VARIATIONS AND MINOR DEPARTURES IN INFANTS
The delicate skin of a newborn is thin, and usually has a pale pinkish cast (even in African American babies) from the blood vessels just beneath the surface. Therefore, the presence of healthy color and turgor may have great significance in your estimate of a newborn infant’s general health. However, the normal range of skin color is great – and the occurrence of essentially nonpathological transitory changes in both color and turgor is common in newborns.

It is understandable that the first anxious questions of many mothers relate to the appearance of their baby’s skin. The illustrations on the following pages offer a brief, graphic summary of some of the conditions frequently encountered in newborns. Birthmarks are included in a separate section. We hope these collections of clinical photographs will be useful for simple review – or to illustrate your explanations to apprehensive parents.

The skin of the newborn has a relatively high circulation rate, which accounts for the general redness of the skin that is a typical characteristic of the healthy term newborn. The skin of premature infants is thin and delicate, and tends to be deep red. In extremely premature infants, the skin may appear almost gelatinous, and bleeds and bruises easily. Vasomotor instability and peripheral circulatory sluggishness are revealed by deep redness or purple lividity in a crying infant, whose color may darken profoundly preceding a vigorous cry. Mottling, another example of general circulatory instability, may be associated with serious illness or related to a transient fluctuation in skin temperature. A transient and harmless, yet dramatic, event known as the harlequin color change is manifested as a division of the newborn’s body from forehead to the pubis into red and pale halves.

Localized cyanosis is differentiated from ecchymosis by the momentary blanching pallor (with cyanosis) that occurs after pressure. The same maneuver can also help in demonstrating icterus (jaundice), which can be significant, but often goes unnoticed if the skin is suffused with blood. Other pathophysiologic conditions that have been determined by studies of skin blood flow (SBF) in newborns include shock, sepsis, and apnea of prematurity. At least one study has postulated that increased SBF can be used as an indicator of pain or discomfort in newborns.
ACROCYANOSIS

Peripheral acrocyanosis (cyanosis of the hands, feet and sometimes the lips) is frequent during the early hours of life. It may also be observed in many older infants, particularly when they have been exposed or chilled. It is generally ascribed to limited development of the peripheral capillary circulation of the skin, and does not usually warrant concern. However, it is important to distinguish acrocyanosis from central cyanosis. Central cyanosis has respiratory, cardiac, central nervous system, hematologic, and metabolic causes, and is usually accompanied by obvious signs of respiratory difficulty.

PHYSIOLOGICAL JAUNDICE

Hyperbilirubinemia (jaundice) is a common and, usually, benign problem in newborns. Jaundice is observed during the first week of life in approximately 60% of term infants and 80% of preterm infants. The color results from the accumulation in the skin of unconjugated, nonpolar, lipid-soluble bilirubin pigment. Jaundice resulting from deposits of indirect bilirubin in the skin tends to appear bright yellow or orange, and jaundice of the obstructive type (direct bilirubin) has a greenish or muddy yellow cast. This difference is usually apparent only in severe jaundice. Severely affected infants may be lethargic and may feed poorly. Jaundice usually begins on the face and, as serum levels increase, progresses to the abdomen, and then the feet. It may be present at birth or appear at any time during the neonatal period, depending on the cause.

Regardless of the benign nature of physiological jaundice, untreated, severe indirect jaundice is potentially neurotoxic, and conjugated-direct jaundice often signifies a serious hepatic or systemic illness. Therefore, jaundice that appears before 24 hours of age, jaundice that is >95th percentile according to the infant’s age in hours in a term infant (lower levels in a preterm neonate), jaundice treated with phototherapy or where total serum bilirubin is rising rapidly and where not explained by history and physical examination, jaundice approaching exchange transfusion levels or not responding to phototherapy, elevated direct bilirubin level, and jaundice persisting beyond 3 weeks or in a sick infant should be evaluated further.

ERYTHEMA TOXICUM

Toxic erythema of the newborn is characterized by a rash of red speckles with yellowish-white bumps in the centers. It is the most common neonatal skin lesion, and may occur any time during the first week, although the second day is most common. It is usually found where clothing rubs the arms, legs, and back, and only rarely occurs on the infant’s face. Erythema toxicum generally disappears without treatment within the first week or so. However, in severe cases, a family history of allergies should be sought, and, if found, use of lotions, powders, perfumed soaps, and plastic should be avoided.

MILIA

Milia are very common, benign, keratin-filled cysts that appear as superficial, uniform, pearly-white to yellowish-domed lesions measuring 1-2 mm in diameter, usually on the face, especially the nose. Primary milia are believed to arise in sebaceous glands that are not fully developed. No topical or systemic medications are effective on primary milia, and they tend to spontaneously disappear within the first few weeks of life.

FORCEPS MARKS

When forceps are used to help during a delivery, the baby may have red marks or even superficial scrapes on the face and head where the metal pressed against the skin. The marks are usually only important because mothers notice them and may be alarmed. These should disappear within a few days. Sometimes a firm flat lump develops in one of these areas, due to minor tissue damage under the skin, but this is likely to go away within two months. Erythema, abrasions, ecchymoses, and subcutaneous fat necrosis of facial or scalp soft tissues may be noted after forceps or vacuum-assisted deliveries. Their location depends on the area of application of the forceps. Facial palsy can also result from pressure over the facial nerve when forceps are used during delivery. The prognosis depends on whether the nerve was injured by pressure or if nerve fibers were torn. If the nerve was injured by pressure, improvement should occur within a few weeks.
MOLDING

Molding of the skull bones during the birth process is a common cause of temporary asymmetry of the newborn’s head. During a head-first birth, pressure on the head caused by the tight birth canal may “mold” the head into an oblong shape, which returns to normal within a few days. This is particularly likely if the infant is a first-born and if the head has been engaged for a considerable time. The head of an infant born by cesarean section or from a breech presentation is characterized by its roundness.

