The Normal Newborn Exam, or Is It?

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In today’s modern world of high technology imaging and sophisticated laboratory examinations, medicine has come to rely on technology much more than in the past. So much so that at times we forget about the power of a thorough physical exam in detecting medical issues. In this article we will explore the normal newborn examination, discuss the importance of knowing normal versus abnormal findings and discuss some common and not so common findings on the newborn examination. In healthy babies, 15% to 20% will have at least one minor anomaly with an associated 3% chance of having a major anomaly. Two, three, or more minor anomalies are found in 0.8% and 0.5% of healthy babies, respectively. In these cases the chances of major anomalies rises to 10% and 20%, respectively [1].

Newborn infants may present to the emergency department for a variety of reasons. Almost all of these derive from the parent’s perception that something is wrong. In each of these cases, the role of the emergency physician is to recognize abnormality and, if no abnormality exists, to alleviate concerns of the parent. Mostly, parental concern stems from conditions that are self-limited or are variants without physiologic consequence. Less common are that these concerns are the presentation of a medical condition that has the potential to worsen or represents underlying illness. Detection of the latter can be life saving. Unfortunately, illness in the newborn is often subtle and difficult to detect. The primary difficulty lies in that the daily activities of a newborn and the newborn’s interaction with the environment are extremely limited. It is therefore imperative that the emergency physician becomes familiar and comfortable with performing a newborn exam.

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Performing a complete and thorough exam on an infant can sometimes be monotonous where elements of the exam can be missed. Therefore, for the benefit of the reader, Box 1 contains a checklist that can be used as a guide while performing such an exam. A complete physical examination may not be possible in the emergency department because of time constraints. However, a systematic approach can allow a thorough examination in a matter of minutes. Focus should be placed on the heart, lungs, and abdomen as well as a general sense of the wellness of the baby. The neurological examination, in particular, is involved and often a complete neurological examination is not necessary. Generally, testing two to three of the primitive reflexes (such as the moro and sucking reflexes) and observing for spontaneous movement of the limbs will give a good sense of the neurological status of the child.

General evaluation

One of the greatest indicators of the wellness of a newborn is the general observation or gestalt that a clinician obtains by simply watching a baby before the start of a physical exam. This can be done while observing the baby in the basinet or in a parent’s arms. Signs of distress such as labored breathing, persistent crying, and so forth are often the first indicators of an impending problem. The newborn infant will often be soundly asleep at the start of the examination and should be sleeping peacefully. Infants who are moaning or grunting during sleep may have underlying illness that should be sought out.

When awakened, it is important that the baby be vigorous and alert. Crying should be energetic and strong. Watch for signs of listlessness and weak cries as harbingers of problems. A baby who does not appear vigorous can have any number of reasons; all of which should raise concern.

Watch the movement and positioning of a child. A newborn, full-term child will lie with his or her upper and lower extremities flexed inward thus showing good tone. A premature or impaired child is more likely to lie at rest with his or her extremities extended from the body showing decreased tone. In addition, examine the movement of the extremities. Each limb should show spontaneous movement. Disuse of an extremity may be the first indication of an underlying pathologic problem.

Vital signs

One of the most difficult parts of pediatrics is recognizing normal versus abnormal because of the great degree of variability in the “normal” ranges of values. This particularly holds true for vital signs. A list of normal ranges can be seen in Table 1. Other clinical indicators should be correlated with the vital signs to aid in determination of significant abnormalities. For
Box 1. The normal newborn exam: a head-to-toe approach

