).

Polydactyly: A congenital anomaly marked by one or more extra digits on the hands or feet. There appears to be a familial tendency to polydactyly, and it is more common in African American infants than in those of other races. The extra digit may be only a skin tag or a floppy appendage or it may appear normal.

Ponderal index: Cube root of body weight times 100 divided by height in centimeters.

Popliteal angle: The angle between the lower leg and the thigh posterior to the knee. The popliteal angle decreases with advancing gestational age.

Positional deformity: A flexible deformity of an extremity with no bony abnormality involved. It is caused by intrauterine positioning and generally corrects without treatment.

Postterm: An infant born at 42 or more weeks gestation.

Precordium: The area of the anterior chest over the heart. In the full-term newborn after the first few hours of life, the precordium should be quiet; a bounding precordium is characteristic of left-to-right shunt lesions (PDA or ventricular septal defect). In premature infants, the precordium is more active due to decreased subcutaneous tissue.

Preterm: An infant born at less than 38 weeks gestation.

Priapism: Constant erection of the penis, an abnormal finding that may indicate a spinal cord lesion.

Pronation: The turning of the body or a body part face down, as of the hand so that the palm is facing down.

Prone: Positioned face down.

Proptosis: Abnormal protrusion or bulging of the eyeball. Also called *exophthalmos.*

Prune belly syndrome: A rare congenital deficiency of the abdominal muscles marked by a protruding, thin-walled abdomen covered with wrinkled skin. It occurs more frequently in males and is associated with renal and urinary tract abnormalities.

Pseudostrabismus: The appearance of strabismus (crossed eyes) due to a flat nasal bridge or the presence of epicanthal folds.

Ptosis: Paralytic drooping of the eyelid in which the upper lid droops when the eyes are fully open.

Pulse deficit: A difference between the heart rate counted by a peripheral pulse and that counted by auscultation. A pulse deficit is frequently seen with ectopic rhythms.

Pulse pressure: The difference between the systolic and the diastolic blood pressure. For term infants, the average difference is 25 to 30 mmHg; for premature infants, it is 15 to 25 mmHg. Wide pulse pressures can indicate large aortic runoff, as in PDA. Narrow pulse pressures are seen with peripheral vasoconstriction, heart failure, and low cardiac output.

Pupillary reflex: Contraction of the pupil when the eye is exposed to bright light.

Pustule: An elevation of the skin filled with cloudy or purulent fluid.

Q-switched ruby laser: Laser used in removal of nevi or tattoos. Emits a short pulse of intense red light that is selectively absorbed by melanin located in the epidermal and dermal skin layers. The laser energy causes the melanin to break down into smaller pigment particles that are removed by the body's immune system.

Quiet alert state: A wakeful state of infant consciousness characterized by interaction with the environment. The infant focuses on stimuli and presents an alert appearance, especially in the eyes. Motor activity is minimal. In this state, preterm infants may show hyperalertness (inability to terminate fixation on a stimulus).

Rachitic rosary: In a newborn with rickets, a series of small lumps can be felt along the edge of the sternum during palpation. The lumps are enlarged costal cartilages.

Radiation: The transmission of a body sound to another location.

Rales: See **Crackles**.

Ranula: A cystic tumor, presenting as a translucent or bluish swelling beneath the tongue, caused by obstruction of a salivary duct or a mucous gland.

Recoil, arm or leg: Test used to evaluate phasic muscle tone. The infant's arm or leg is gently extended. In premature infants of 28 or fewer weeks gestation, minimal resistance is normal. Resistance increases with gestational age.

Red retinal reflex: Reflection of a clear red color from the retina when a bright light is directed at the newborn's lens. The reflex is a pale color in dark-skinned infants.

Reflex, anocutaneous: An autonomic nervous system reflex. *Stimulus*: Cutaneous stimulation of the perianal skin. *Normal response*: Contraction of the external sphincter. Also called *anal wink*.

Reflex, Babinski: A developmental reflex. *Stimulus*: Stroke the sole of the infant's foot. *Positive response*: Extension of the great toe and spreading of the other toes.

Reflex, biceps: A motor reflex that is most active during the first 2 days after birth and when the newborn is alert. *Stimulus*: Place thumb over insertion of infant's biceps tendon, flex newborn's arm, and tap thumb with reflex hammer. *Normal response*: Infant flexes biceps muscle.

Reflex, corneal: A peripheral sensory reflex. *Stimulus*: Blow air on the infant's eye or touch the cornea gently with a piece of cotton. *Normal response*: The infant blinks.