Diagram 1 shows a neonatal skull on the first day of life, demonstrating molding of the skull when the parietal bones override the occipital and frontal bones, causing a narrowing of the sutures and elongation of the head.

Diagram 2 is of the third day of life, and shows reexpansion of the cranial and widening of the sutures and fontanels. The parietal, occipital and frontal bones have returned to normal positions.
FACIAL ASYMMETRY

Although frequently attributed to forceps pressure, most trauma to the facial nerve probably results from pressure on the nerve in utero, which may be due to fetal positioning (from the head lying against the shoulder) or to pressure against the nerve by the sacral promontory or a uterine fibroid. Facial nerve injury usually occurs at or distal to its exit from the stylomastoid foramen, resulting in facial asymmetry, especially when crying. When the jaw has been held against a shoulder or an extremity during the intrauterine period, the mandible may deviate strikingly from the midline. However, muscle innervation is intact, and both sides of the face can move. Comparing the maxillary with the mandibular occlusal surfaces, which should be parallel, differentiates this from a true facial nerve injury. Testing or treatment is not needed for peripheral facial nerve injuries or mandibular asymmetry, since they usually resolve by age 2 to 3 months. Symmetric facial palsy, however, suggests absence or hypoplasia of the 7th nerve nucleus (Möbius syndrome), a rare genetic disorder characterized by facial paralysis and an inability to suck.

CAPUT SUCCEDANEUM

Caput succedaneum is a diffuse, edematous, sometimes ecchymotic swelling of the soft tissues of the scalp involving the portion of the head presenting during vertex delivery. The swelling is not sharply defined and may extend across the midline and across suture lines. Analogous swelling, discoloration and distortion of the face are seen in face presentations. The edema usually disappears within the first few days of life. No specific treatment is needed, but if extensive ecchymoses are present, hyperbilirubinemia may develop.

CEPHALHEMATOMA

Caput succedaneum may temporarily mask the presence of cephalohematoma, a subperiosteal hemorrhage. No discoloration of the scalp occurs, and swelling is not usually visible until several hours after birth because subperiosteal bleeding is a slow process. An underlying skull fracture, usually linear and not depressed, is occasionally associated with cephalohematoma. Most cephalohematomas are resorbed within 2 weeks to 3 months, depending on their size. They require no treatment, although phototherapy may be necessary to ameliorate hyperbilirubinemia.

PLAGIOCEPHALY

When present at birth, this type of craniofacial asymmetry is probably due to a restrictive intrauterine environment. A breech orientation can lead to abnormal head shape when the infant’s head becomes wedged under the mother’s ribs. Premature delivery also makes the already soft cranium particularly susceptible to molding. Other risk factors include male gender, firstborn, and limited passive neck rotation at birth. The majority of cases will have resolved by age 2 years.
Most parents, seeing their new baby for the first time, are apt to look first at his face, and, especially, his eyes, since they are by far the most dominant facial feature in the neonate. At birth, the eye of a normal full-term infant is approximately 65% of adult size. Small wonder that any one of several minor variations from normal may create anxiety among parents, and the need will exist for prompt reassurance. While the healthcare professional’s prime concern is to assess and safeguard the infant’s visual function, the parents are often more concerned with what they believe to be the immediate visible problems.

Many of these worrisome eye abnormalities are transient phenomena related to ocular or neuromuscular immaturity or the trauma of birth. Clinical illustrations of some of these variations are presented on the following pages.

EYE COLOR

Most Caucasian babies are born with dark gray-blue eyes that can take weeks or months to reveal their true color. Many African-American, Asian, and Hispanic babies are born with dark gray-brown eyes that don’t change color significantly, but some may start out with hazel eyes that get darker as they approach 6 months. A gradual deposition of pigment produces the final eye color of the individual by 6 to 9 months of age.31

A speckling of the iris, known as Brushfield spots, is characterized by white spots slightly elevated on the surface of the iris arranged in a ring concentric with the pupil.32 These spots occur in normal children, but are far more frequent in Down syndrome (trisomy 21).32,33 They were described in 1924 by Thomas Brushfield, and are due to aggregation of a normal iris element (connective tissue).32

Until recently, it was believed that the lacrimal apparatus of the neonate is nonfunctioning, and that tears are not produced with crying until 1 to 3 months of age. However, recent research has shown that term infants do manifest normal lacrimal secretions.34 Preterm infants also secrete tears, but to a lesser extent, the quantity of secretion being proportional to birth weight.34 The precorneal tear film performs the essential function of maintaining an optically uniform corneal surface essential for normal visual development and central fixation, which develop during the first 3 weeks of life.34

REFLECTIONS OF ANATOMIC IMMATURETY

Development of the eyes begins very early in embryonic life, and is relatively far advanced at birth. During the embryonic period, which occurs from 2 to 8 weeks after conception, the outer layer of cells forms what will become the nervous system, the sensory receptors (eyes, nose, ears), and the skin.35 At 10 weeks, the eyelids fuse the eyes shut until the 7th month, when the eyes open and blink in response to sound.36 There is evidence to suggest that some sense of light and dark may be associated with the development at this time of cycles of activity.36

Growth and anatomic immaturity of the eyes, therefore, seem to be relatively advanced in the newborn infant, compared to visual function. Newborn infants can see, respond to changes in illumination, and track moving objects.36 Recent findings indicate that visual function, rather than visual acuity, is the primary concern of parents.36 However, several visible signs reflect these functional immaturities.

VISIBLE STRUCTURES WITHIN THE EYE

In an infant, the sclera is thin and translucent, with a bluish tinge.37 The cornea is relatively large, and attains adult size by 2 years of age.37 In infants born prematurely, the cornea may have a transient opalescent haze.37 The lens of a newborn is more spherical than that of an adult. Its greater refractive power helps compensate for the relative shortness of the young eye.37 The macular landmarks, particularly the foveal light reflexes, are less well defined in the newborn, and may not be readily apparent.37 Superficial retinal and conjunctival hemorrhages may also occur at birth.37 These are usually resolved spontaneously without consequence.