A. Head
- Shape
- Fontanelle
- Lesions/swelling

B. Eyes
- Red reflex
- Extraocular movement
- Pupillary shape/size

C. Ears
- Positioning
- Tags

D. Nose
- Nasal patency

E. Mouth
- Palate
- Dentition
- Oral lesions

F. Neck
- Swelling/cysts

G. Chest
- Asymmetric chest rise with respirations
- Retractions
- Accessory respiratory muscle use

H. Lungs
- Symetric aeration
- Breath sounds

I. Heart
- Murmur

J. Abdomen
- Hepatosplenomegaly
- Cord vessels at birth

K. Genital
- Inguinal masses
- Testicular/scrotal asymmetry
- Genital hypertrophy/lesions
- Rectal patency

L. Extremities
- Tenderness
- Extremity use/range of motion
- Additional digits/tags
- Hip click

M. Neurology
- Tone
- Suckling
- Palmar/pantar
- Moro
example, a respiratory rate of 50 in a newborn who is sleeping comfortably and showing no signs of retractions, and so forth, is likely ok; however, a respiratory rate of 50 associated with retractions, grunting, and nasal flaring indicates a neonate in respiratory distress.

Temperature should always be included in the vital signs of newborns. It has been well documented that temperatures higher than 38°C are associated with increased risk of serious bacterial illness in infants less than 2 months of age. These include infections such as sepsis, meningitis, urinary tract infection, enterocolitis, and osteomyelitis. The latter two often have symptoms that can lead one to suspect them. The first three can be more insidious. As a result, countless infants are hospitalized each year for a “rule out sepsis” work-up to look for these infections.

While a fever is a flag for infection work-up, one must always remember that newborns are just as likely to develop low temperatures or temperature instability in response to infections as well. Our newborn nursery uses temperature ranges from 36.5 to 37.5°C as a “normal” temperature for newborns in the first several days of life. Swings above or below normal should be a signal to evaluate the child closely for the possibility of an underlying infection or other problem.

Hypothermia and hyperthermia may be indicators of sepsis; however, infection is not the only cause that should be pursued. Hypoglycemia, hypothyroidism, and hypoxia can also present with low temperatures. Hyperthermia may be a manifestation of drug withdrawal, intracranial hemorrhage, or adrenal hemorrhage [2].

While fevers/hypothermia are indicators of illness, one must remember that infants are extremely susceptible to outside influence on temperature. A neonate may easily become hyperthermic as a result of an overly warm incubator, or may become hypothermic if left unwrapped in a cool room. After these issues are corrected, continued swings in temperature should yield an infection work-up. It should be noted that bundling an infant will not produce an elevation in the core body temperature [3]. If bundling is suspected as the source of a fever, a rectal temperature should be checked. A rectal temperature higher than 38°C should prompt a sepsis evaluation.

<table>
<thead>
<tr>
<th>Age</th>
<th>Heart rate</th>
<th>Respiratory rate</th>
<th>Systolic blood pressure</th>
</tr>
</thead>
<tbody>
<tr>
<td>Newborn</td>
<td>90–180</td>
<td>40–60</td>
<td>60–90</td>
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<tr>
<td>1 mo</td>
<td>110–180</td>
<td>30–50</td>
<td>70–104</td>
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<tr>
<td>3 mo</td>
<td>110–180</td>
<td>30–45</td>
<td>70–104</td>
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<td>6 mo</td>
<td>110–180</td>
<td>20–35</td>
<td>72–110</td>
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Height, weight, and head circumference (Occipital Frontal Circumference or OFC) are critical to evaluate in the newborn. They can often give clues about other potential problems or abnormalities. In particular, weight is important to evaluate. The average weight of a term neonate at birth is 3.4 kg. Standardized growth charts that can be used when making determinations about small or large for gestational age (SGA or LGA, respectively) infants are available for download from http://www.cdc.gov/growthcharts. Infants are considered appropriate for gestational age (AGA) if they are within 2 standard deviations from the mean. It is important to know the gestational age as accurately as possible, as 1 or 2 weeks’ difference can have significant effects on SGA or LGA determinations. Gestational age can be estimated based on physical characteristics of the neonate including skin creases, external genitalia, ears, breasts, and neuromuscular development[2]. One should also remember to adjust for prematurity when plotting weights of infants during an evaluation. This correction typically continues until the child is 2 years of age. LGA or SGA status should alert the practitioner to evaluate the child carefully for other possible anomalies. Twenty percent of infants with serious congenital anomalies are SGA [2].