Reflex, Moro: A developmental reflex that is normally completely present at birth only in term neonates. This reflex may be elicited by sudden movement of surface infant is on, loud noise, or causing the head to drop approximately 30°. *Stimulus*: Hold infant supine, supporting head a few centimeters above bed, withdraw supporting

hand and allow the infant to "fall back" to it. *Normal response*: As infant's head falls, infant extends and abducts arms and opens hands; then adducts and partially flexes arms, and makes fists. Infants greater than 32 weeks gestation may cry out. Premature infants do not complete the reflex by adducting and flexing the extremities, nor do they cry. Complete lack of the reflex is abnormal; asymmetric response may indicate a localized neurologic defect (such as a brachial plexus injury).

Reflex, palmar grasp: A developmental reflex that is normally present at birth in both term and preterm infants. *Stimulus*: Stroke infant's palm with a finger. *Normal response*: Infant grasps finger and tightens grasp on attempt to withdraw finger.

Reflex, patellar (knee jerk): A motor reflex that is most active during the first 2 days after birth and when the newborn is alert. *Stimulus*: Tap infant's patellar tendon just below kneecap, with newborn's knee flexed and supported. *Normal response*: Infant extends knee and contracts quadriceps.

Reflex, rooting: A developmental reflex that is normally present at birth in both term and preterm infants. *Stimulus*: Stroke infant's cheek and corner of mouth. *Normal response*: Infant turns head toward stimulus and opens mouth.

Reflex, startle: Resembles Moro reflex but is more limited. See **Reflex, Moro**.

Reflex, stepping: A developmental reflex that becomes more active 72 hours after birth. *Stimulus*: Hold infant upright with soles of feet touching a flat surface. *Normal response*: Alternate stepping movements.

Reflex, sucking: A developmental reflex that is normally present at birth in both term and preterm infants. (It is weaker in preterm neonates.) *Stimulus*: Touch infant's lips. *Normal response*: Infant opens mouth and makes sucking movements.

Reflex, tonic neck: A developmental reflex that is normally present at birth only in term neonates. *Stimulus*: With infant supine, turn infant's head to one side. *Normal response*: Extension of upper extremity on side to which head is turned and flexion of upper extremity on opposite side. Lack of response or a marked response with minimal head turning is abnormal. Also called *fencing position.*

Reflex, truncal incurvation (Galant): A developmental reflex. *Stimulus*: Suspend infant ventrally, supporting anterior chest wall in palm of hand. Apply firm pressure parallel to spine in thoracic area with thumb or cotton swab. *Positive response*: Infant flexes pelvis toward side of stimulus.

Reflexes, developmental: Reflexes that do not require functional brain above the diencephalon. Those present in most newborns include the sucking, rooting, palmar grasp, tonic neck, Moro, stepping, truncal incurvation, and Babinski reflexes. Also called *primary* or *primitive* reflexes.

Respiratory distress syndrome (RDS): A condition of the newborn most often occurring in premature infants, but may be seen at term, marked by breathing difficulties and cyanosis and having as its origin a deficiency of surfactant and structural immaturity.

Retinoblastoma: A retinal tumor. Lack of a red reflex in a newborn could indicate its presence.

Retractions: Drawing in of the chest during respiration. Immediately after birth, substernal or intercostal retractions are common; if they persist, they may indicate respiratory problems. Suprasternal retraction, especially with gasping or stridor, may indicate obstruction of the upper airway.

Rhonchi: Adventitious breath sounds lower in pitch and more musical than crackles. Although seldom heard in the neonate, they may be auscultated if secretions or aspirated foreign matter are present in the large airways.

Rotation, neck: Turning of the face to the side.

Rub: Adventitious breath sound resembling the rubbing of a finger over a cupped hand held near the ear. Rubs are often heard in newborns during mechanical ventilation; they are also associated with inflammation of the pleura.

Ruga (plural: rugae): A ridge, wrinkle, or fold. Rugae first appear on the front of the scrotal sac at about 36 weeks gestation,

covering the sac by 40 weeks. In the term male infant, the scrotum is fully rugated; the preterm male has few or no rugae.

Scale: Exfoliation of dead or dying bits of skin. Scale can also result from excess keratin production.

Scaphocephaly: Long, narrow head shape caused by craniosynostosis (premature fusion) of the sagittal suture, limiting lateral growth of the skull.