THE INFANT’S EYES
DOLL'S EYE MANEUVER

Complete ocular movement may be demonstrated as early as 25 weeks gestation using the doll's eye maneuver. This technique is used to examine horizontal and vertical eye movements in an infant. If the head is suddenly turned to the right, the eyes look to the left in a symmetric fashion. Normal infants follow a toy or interesting object in all directions. Premature infants tend to have slightly disconjugate eyes at rest, with one eye horizontally displaced from the other by 1 or 2 mm. Skew deviation of the eyes (vertical displacement) is always abnormal and requires investigation.

INCOORDINATE EYE MOVEMENTS

Many normal infants may have imperfect coordination of their eye movements and alignment during the early days and weeks of their lives. In the newborn period, transient nystagmus is common. These involuntary rapid movements of the eye may be horizontal, vertical, rotatory, pendular, or mixed. Proper coordination should be achieved by 3-6 months, probably sooner. Persistent deviation of an eye in an infant requires evaluation.

REFLECTIONS OF NEUROMUSCULAR IMMATUREITY

At birth, the forebrain is so immature that its influences are limited to signaling distress in reaction to hunger or thirst, a function of the immature hypothalamus. Although various other nuclei become functionally mature over the course of the first several postnatal months and years, this immaturity explains many of the temporary neurobehavioral symptoms described below.

PSEUDOOSTRABISMUS

Pseudostrabismus (pseudoesotropia) is one of the most common reasons a pediatric ophthalmologist is asked to evaluate an infant. This condition is characterized by the false appearance of strabismus when the visual axes are accurately aligned. It may be caused by a flat, broad nasal bridge, prominent epicanthal folds, or a narrow interpupillary distance. Pseudostrabismus can be differentiated from true strabismus when the corneal light reflex is centered in both eyes, and when the cover-uncover test shows no refixation movement. However, since true esotropia can develop later in children with pseudoesotropia, healthcare professionals and parents should be cautioned that reassessment is required if the apparent deviation does not improve.

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SETTING-SUN SIGN AND GRAEFE’S SIGN

These two signs should be distinguished from each other in spite of their similar presentations. Graefe’s sign is a band of white sclera above the iris when the eyes are not moving. In the absence of other symptoms, Graefe’s sign is not indicative of intracranial hypertension. It is frequently seen in premature infants with esotropia, exotropia, and heterotropia. Setting sun sign is elicited by repeated head nods. The irises deviate downward and appear to sink beneath the lower lids due to impingement of the dilated suprachiasmal recess on the tectum. Although further evaluation is warranted, a benign form of the setting sun phenomenon might exist until 6 or 7 months of age in normal infants.

EYELIDS

Newborn infants tend to keep their eyes closed much of the time, but normal newborns can see, respond to changes in illumination, and fixate points of contrast. Normal 28-week-gestation premature infants blink when a bright light is directed to the eyes, and, by 32 weeks, infants maintain eye closure until the light source is removed. At 37 weeks, normal premature infants turn their head and the eyes to a soft light, and, by term, visual fixation and the ability to follow a brilliant target is recognized.

Any attempt to force the eyelids open will likely be met with resistance on the part of the infant. However, the eyes often open spontaneously if the infant is held up and tipped gently forward and backward. The success of this maneuver is a result of labyrinthine and neck reflexes. Retinal hemorrhages are common, especially with vacuum-assisted deliveries, but they resolve in most infants by 2 weeks, and in all infants by 4 weeks of age. Papillary reflexes are present after 28-30 weeks of gestation. The iris should be inspected for colobomas and heterochromia.

Pink, red or salmon-colored patches that appear on the eyelids are often referred to as “stork bites.” These patches tend to become lighter during the first two years, becoming noticeable only when the child cries or exercises himself.

DOLLS EYE MANEUVER

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SUBCONJUNCTIVAL HEMORRHAGE

Subconjunctival hemorrhage is the breakage of small blood vessels in the eyes of the newborn. One or both of the eyes may have a bright red band around the iris. Although likely to have occurred during the birth process, it may occasionally result from severe sneezing or coughing. Rarely it may be a manifestation of a blood dyscrasia. The condition is similar to a skin bruise that goes away after several days, without permanent damage to the infant’s eyes.

CHEMICAL CONJUNCTIVITIS

Another common lesion which may concern parents is the result of the irritation induced by silver nitrate drops used in the prevention of gonococcal ophthalmia. This chemical conjunctivitis is acute, but benign, usually occurring within 6-12 hours after birth, and clearing by 24-48 hours. It is characterized by redness and swelling of the conjunctiva, edema of the eyelids, and discharge, which may be pusulent. Additional confirmation of the chemical etiology of the conjunctivitis can be provided by negative cultures, if warranted. Before the institution of topical ophthalmic prophylaxis at birth, gonococcal ophthalmia was a common cause of blindness or permanent eye damage in infants.

THE INFANT’S BIRTHMARKS

Vascular and pigmented nevi, or birthmarks, are frequent abnormalities in newborn infants. Because they are so common, and usually so conspicuous, their importance tends to be overemphasized. Their causation is often the subject of many misconceptions and superstitions, even among well-educated parents.

Not uncommonly, a parent may believe the infant was “marked” by some traumatic incident during pregnancy, and may even feel some guilt for the blemish. Even enlightened parents may find it hard to resist this notion, especially if their own parents have strong convictions as to the cause.