LGA infants may be large simply due to familial inheritance. Simply stated, large parents will often have large infants. Additionally, increased maternal weight gain during pregnancy may translate into an LGA infant. Of all causes of LGA, diabetes in the mother is most common. Infants are at particularly increased risk if the mother’s blood sugars are poorly controlled during the last trimester. (Of note, diabetes mellitus types 1 and 2 are linked to significantly increased risk of perinatal mortality and morbidity[4]. Associated conditions include postnatal hypoglycemia, cardiac septal hypertrophy, small left colon syndrome, and meconium plug, as well as others.)

While being an infant of a diabetic mother is the most common cause of LGA, there are several genetic syndromes that can cause babies to be LGA. Among them are cerebral gigantism (Soto’s syndrome), Beckwith-Wiedemann syndrome, Simpson-Golabi-Behmel syndrome, and 11p trisomy. These syndromes all have their own characteristics and discussing them is beyond the scope of this paper; however, it is important to consider such syndromes when assessing an LGA baby. For a more thorough discussion of syndromes and their associated clinical findings, one can refer to Smith’s Recognizable Patterns of Human Malformation [5].

OFC should be examined to assess for microcephaly or macrocephaly. The OFC can be particularly helpful when there is concern for hydrocephalus. Macrocephaly can be an isolated finding or associated with other anomalies. As an isolated finding, it is often familial with an autosomal dominant inheritance. When this is in question, the OFC of the parents can be determined and plotted on nomograms. Nomograms for head circumferences of 18 year olds can be used to extrapolate to the parents’ OFC percentile. Head growth for infants with familial macrocephaly should follow standard growth. While familial macrocephaly may be the cause, one
should not assign this diagnosis without ensuring that other causes are not present. In particular, hydrocephalus is the most common cause of macrocephaly and should therefore trigger an evaluation [6]. Hydrocephalus often presents with widening sutures, full feeling, possibly bulging fontanelles, and a rapidly expanding OFC. Macrocephaly can also be associated with intracranial hemorrhage (eg, subdural or epidural bleeds), enlarged brain tissue (macrencephaly), or thickening of the skull bones [6]. Daily examination while the neonate is in the hospital can identify widening suture lines or a fontanelle that is becoming more full and tense. These can be earlier indicators of a quickly enlarging head and thus an underlying pathologic problem.

Head

The newborn head shows a great degree of variability in the “normal” examination, as well as a large number of findings that are due to molding in the vaginal canal and birth trauma. These normal variants and benign birth trauma findings can make it difficult to find true pathological lesions.

A brief review of anatomy reveals that the newborn skull is composed of many bones. The “neurocranium” (the portion of the skull that encompasses the brain) is composed of eight bones. The facial skull is composed of 14 irregular bones. Most of these bones are not fused together at birth and are separated by fibrous webs, known as suture lines. Most physicians are aware of the anterior and posterior fontanelles; however, there are a total of six fontanelles. In addition to the two previously mentioned fontanelles, there are two pairs of lateral fontanelles known as the sphenoidal or anterolateral fontanelles and the mastoid or posterolateral fontanelles. The four lateral fontanelles generally fuse in infancy and are less clinically significant. The posterior fontanelle begins to close after the first few months and is generally not appreciable by 1 year of age. The anterior fontanelle is generally no longer palpable by 18 months of age [7]. Sutures are generally closed by age 12, but complete fusion continues until into the third decade [8].

Often, most commonly following vaginal birth, the newborn skull will show a significant degree of molding. While this can be distressing to new parents, reassurance is the rule, as molding will resolve relatively rapidly, often over the course of 3 to 5 days. The suture lines allow the bones to shift during the birth process. Often, they are molded into a cone shape allowing for an easier passage of the head through the birth canal. Molding can be significantly more pronounced after a long labor and delivery course. It is important to recognize molding as the OFC can increase by up to 1 cm as the head shape resolves [2].