Scapula: One of the pair of triangular bones at the back of the shoulder articulating with a clavicle. Commonly called the *shoulder blade.*

Scarf sign: A test for newborn gestational age involving gently pulling the arm of a supine infant across the chest and around the neck as far as possible posteriorly. The older the infant, the greater the resistance to this maneuver.

Sclera: The white portion of the eyeball.

Sclera, blue: Unusual blue coloring of the white portion of the eyeball. It is seen with a variation of osteogenesis imperfecta and certain other abnormalities.

Scoliosis: A congenital deformity of the spine in which vertebrae fail to form and/or segment. It is embryonic in origin. If undetected, it can affect neurologic function as well as appearance. Congenital scoliosis may be associated with genitourinary tract anomalies, Klippel–Feil syndrome, and Sprengel deformity.

Seizure: An abnormal series of movements (neurologic in origin) characterized by alternate muscular contraction and relaxation of unequal amplitude. The movements are less rapid than those seen in jitteriness. They do not occur in response to stimuli and cannot be stopped by flexing and holding the affected limb; may be accompanied by abnormal eye movements. *See also* **Jitteriness**.

Sensory threshold: An infant's level of tolerance for stimuli. Infants who exceed their sensory thresholds (are overstimulated) exhibit signs and symptoms of stress. Low sensory thresholds are generally seen in preterm infants and in infants with neurologic impairment.

Sequence: A single defect as the cause for other anomalies (the "snowball" effect).

Simian crease: A single transverse palmar crease. It may be found in normal newborns, but when seen with short fingers, an incurved little finger, and a low-set thumb, it is suggestive of Down syndrome.

Sinus, dermal: A posterior neural tube defect occurring along the midline of the back, frequently in the lumbar region. It may present as a dimple surrounded by tufts of hair or lipomas or may terminate in subcutaneous tissue, a cyst, or a fibrous band. It may also extend into the spinal cord and be associated with spina bifida.

Sinus, preauricular: An abnormal channel located in front of the pinna (auricle) of the ear. If it communicates with the internal ear or the brain, chronic infection is likely.

Skin tag: A small skin outgrowth. Those occurring anterior to the tragus are thought to be embryological remnants of the first branchial arch. Skin tags may be familial or associated with other anomalies.

Small for gestational age (SGA): An infant whose weight falls below the 10th percentile for gestational age when plotted on a standard growth chart.

Spina bifida: The mildest form of posterior neural tube defect, with the malformation arising from lack of or incomplete closure of the posterior portion of the vertebrae. The meninges and spinal cord are normal. The defect is covered with skin (which may be dimpled), and it is most common in the lower lumbar and lumbosacral area. *See also* **Sinus, dermal**.

Split: Term describing a heart sound in which two components can be heard. The "splitting" is caused by the asynchronous closure of the two valves that create the sound. The rapid newborn heart rate makes splitting difficult to auscultate. In newborns splitting is usually not heard in S1. S2 is generally single at birth but is split in two-thirds of infants by 16 hours of age and in four-fifths by 48 hours of age. Wide splitting of S2 is abnormal, however.

Sprengel deformity: A congenital anomaly of the shoulder girdle, marked by some hypoplasia and malrotation of the scapula, which gives it an elevated appearance.

Shoulder abduction and flexion are limited. This deformity is more often unilateral, but can be bilateral. It is often associated with congenital spinal anomalies, Klippel–Feil syndrome, and renal anomalies.

State (of consciousness): One of six levels of consciousness seen in infants. There are two sleep states (*deep* and *light*), one transitional state (*drowsiness*), and three awake states (*quiet alert, active alert*, and *crying*). Behavioral assessment evaluates the newborn's ability to control his state, move smoothly from one state to another, and maintain alertness when in an awake state. Term infants may use changes in state to control environmental input.

Stenosis, anal: Constriction or obstruction of the anorectal canal. Signs include passage of small, thin stools; progressive abdominal distention; and finally, vomiting.

Stenosis, pyloric: Obstruction of the pyloric orifice of the stomach, usually presenting about 3 weeks of age, rarely before the fourth or fifth day of life. If present, it is palpable immediately after feeding or vomiting as an ovoid mass between the umbilicus and the right lower costal margin. Signs of pyloric stenosis (upper quadrant distention and visible peristalsis) in the absence of a palpable mass may indicate duodenal or jejunal obstruction in the neonate.

Sternocleidomastoid muscle: The muscle that flexes the vertebral column and rotates the head.