The understanding healthcare professional can do much to allay parental anxiety through explanation, stressing the high prevalence of nevi, and the lack of scientific evidence implicating prenatal influences in their causation. In many cases, the likelihood of spontaneous regression will provide further reassurance, and should be stressed. Some of the most common birthmarks are described and illustrated on the succeeding pages.
STRAWBERRY MARK

Nevus vasculosus or strawberry mark is a bright or dark red, raised or swollen, bumpy area that looks like a strawberry. Hemangiomas are formed by a concentration of tiny, immature blood vessels. Favored sites are the face, scalp, back, and anterior chest. They may not appear at birth, but often develop in the first two months. Strawberry hemangiomas are more common in premature babies, and in girls more often than boys. This birthmark is likely to grow in size for several months, then gradually begin to fade. Most strawberry hemangiomas completely disappear by 9 years of age.

CAVERNOUS HEMANGIOMA

This birthmark is less common than the strawberry hemangioma, affecting only 1 or 2 out of every 100 babies. It is composed of larger more mature vascular elements, and involves deeper layers of skin. The cavernous hemangioma is likely to be a blue or blue-red lumpy mass that has less distinct borders than a strawberry. Favored sites are the face, scalp, back, and anterior chest. It may not appear at birth, but often develop in the first two months. Strawberry hemangiomas are more common in premature babies, and in girls more often than boys. This birthmark is likely to grow in size for several months, then gradually begin to fade. Most strawberry hemangiomas completely disappear by 9 years of age.

SALMON PATCH

True port-wine stains should be distinguished from the most common vascular malformation, the salmon patch of neonates, which is a relatively transient lesion. The salmon patches (sometimes called stork bites) are small pink or red patches found on the infant’s neck, eyes, or upper lip. They are caused by a concentration of immature blood vessels, and may be visible when the baby is crying. Most of these marks fade and disappear completely.

PORT WINE STAIN

Nevus flammeus, or port-wine stains, are present at birth. These vascular malformations consist of mature dilated dermal capillaries, and represent a permanent developmental defect. The lesions are macular, sharply circumscribed, pink to purple, and varied in size. The head and neck regions are the most common areas for the port-wine stains, and most lesions are unilateral. In the absence of associated anomalies, morbidity from these lesions may include a poor self image, hypertrophy of underlying structures, and traumatic bleeding.

VASCULAR NEVI

Developmental vascular anomalies may occur as isolated defects or as part of a syndrome. They can be separated into two major categories: hemangiomas and vascular malformations. Most hemangiomas are not present at birth, or are very faint red marks. However, shortly after birth they begin to grow rapidly. Over time they become smaller and lighter in color, but this process may take several years. Hemangiomas are the most common tumor of infancy, occurring in 1% of newborns and 5% of Caucasian infants in the 1st year of life. Vascular malformations are present at birth, and are derived from capillaries, veins, arteries, or lymphatics. Hemangiomas do not regress but usually enlarge over time.

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ACCESSORY NIPPLES

Solitary or multiple accessory nipples may occur in a unilateral or bilateral distribution along a line from the anterior axillary fold to the inguinal area. They are more common in African American infants (3.5%) than in Caucasian infants (0.6%). Accessory nipples may or may not have an areola and may be mistaken for congenital nevi. They only rarely undergo malignant change.

MONGOLIAN SPOTS

Mongolian spots are blue to slate gray and may resemble bruises. These well-demarcated areas of pigmentation may appear over the buttocks, back, and other parts of the body in more than 50% of African American, Native American and Asian infants. These patches are also fairly common in infants of Mediterranean ancestry, but are rare in blond-haired, blue-eyed infants. They tend to disappear within the first year.

OTHER PIGMENTED VARIATIONS

CONGENITAL PIGMENTED NEVI

These moles vary in color from light brown to black and may be hairy. Small ones are very common and may occur on the lower trunk, upper back, shoulders, chest, or proximal limbs. Removal of all small congenital nevi is not warranted, particularly since the development of melanoma in a small congenital nevus is a very rare event before puberty.

NEVUS SPILUS

This nevus is a brown patch within which are darker flat or raised brown melanocytic elements. Nevus spilus is rare at birth, and is commonly acquired during late infancy or early childhood. It does not need to be excised unless atypical features or recent clinical changes are noted.

PIGMENTED NEVI (MOLES)

Melanocytic nevi are subdivided into two broad categories: those that appear after birth or acquired nevi, and those that are present at birth, the congenital nevi. Congenital melanocytic nevi are present in approximately 3% of newborn infants.

ACCESSORY NIPPLES

Solitary or multiple accessory nipples may occur in a unilateral or bilateral distribution along a line from the anterior axillary fold to the inguinal area. They are more common in African American infants (3.5%) than in Caucasian infants (0.6%). Accessory nipples may or may not have an areola and may be mistaken for congenital nevi. They only rarely undergo malignant change.
Human feeding and respiratory systems share a portion of the pharynx between the area behind the tongue and the entrance to the larynx and to the esophagus. Since the goal of swallowing is to move food into the digestive system and keep it out of the respiratory system, congenital malformations that may cause problems in this process need to be identified and differentiated from those that require no treatment.

**Newborn Mouth**

Nutritive sucking in neonates first develops at about 34 weeks gestation. The coordinated oral and pharyngeal movements necessary for swallowing solid foods develop within the first few months of life, in term infants. Before this time, the tongue thrust is upward and outward in order to express milk from the nipple. The cheeks of a normal newborn have a fullness on both the buccal and the external aspects as a result of the accumulation of fat making up the sucking pads. These pads, as well as the labial tubercle on the upper lip (sucking callus), disappear when sucking ceases. A marble-sized buccal mass is usually due to benign idiopathic fat necrosis.

**Internal Structures Promote Feeding**

Proportional differences exist between the young infant and the older infants, child, and adult. The oral cavity is small in a newborn, and is totally filled by the tongue, due to a small and slightly retracted lower jaw. The sucking pads already mentioned provide stability during sucking. The soft palate and epiglottis are in contact at rest, providing an additional valve at the back of the oral cavity. The larynx and hyoid cartilage are higher in the neck and closer to the base of the epiglottis, providing added protection of the airway. The infant’s eustachian tube runs horizontally from the middle ear into the nasopharynx, in contrast to the older child and adult, where the angle is vertical. This positioning of the eustachian tube places the infant at greater risk for ear infections from food or liquid that reflux upward into the nasopharynx.