Localized edema of the scalp can occur following birth and is referred to as caput succedaneum. This is the most common scalp injury due to birth trauma [9]. Caput succedaneum is particularly common after vaginal delivery, but can occur following cesarean section delivery as well. This edema is
generally serosanguinous, and occurs due to pressure on the head after being constricted against the uterus, cervix, or vaginal vault [2]. Vacuum-extracted delivery commonly results in large caput areas because of the addition of negative pressure in the area of the vacuum attachment.

Typical caput succedaneum size is only a few centimeters in diameter; however, they can be significantly larger. Spontaneous resolution is the rule for caput succedaneum and, regardless of size, edema generally resolves within 48 hours [2]. Examination of the scalp will show a boggy, somewhat ill-defined area that may cross suture lines. This feature helps distinguish caput succedaneum from another commonly found head lesion following birth, a cephalhematoma.

Cephalhematomas result from injury to the blood vessel found in the subperiosteal area. They can be distinguished from caput succedaneum in that they do not cross suture lines. Cephalhematomas occur in 1.5% to 2.5% of all deliveries. Extraction deliveries are at a greater risk of developing a cephalhematoma with the incidence in forceps deliveries at 4.1% and vacuum extraction ranging from 9.8% to 14.8% [9,10]. Overly large hematomas or those that persist may be the first indication of an underlying bleeding disorder and should prompt evaluation.

While caput succedaneum usually resolves in 48 hours, cephalhematomas may take several days to weeks to fully resolve (further allowing them to be distinguished from a caput). Cephalhematomas will resolve without intervention. Aspiration of the cephalhematoma is generally not indicated, as it leads to an increased risk of infection within the cephalhematoma [11]. The exception to this is a cephalhematoma that is expanding, showing overlying erythema, or having other signs of infection. While infection is rare, it can occur, occasionally in association with scalp electrode placement. Here, aspiration may be required for diagnostic purposes.

Less common, but important to consider, is a subgaleal bleed. Clinical presentation of subgaleal bleeds is generally described as the cranium having a diffusely boggy or soft swelling of the scalp. While subgaleal bleeds may occur following difficult deliveries without extraction assistance, they most commonly will occur following vacuum extraction. Occurrence with vacuum extraction has been shown to be 59 (0.59%) in 10,000 births, compared with 4 (0.04%) in 10,000 births via spontaneous vaginal deliveries without instrumentation [12]. Infants delivered with vacuum extraction should be monitored with serial scalp exams for the development of a subgaleal bleed. These bleeds can be life threatening as large amounts of blood can fill the potential space that lies between the galea aponeurosis and the periosteum. This space extends from the orbital ridges to the superior aspect of the neck and laterally to the ears. Mortality from these bleeds approaches 22% [13]. Infants may require intravenous fluid support, blood transfusions, or antibiotic management of infections that may establish themselves in the bleed area. Additionally, because of the increased red cell breakdown, these infants are at risk of hyperbilirubinemia.
Normally, the bones of the head are separated by suture lines to allow for molding of the skull during birth as well as head growth. Head growth is primarily driven by brain growth. As the brain grows, the neurocranium expands to allow more room. Brain growth is at a tremendous rate in utero and continues to rapidly grow during the first 3 years of life. At birth, the infant brain is about 40% of adult volume. By age 3, it has reached nearly 80% and by age 7 it is at 90% of adult size [8]. When a suture line is prematurely fused, termed craniosynostosis, the growth capacity of the skull in that plane is significantly reduced, but continues in the plane perpendicular to the fused suture.