Stethoscope: An acoustical device used to auscultate (listen to) sounds within the body. In the neonate, these include heart, lung, venous, arterial, and intestinal sounds. A neonatal stethoscope generally has a double head composed of a flat diaphragm (for auscultating high-frequency sounds) and a bell (for auscultating low-frequency sounds). It does not magnify sound but simply eliminates environmental noise.

Stippling: A speckling of fine dots.

Strabismus: The appearance of crossed eyes in the newborn due to muscular incoordination.

Streeter dysplasia: See **Congenital constricting bands**.

Stridor: A high-pitched hoarse adventitious breath sound heard during inspiration or expiration at the larynx or upper airways. In the newborn, stridor indicates partial obstruction of the airway. It may also be heard in infants with edema of the upper airway after extubation.

Structural deformity: A fixed (or rigid) deformity involving bony abnormalities. It generally requires surgical intervention.

Subcutaneous tissue: Layer of fatty tissue underlying the dermis, which insulates the body, protects the internal organs, and stores calories.

Sucking blister: Intact or ruptured vesicle or bulla sometimes seen on the lips, fingers, or hands of the newborn. The cause is vigorous sucking, in utero or after birth.

Sunset sign: Eyelid retraction and a downward gaze in which the sclera is visible above the iris. The sign is often seen in infants with hydrocephalus.

Supination: Turning of the body or a body part face up, as of the hand so that the palm is facing up.

Supine: Positioned face up.

Suprasternal notch: An indentation on the upper border of the sternum.

Suture: A fibrous joint between bones of the skull. The **metopic suture** extends midline down the forehead between the two frontal bones and intersects with the **coronal suture**, which separates the frontal and parietal bones. The **sagittal suture** extends midline between the two parietal bones to the back of the head, where it intersects with the **lambdoidal suture**, which separates the parietal and occipital bones. The **squamosal suture** extends above the ear and separates the temporal from the parietal bone.

Sutures, overriding: Head molding may cause the edge of the bone on one side of a suture to feel as if it is on top of (overriding) the edge of the opposite bone. Overriding is commonly seen in the lambdoidal suture, with the parietal bone on top of the occipital. Overriding sutures must be differentiated from craniosynostosis (premature fusion of a suture).

Symmetrically growth restricted: An infant who has not grown at the expected rate—that is, who ranks at less than the 10th percentile for gestational age on standard growth charts—for *all three* growth parameters: weight, length, and occipital-frontal head circumference.

Symphysis, pubis: Midline cartilaginous joint connecting the pubic bones.

Syndactyly: Congenital webbing of the fingers and toes, frequently familial in origin. The more severe the webbing, the greater the likelihood of underlying bony abnormalities.

Syndrome: A recognizable pattern of anomalies, often of more than one system, that is historically based and leads to a diagnosis.

Systole: The period of contraction of the heart, particularly of the ventricles.

Tachycardia, sinus: A heart rate greater than 180 to 200 beats per minute in the newborn. Simple tachycardia, originating in the sinus node of the heart, normally occurs in response to a stimulus that increases demand on the heart. When the stimulus is removed, the rate gradually returns to normal.

Tachycardia, supraventricular (SVT): A heart rate greater than 200 to 300 beats per minute in the newborn. Without immediate intervention, it can lead to cardiovascular collapse.

Tachypnea: Excessively rapid and shallow respirations. In the neonate, persistent respirations of greater than 60 per minute may indicate TTN, RDS, meconium aspiration, pneumonia, hyperthermia, or pain.

Tap: A sharp, well-localized point of maximum cardiac impulse, usually associated with pressure overload or hypertrophy.

Temperament: The way in which an infant interacts with the environment. S. Chess and A. Thomas identify three categories of temperament: easy, difficult, and slow to warm.

Teratogenic agent: An extrinsic factor—either environmental or maternal—that causes abnormalities in fetal development. Examples include infection, metabolic disorders, and exposure to drugs or alcohol, heavy metals, many chemicals, and radiation.

Term: An infant born between the beginning of week 38 and the end of week 41 of gestation.

Testicular torsion: See **Neonatal torsion of the testis**.

Thorax: The chest. The thoracic cavity is bound by the sternum, 12 thoracic vertebrae, 12 pair of ribs, and the diaphragm.

Thrill: A low-frequency, palpable murmur that resembles holding one's hand on a purring cat. Thrills are uncommon in the newborn. If present, their location can aid in identification of cardiac problems.

Thrush: An oral fungal infection characterized by adherent white patches on the tongue and mucous membranes and caused by *Candida albicans.*

Thyroglossal duct cyst: A cyst on the embryological duct between the thyroid and the back of the tongue.