The soft and hard palate should be inspected and palpated for a complete or submucosal cleft, and the contour noted if the arch is excessively high or the uvula is bifid. The throat of a newborn infant is hard to see because of the low arch of the palate. However, it should be clearly viewed because it is easy to miss posterior palatal or uvular clefts. The tonsils will be small.

**The Infant’s Mouth**

Many findings made during examination of a newborn’s mouth are of clinical value because they reflect the stage of anatomic development and, to some extent, the effects of the fetal environment.

Human feeding and respiratory systems share a portion of the pharynx between the area behind the tongue and the entrance to the larynx and to the esophagus. Since the goal of swallowing is to move food into the digestive system and keep it out of the respiratory system, congenital malformations that may cause problems in this process need to be identified and differentiated from those that require no treatment.

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THRUSH

Oropharyngeal infection with Candida albicans (thrush, moniliasis) is common to neonates from contact with the organism in the birth canal. The lesions appear as plaques covering all or part of the oropharyngeal mucosa. The plaques are removable from the underlying surface, which is characteristically inflamed with pinpoint hemorrhages. Thrush is usually self-limited in the healthy newborn infant, but treatment with nystatin will hasten recovery and reduce the risk of spreading to other infants.

RETENTION CYSTS

Retention cysts may be seen on the gums. These will usually disappear within a few weeks. Clusters of small white or yellow follicles or ulcers on an erythematous base may be found on the anterior tonsillar pillars, most frequently on the 2nd to 3rd day of life. Their cause is unknown, but they usually clear without treatment in 2 to 4 days.

SHORT LINGUAL FRENUM

A number of normal anatomic variations may be noted in the mouth. Ankyloglossia or “tongue-tie” is characterized by a short lingual frenum that hinders the tongue movement. It generally requires no treatment. However, if the extent of the ankyloglossia is severe, speech may be affected, and surgical correction indicated. A bifid uvula may be normal or associated with a submucous cleft of the soft palate. Geographic tongue (migratory glossitis) or fissured tongue (scrotal tongue) may present as surface furrowing of the tongue. Geographic tongue is characterized by one or more smooth, bright-red patches, often showing a yellow, gray, or white membranous margin on the dorsum of an otherwise normally roughened tongue. The condition has no known cause, and no treatment is indicated. Fissured tongue is manifested by numerous small furrows or grooves on the dorsal surface.

NATAL AND NEONATAL TEETH

A normal mouth may have precocious dentition, with natal (present at birth) or neonatal (eruption after birth) teeth in the lower incisor position or aberrantly placed. These teeth are shed before the deciduous ones erupt. Rarely, these teeth may be indicative of Ellis-van Creveld, Hallermann-Streiff, or other syndromes. If the teeth are loose, they may need to be removed, since some believe they can present a choking hazard in an infant. An X-ray may be done to determine if the teeth are extra, or, as is more commonly the case, if they are part of the normal primary set of teeth (baby teeth) and have come through the gums early.

GEOGRAPHIC TONGUE

Geographic tongue (migratory glossitis) or fissured tongue (scrotal tongue) may present as surface furrowing of the tongue. Geographic tongue is characterized by one or more smooth, bright-red patches, often showing a yellow, gray, or white membranous margin on the dorsum of an otherwise normally roughened tongue. The condition has no known cause, and no treatment is indicated. Fissured tongue is manifested by numerous small furrows or grooves on the dorsal surface.
MUCOCELE

Mucus retention cysts (mucocele) are painless, bluish, fluctuant, tense, 2-10 mm papules on the lips, tongue, palate, or buccal mucosa. Traumatic severance of the duct of a minor salivary gland leads to submucosal retention of mucus secretions. Those on the floor of the mouth are known as ranulas when the submaxillary or sublingual salivary ducts are involved. Fluctuations in size are usual, and the lesions may disappear temporarily after traumatic rupture. Recurrence is prevented by excising the mucocele.

EPSTEIN’S PEARLS

Bohn nodules (Epstein’s pearls) are small developmental anomalies located along the buccal and lingual aspects of the mandibular and maxillary ridges, and in the hard palate of the neonate. These lesions arise from remnants of mucus gland tissue. Treatment is not necessary, as the nodules disappear within a few weeks.

Dental lamina cysts are small cystic lesions located along the crest of the mandibular and maxillary ridges of the newborn. These lesions arise from the epithelial remnants of the dental lamina. Treatment for these cysts is not necessary, as they will also likely disappear within a few weeks.

MACROGLOSSIA

Macroglossia is tongue enlargement that leads to functional and cosmetic problems. Although this is a relatively uncommon disorder, it may cause significant morbidity. Macroglossia can be classified as localized or generalized based on the extent of tongue involvement. Congenital causes of localized macroglossia include hemangioma, lymphangioma, and lingual thyroid. Congenital causes for generalized macroglossia include cretinism, primary idiopathic macroglossia, Robinow syndrome, Beckwith-Wiedemann syndrome, and Down syndrome.

The majority of cases of macroglossia are treated surgically. Indications for surgery include airway obstruction, speech difficulties, dysphagia, and cosmetics. The procedure of choice is partial glossectomy, but it probably will not occur until the child is about 2 years of age.

CONGENITAL EPULIS

One type of granular cell tumor that is benign, and only found in the newborn, is the congenital epulis. It is usually found in the anterior maxilla, but occasionally is found in the anterior mandible. It will likely be attached to the alveolar mucous membrane by a pedicle. This smooth-surfaced, nonhemorrhagic tumor occurs more frequently in females than in males. Treatment consists of surgical excision, including the base of the stalk.
THE INFANT'S EXTERNAL EAR

The appearance of the newborn’s external ear is often of great interest to parents, and there is wide variation in sizes, configurations, and positions of the external ear within the range of normal. Although minor variations from “normal” are the primary concern of this booklet, descriptions of more significant aberrations are also included, because they may signal malformations of other organ systems or chromosomal aberrations.