Craniosynostosis can occur in nonsyndromic and syndromic patterns. Its incidence is 1 in 1700 to 2500 births and 1 in 25,000 births, respectively [14]. Simple craniosynostosis involves a single suture, and compound craniosynostosis involves two or more suture lines. Most commonly involved is the sagittal suture that is affected 40% to 60% of the time, followed by the coronal (20% to 30%), metopic (less than 10%) and finally the lambdoid suture. Isolated lambdoid synostosis occurs in approximately 3 of every 100,000 births [8,14].

The major forms of hereditary craniosynostosis exhibit autosomal dominance, but can have a significant degree of variability in penetrance and expressivity. Fibroblast growth factor receptors (FGFRs) have been shown to be associated with suture formation and mutations in these genes have been implicated in several diseases. For example, mutations in the \textit{FGFR2} gene cause Apert’s syndrome and Crouzon’s disease [15].

Much more common than craniosynostosis is positional plagiocephaly (simple cranial shaping). At first glance, lambdoid synostosis and occipital plagiocephaly can look almost identical to each other. However, a thorough physical exam can readily distinguish between the two entities. The variance in head shape is best appreciated from above. With positional plagiocephaly, the affected side will show forward positioning of the ear in response to the bones being pushed forward by repeated positioning on the affected side. In craniosynostosis, the affected side will have a posteriorly displaced ear, because of failure of bone growth to move the ear forward to its normal position. Mild plagiocephaly often can be treated with simple alternating of the baby’s sleep position such that the head is in a different position from night to night. More severe plagiocephaly may require a specially fitted helmet to correct the misshapen skull. The helmet molds the skull bones back into place as the skull grows by gently putting pressure on the areas that are misshapen to move them back to correct alignment. After the initial fitting period the helmet is worn for 23 hours a day. For this to be effective, it should be done before the sutures begin to fuse, ideally by age 4 to 6 months. Rarely, these devices may fail and surgical correction may be needed. If so, the optimal time to do so is before 1 year of age [16].

Fontanelle size is often not appreciated, but can be an indicator of underlying pathology. Anterior fontanelle size is measured by averaging the
anteroposterior and transverse lengths. Fontanelle size is influenced by brain growth due to its stimulation of skull bone growth, suture development, and subsequent osteogenesis [17]. On the first day of life, the average infant fontanelle size is varied from 0.6 cm to 3.6 cm [18]. A large number of disorders are associated with an abnormally large fontanelle. Most commonly, a large fontanelle is found in congenital hypothyroidism, achondroplasia, Down syndrome, and increased intracranial pressure.

Small fontanelles can be a sign of normal early closure, as 1% of fontanelles will close by age 3 months. Additionally, molding can make it difficult to appreciate a truly open fontanelle. However, a small fontanelle can be the harbinger of a pathologic problem and should be investigated. Typically, a small fontanelle is associated with microcephaly. It can be associated with craniosynostosis as previously discussed, but can also be seen with abnormal brain development, fetal alcohol syndrome, congenital infections, and many genetic syndromes [17]. Conversely, soft tissues within the fontanelle may shift depending on intracranial pressure such that bulging of soft tissue within the fontanelle implies increased pressure within the cranium and sunken soft tissues implies dehydration.

Eyes

A pediatrician often examines the red reflexes of the eye at nearly every well-child visit in search of the all elusive retinoblastoma. However, the red reflex can give much more information than simply screening for tumors. A white reflex, known as leukocoria, from the eye can be associated with many other conditions including cataracts, retinal detachment or dysplasia, papillary membranes, and vitreous opacities. The differential expands much beyond this, and as such, findings of leukocoria require referral to an ophthalmologist for further evaluation. Bear in mind that babies with darker skin tones can have reflexes that do not look bright red; however, they should not be white. In addition to examining for leukocoria, the clinician can look for colobomas, examine the pupillary reflex, and look for strabismus using the ophthalmoscope.

Retinoblastoma does, indeed, present with leukocoria. It is the most common malignant intraocular tumor in childhood, with an incidence of 1 in 17,000 [19]. It may present unilaterally or bilaterally, with 30% of cases being bilateral. It is associated with mutations or deletions of the q14 band of chromosome 13 [20].