Tonic: Of or relating to or producing normal tone or tonus in muscles or tissue.

Torticollis: An anomaly affecting the sternocleidomastoid muscle, more often on the right side. It is not usually seen until about 2 weeks of age, when it can be palpated as a firm, immobile mass 1 to 2 cm in diameter in the midportion of the muscle. Undetected, it produces facial asymmetry and limited neck rotation.

Traction response (pull-to-sit maneuver): Tests the ability of the infant's muscles to resist the pull of gravity. When the normal-term infant's hands are grasped and he is slowly pulled from a supine to a sitting position, the infant contracts the shoulder and arm muscles, and then flexes the neck, with the head lagging behind the body only minimally. The infant also flexes the elbows, knees, and ankles. When the infant reaches the sitting position, the head remains erect briefly before falling forward. Neck flexion is not seen in infants of less than 33 weeks gestation.

Tragus: A small cartilaginous flap in front of the external opening of the ear.

Transient tachypnea of the newborn (TTN): A condition of the newborn marked by excessively rapid respirations. The underlying pathology is usually retained fetal lung fluid; tachypnea subsides as fluid is absorbed or expelled.

Transillumination: A physical assessment technique involving shining light through body tissues. The area being examined is placed between the light and the observer. If a pneumothorax is suspected in the neonate, transillumination can allow comparison of the left, right, upper, and lower aspects of the chest and illuminate air pockets with a lantern-like glow. If the infant's head is an unusual size or shape or if the neurologic examination is abnormal, transillumination can help identify the reason. It is useful for identification of abnormal findings in the scrotum. Masses filled with clear fluid appear translucent with illumination; solid or blood-filled masses do not transilluminate.

Trisomy: A form of chromosomal aneuploidy characterized by the presence of 47 chromosomes. It is generally caused by nondisjunction (failure of the egg or sperm to divide the genetic material equally). The most common autosomal trisomies are of chromosome 21 (Down syndrome), chromosome 18, and chromosome 13.

Urethral meatus: Opening of the urethra on the body surface, located directly below the clitoris in female infants and at the center of the end of the penile shaft in males. Anterior displacement of the meatus at or on an enlarged clitoris suggests pseudohermaphroditism in a female infant. Hypospadias and epispadias are placement abnormalities seen in male infants.

Uvula, bifid: A split uvula. This may be associated with other congenital anomalies.

Valgus: Bent outward or twisted away from the midline of the body.

Varus: Turned inward.

Vernix caseosa: A greasy white or yellow material, composed of sebaceous gland secretions and exfoliated skin cells, that covers the newborn infant's skin. Vernix develops during the third trimester of intrauterine growth, gradually decreasing in amount as the fetus approaches 40 weeks gestation. Because vernix may be present in the newborn's auditory canal, otoscopic examination is not done immediately after birth.

Very low birth weight (VLBW): An infant who weighs less than 1,500 g at birth.

Vesicle: An elevation of the skin filled with serous fluid and less than 1 cm in diameter.

Vesicular breath sounds: Normally found over the entire chest, except over the manubrium and the trachea. During expiration, vesicular sounds are soft, short, and low-pitched; during inspiration, they are louder, longer, and higher pitched.

Weaver curve: Weaver curves are reference curves used to determine whether genetic influences account for a child's macrocephaly by comparing a reference score for the average parents' OFC with the child's reference score for OFC. A genetic cause for macrocephaly is confirmed if the child's standard score is within the range determined by the average parental score on the Weaver curve.

Werdnig–Hoffmann disease: A disorder of the lower motor neurons, causing flaccid weakness of the extremities as well as tongue fasciculation.

Wheal: A reddened, solid elevation of the skin caused by a collection of fluid in the dermis.

Wheezes: Adventitious breath sounds, usually louder on expiration than on inspiration, that are higher in pitch than rhonchi. They are usually heard only in newborns with CLD.

Whorl, hair: Spiral hair growth pattern commonly seen in the posterior parietal region of the newborn's scalp. Absence of or abnormal location of a whorl can indicate abnormal brain growth.

Witch's milk: Milky secretions engorging the breasts of some infants at birth, due to the influence of maternal estrogen. The secretions generally disappear in 1 to 2 weeks, and the enlargement subsides over several months.

Xiphoid process: Sword-shaped cartilaginous projection at the end of the sternum.

Glossary Compiled and Verified Using the Following Sources

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