Thorough familiarity with normal as well as minor abnormalities, which can often be traced to familial features, can alert the healthcare professional to those infants who may warrant further investigation.

The external and middle ear are derived from the first and second branchial arches. By the fifth week of gestation, the external ear begins to develop as small buds of mesenchyme along these arches. These hillocks fuse during the 12th week. Following a precise pattern, these six hillocks, together with an auricular fold of the hyoid integument (AF in Figure 2), develop into the auricle. The named hillocks include:

- #1 tragus
- #2, 3 helix
- #4, 5 antihelix
- #6 antitragus

As the face develops, the auricle gradually is repositioned to a more lateral cephalic position from the original location on the side of the neck. At birth, the height of the ear is 66% of adult size. The ear is approximately 85% of full size by age 6 years and 90% by age 9 years. Because the final adult size is nearly achieved by 6 years of age, the contralateral normal ear serves as a reasonable template of the final auricular height.

Figure 1 — The normal newborn ear (Anatomic nomenclature from Dorland's Illustrated Medical Dictionary — 24th ed.)
Normal Variations and Minor Malformations

The complex embryology of the ear, together with inherited familial characteristics, may lead to numerous variations of size, shape, and attachment. Therefore, it is important to inquire whether the condition appears in other family members. Parents can usually be reassured that minor variations have little or no medical significance. Severe malformations of the external ear are rare, but isolated abnormalities occur in approximately 1% of children.

OVERLY PROMINENT OR PROTRUDING EARS

There appears to be a strong familial tendency to prominent ears. In addition, it has been theorized that the high level of circulating maternal estrogens in neonates makes the auricular cartilage soft and malleable. Shortly after birth, the competing muscle forces can affect the shape of the ear. The force of a weak posterior auricular muscle can be overwhelmed by forces in the intrinsic muscles of the anterior surface of the ear. As estrogen levels diminish, the cartilage acquires more elastic resilience, a more retentive memory, and the shape of the cartilage is altered permanently.

The most common causes of protrusion of the external ear are 1) an underdeveloped or flat antihelix; 2) an overdeveloped deep concha; or 3) a combination of both of these features.

ABSENT ANTIHELIX

The antihelix normally forms a symmetric Y in which the gently rolled or folded crest of the root of the antihelix continues upward as the superior crus. An unusually prominent or “lop” ear results from lack of bending of the cartilage that creates the antihelix. It may be improved cosmetically in the neonatal period by applying a firm framework to the pinna for a period of several weeks to several months. Otoplasty for cosmetic correction can be considered in children ages 5 and older because, by this time, the pinna has reached about 80% of adult size.

DARWIN’S TUBERCLE

The auricular tubercle of Darwin is frequently seen where the helix turns downward along the posterior segment. This tubercle is very evident about the sixth month of fetal life, and is believed to be a vestige of the top of formerly erect and pointed ears.

PREMATURE INFANT EAR DEVELOPMENT

Part of the process of determining gestational age can be made by examining the level of development of the external ear of the newborn. Before 34 weeks, the cartilage is not present in any part of the ear, and the pinna is flat, formless and remains folded. At 34 to 37 weeks, the pinna will be curved, soft, and able to recoil. At 37 to 40 weeks, the pinna will be fully formed, firm, and able to recoil instantly. After 40 weeks, there will be thick cartilage, and the ear will be stiff.
Malformations of the External Ear With Potential Teratogenic Significance

When deviations from normal formation or position of the external ear are encountered, they may range from normal variants to obvious abnormalities. Healthcare professionals need to know what signs can alert them to possible abnormalities of internal organs (chiefly urogenital) that were developing embryonically during the same period as the ear. Abnormalities of the ear are also found in a number of clinical syndromes and clinical abnormalities.

Malformed ears are sometimes associated with trisomy 13 (Patau syndrome). Large prominent ears may indicate Fragile X syndrome. The positioning of the ears (low-set) may be an important indicator of urinary tract abnormalities, chromosomal trisomies, or dysmorphic syndromes. Malformed auricles may indicate abnormalities of the internal otic structures that could preclude normal hearing.

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True low-set ears are characteristically found with other significant abnormalities in Fragile X syndrome, trisomy 13 (Patau syndrome), and trisomy 8 (mosaicism) among others. Unilateral renal agenesis is often discovered during the course of the newborn evaluation, and low-set ears are a definitive indicator.

A newborn evaluation should include checking the ears for unilateral or bilateral accessory tragi (skin tags). They usually appear as single, pedunculated, flesh-colored papules in the preauricular region anterior to the tragus. In contrast to the rest of the pinna, which develops from the second branchial arch, the tragus and accessory tragi derive from the first branchial arch. Accessory tragi may occur as isolated defects or in chromosomal first branchial syndromes that include anomalies of the ears and face, such as cleft lip, cleft palate, and mandibular hypoplasia. Accessory tragi are consistently found in Goldenhar syndrome (oculoauriculovertebral syndrome). Surgical excision is appropriate.

Preauricular skin tags in oculoauriculovertebral dysplasia (Goldenhar's syndrome)
BOWING OF LEGS

In the newborn, the imprint of the in utero position may be evident, and can be confused with an abnormality. In utero positioning causes temporary joint and muscle contractions, and affects the torsional alignment of the long bones, especially those of the lower extremities. When this is the problem, there are usually other clinical manifestations, such as increased muscle tone and tendon reflexes, decreased spontaneous movements of the affected limbs, and the posture is usually fixed to the point the infant will not open his hand spontaneously. Flexion abnormalities have also been associated with trisomy 18 (Edward's syndrome). In examining the extremities, the effects of fetal posture should be noted so their cause and usual transitory nature can be explained to parents. This explanation may be particularly important after a breech delivery, because the legs may have been trapped between the body and the uterine wall. Observing the extremities in spontaneous or stimulated activity is the most common way suspicion is aroused that a fracture or nerve injury might have occurred during delivery.

In the typical in utero position, the hips are flexed, abducted, and externally rotated. The knees are flexed, and the lower legs are internally rotated. The combination of external rotation of the hip and internal rotation of the lower legs produces a bowed appearance. This is not true bowing, but, rather, a torsional combination, and will resolve with 6 to 12 months of independent ambulation.

PALMAR CREASES

Prominent creases (flexion creases) appear on the palms of the hands and soles of the feet. The palm normally has three flexion creases. Sometimes, the two horizontal creases fuse to form a single crease, called a single palmar or "simian crease." Palmar creases develop early, by the 11th to 12th week of gestation. Therefore, abnormalities in palmar creases may indicate problems with early development, including disorders such as Down syndrome. However, a single palmar crease appears in approximately 1 of 10 people, so is not a concern in the absence of other clinical symptoms.

PLANTAR CREASES

The plantar surface of the newborn can serve as a useful tool in assessing gestational age of the infant. Before 28 weeks, there will not be any creases on the sole of the foot, and the foot length will be 40 to 50 mm. At 28 to 32 weeks, there will still not be any sole creases, but there may be faint red lines over the anterior aspect of the foot. At 34 to 37 weeks, there will be 1 to 2 anterior transverse creases on the surface. At 37 to 39 weeks, creases will cover the anterior 2/3 of the sole. At term, there will be creases covering the entire plantar aspect of the foot and involve the heel.

CORTICAL THUMB

In early infancy, the thumb may be enclosed within the palm (thumb-in-fist). This phenomenon, known as "cortical thumb," presents no problem, in the absence of other abnormal neurological signs, and is likely to resolve by 7 months of age. In some cases, however, the thumb-in-fist position has been associated with upper motor neuron lesion. When this is the problem, there are usually other clinical manifestations, such as increased muscle tone and tendon reflexes, decreased spontaneous movements of the affected limbs, and the posture is usually fixed to the point the infant will not open his hand spontaneously. Cortical thumb may be particularly important after a breech delivery, because the legs may have been trapped between the body and the uterine wall. In this situation, the fetus is unable to kick optimally, and the incidence of deformations is increased. Observing the extremities in spontaneous or stimulated activity is the most common way suspicion is aroused that a fracture or nerve injury might have occurred during delivery.

In the typical in utero position, the thumbs are flexed, abducted, and externally rotated. The thumbs are flexed, and the lower limbs are internally rotated. The combination of external rotation of the thumb and internal rotation of the lower limbs produces a bowed appearance. This is not true bowing, but, rather, a torsional combination, and will resolve with 6 to 12 months of independent ambulation.
METATARSUS VARUS

Congenital metatarsus adductus, a common problem among infants and young children, is also known as metatarsus varus if the forefoot is supinated as well as adducted. It occurs equally in males and females, and is bilateral in approximately 50% of patients. There appear to be hereditary tendencies. It also tends to occur more often in firstborn compared to later children as a result of the increased molding effect from the primigravida uterus and abdominal wall. Approximately 10% of children with metatarsus adductus may have acetabular dysplasia. Careful examination of the hips is necessary in any child with metatarsus adductus. The treatment is predominantly nonoperative, and varies based on forefoot flexibility. There are limited indications for surgery in the most severe cases.

SYNDACTYLY

Syndactyly is an abnormal fusion of the digits, either partial or complete, which may consist of interdigital webbing of the skin only, of the bony structure, or both. Potential sharing of common important structures between the digits, such as the neurovascular bundle, may occur, and must be considered. There is also a tethering effect on the growth of the affected digit. Referral for delineation of a specific disease and development of treatment strategies is indicated when the condition is recognized.

 Syndromes associated with syndactyly include Apert syndrome, Carpenter syndrome, de Lange syndrome, Holt-Oram syndrome, trisomy 21, trisomy 13, trisomy 18, and Laurence-Moon-Biedl syndrome.

CAVUS FEET

Cavus feet represent an exaggeration in the medial longitudinal arch associated with hindfoot varus and, occasionally, adduction of the forefoot. This type of deformity appears most commonly during the middle childhood years and is usually progressive, leading to considerable compromise of foot function. Since these deformities tend to be rigid, the most important aspect of the patient evaluation is to establish an accurate diagnosis. Possible causes include spinal cord disease and peripheral neuropathies, such as Charcot-Marie-Tooth disease. Aggressive treatment is usually necessary for moderate to severe cavus feet and involves reconstructive surgery.

POLYDACTYLY

Extra digits, or polydactyly, occur as both simple and complex deformities. Skin tags and digit remnants are typically seen near the metacarpophalangeal joint of the small finger or the thumb. Since they do not have palpable bone in the base, or possess voluntary motion, they may simply be ligated or excised in the newborn period. More complex varieties must be formally amputated, which is usually performed at approximately 1 year of age.

Syndromes in which polydactyly commonly occurs include Carpenter syndrome, Ellis-van Creveld syndrome, Meckel-Gruber syndrome, trisomy 13, and Rubinstein-Taybi syndrome.
FLEXIBLE FLATFEET

Hypermobile flatfeet or pronated feet are common sources of concern to parents. In general, these children are asymptomatic, and have no functional limitations. Flatfeet are common in neonates and toddlers because of an associated laxity in the bone-ligament complexes of the feet and fat in the area of the medial longitudinal arch. The treatment of flexible flatfoot is conservative, and these children usually demonstrate significant improvement by 6 years of age. It should be emphasized that the diagnosis of flexible flatfoot is usually not possible before this time. Modified shoes and orthoses do not significantly alter the clinical appearance of the feet. If necessary, feet that are symptomatic usually respond readily to the use of commercially available medial longitudinal arch support.

GENU RECURVATUM

Congenital hyperextension of the knees can be of several types, and genu recurvatum refers specifically to a deformity that manifests as hyperextension only. The diagnosis can be established by a thorough orthopedic examination of the newborn infant. Various causes have been postulated, including environmental and genetic conditions, as well as mechanical factors such as intrauterine trauma of the fetus, lack of amniotic fluid, lack of intrauterine space, and malposition of the fetus. Parents can be reassured that genu recurvatum deformities almost always respond to conservative manipulation of the involved extremity, if the diagnosis is timely and treatment starts soon after birth.

TIBIAL TORSION

Internal tibial torsion is one of the most common causes of in-toeing in children younger than 2 years of age and is secondary to normal in utero positioning. The condition is commonly seen during the 2nd year of life and may be associated with metatarsus adductus. The degree of tibial torsion can be measured by the prone thigh-foot angle (torsional profile). Treatment of internal tibial torsion is by observation. This is a physiologic condition, and spontaneous resolution with normal growth and development can be anticipated, although improvement usually does not occur until the child begins to pull to stand and walk independently.

External tibial torsion is also relatively common and is frequently associated with a calcaneovalgus foot. It is secondary to a normal variation in positioning in utero. External tibial torsion is indicated by an abnormally positive thigh-foot angle (torsional profile), typically 30-50 degrees. Treatment is by observation, following the same clinical course as that of internal tibial torsion.

FEMORAL TORSION

Internal femoral torsion is a common cause of in-toeing in children 2 years of age or older. It occurs more commonly in girls than boys (2:1). The majority of children with this condition have generalized ligamentous laxity. The cause of femoral torsion is controversial. Some believe it is congenital and a result of persistent infantile femoral anteversion, whereas others believe it is acquired secondary to abnormal sitting habits. Clinical features of internal female torsion demonstrate that the entire lower leg is inwardly rotated during gait. The treatment is predominantly by observation. Correction of abnormal sitting habits usually allows the torsion to resolve with normal growth and development, although it may take 1 to 3 years for complete correction to occur.

External femoral torsion, also known as femoral retroversion, is an uncommon disorder unless associated with a slipped capital femoral epiphysis (SCFE).
Hip stability as well as acetabular development can be assessed accurately in neonates and young infants by dynamic ultrasonography. When an unstable hip is recognized at birth, maintenance of the hip in the position of flexion and abduction (“human position”) is usually sufficient. Methods to accomplish this include the Pavlik harness and the Frejka splint. Double and triple diapers, although controversial, are commonly used in newborns with dislocatable hips for 2 to 3 weeks because the splints and harnesses usually do not fit satisfactorily initially.

The Ortolani test is a maneuver to reduce a recently dislocated hip. It is most likely to be positive in infants who are 1 to 2 months of age, because adequate time must have passed for a true dislocation to have occurred. In performing this test, the thigh is flexed and abducted, and the femoral head is lifted anteriorly into the acetabulum. After 2 months of age, manual reduction of a dislocated hip is usually not possible because of the development of soft tissue contractures.

A positive family history and generalized ligamentous laxity are related factors in the occurrence of DDH. Approximately 60% of children with typical DDH are firstborns, and 30% to 50% were in the breech position. The frank breech position, with the hips flexed and the knees extended, is the position of highest risk. All neonates should be screened for DDH. The Barlow test is the most important maneuver in examining the newborn hip. The test actually dislocates an unstable hip. After release of the posterior force, the hip usually relocates spontaneously. It has been estimated that only 1 in 100 newborn infants have clinically unstable hips, whereas only 1 in 800-1000 of these infants eventually experience a true dislocation.
ANNULAR BANDS

Annular bands or constriction rings are relatively common congenital disorders that involve one or more of the extremities. They may consist of simple constriction rings, or rings with deformity of the distal part of the extremity, with swelling and lymphedema. Occasionally, the rings may be deep enough to have produced an amputation. Sometimes there will be an associated syndactyly with the adjacent toe. Annular bands of the lower extremities are frequently associated with clubfoot. Treatment of annular or constriction bands is predominantly observation. If there are deep rings with swelling and lymphedema, surgery may be necessary to relieve the congestion.

TALIPES EQUINOVARUS

Talipes equinovarus (clubfoot) is a common foot deformity that involves not only the foot, but also the entire lower leg. It can be classified as congenital, teratologic, or positional. The congenital clubfoot is usually an isolated abnormality, whereas the teratologic form is associated with a neuromuscular disorder. The positional clubfoot is a normal foot that has been held in a deformed position in utero. The cause of clubfoot is unknown, but inheritance factors are considered likely. The congenital form of clubfoot constitutes 75% of all cases. Examination of the clubfoot demonstrates hindfoot equines, hindfoot and midfoot varus, forefoot adduction, and variable rigidity. Conservative treatment is initiated in all infants, although a significant proportion of children later require surgery.

CALCANEOVALGUS FEET

The calcaneovalgus foot is a relatively common finding in the newborn, and, like clubfoot, is secondary to in utero positioning. A hyperdorsiflexed foot with forefoot abduction and increased heel valgus manifests the condition. It is usually associated with external tibial torsion. It is often unilateral, but occasionally may be bilateral. In utero, the plantar surface of the foot was against the wall of the uterus, forcing it into a hyperdorsiflexed, abducted, and externally rotated position. The position also produces the external tibial torsion. When these two conditions are combined with the normal newborn increased external rotation of the hip, it results in a lower extremity that appears excessively externally rotated.

The typical calcaneovalgus foot requires no treatment. The hyperdorsiflexion of the foot resolves during the first 6 months of life. The external tibial torsion, however, persists, and follows the same natural history as internal tibial torsion.