Congenital cataracts are present from birth, by definition; however, they may not be picked up until later in the first years of life. This is often because some cataracts are progressive and become larger with time. Congenital cataracts occur in 0.44% of live births, and account for 11.50% of blindness in preschool [21]. Of these, 23% are inherited (most commonly autosomal dominant with complete penetrance) and as many as 60% of bilateral cataracts are associated with metabolic (as in galactosemia) and systemic...
disease. Associated systemic diseases include congenital infections (rubella, toxoplasma, cytomegalovirus, and herpes simplex virus in particular), trisomies (13, 18, and 21), hypoglycemia, and prematurity [22]. On physical exam, one may see leukocoria, strabismus, nystagmus, light sensitivity or decreased visual acuity (particularly later in life). Smaller cataracts may appear as black spots in the red reflex.

When examining the eyes for red reflexes, one may notice a dysconjugate gaze. Gaze can be assessed by visualizing the papillary light reflex (using the ophthalmoscope, but not looking through its lens). At birth, dysconjugate gaze is not particularly concerning. Eyes may often be crossed or divergent. These findings generally correct without intervention by 2 months of age [23]. Dysconjugate gaze beyond this time frame may be an indication of an underlying defect in the function of one or both eyes. Dysconjugate gaze as a function of eye tracking is termed strabismus. In this condition, the weaker of the two eyes is typically patched to allow for appropriate development. In cases where strabismus is not corrected, visual acuity will be lost in the weaker eye. This condition of lost vision is termed amblyopia.

Following birth, subconjunctival hemorrhages are common. They represent a burst blood vessel and do not present a danger to the infant. They typically reabsorb in 1 to 2 weeks. After birth, they can be caused by increasing intraocular pressure, such as through coughing or sneezing, or they may occur spontaneously. One should be aware that a subconjunctival hemorrhage may be a sign of trauma that a parent may not have witnessed or may not be forthcoming so the eye should be examined thoroughly for other signs of injury.

Blue sclera is often one of the telltale signs of osteogenesis imperfecta. While this is the most commonly thought of diagnosis, blue sclera can appear in many different syndromes, including Ehlers-Danlos syndrome and Hallermann-Streiff syndrome. In addition, healthy infants may have a bluish tint to the sclera since the sclera is thinner than in adulthood [21].

While examining the eye, it is important to examine the iris as well as the sclera and pupil. The normal iris of neonates is typically blue or bluish-gray in light-skinned babies and can be darker gray or brown in infants with darker skin [21]. The iris should be examined for colobomas, which appear as a break in the continuity of the iris. Colobomas may be an isolated finding; however, they often are associated with other congenital anomalies, so their presence should prompt a full examination and work-up for other findings [21]. Brushfield spots are areas of stromal hyperplasia surrounded by areas of hypoplasia giving a speckled look to the iris. These can be seen in healthy patients, but are much more common in those affected by Down syndrome. Upwards of 90% of Down syndrome patients will have Brushfield spots [24], which can be seen in Zellweger’s syndrome as well.

Nystagmus is repetitive, involuntary, rhythmic movements of the eye in a particular direction. Several types of nystagmus exist, including horizontal,
vertical, and rotary nystagmus. Nystagmus in neonates may be benign or may represent a pathological process. It can be secondary to retinopathy of prematurity, prematurity itself, or normal physiologic reflexes. An entity known as transient neonatal nystagmus typically develops before age 10 months (mean 2.7 months) and resolves spontaneously by age 12 months. The cause is not yet determined [25].

When examining the eyes, one should be on the lookout for opsoclonus in neonates. Opsoclonus is rapid, irregular, and nonrhythmic movements of the eyes. It should not be confused for nystagmus, which is a rhythmic beating of the eyes. Opsoclonus shows beating in horizontal and vertical directions. It may be seen in association with an acute febrile illness, especially those caused by Epstein-Barr virus, varicella, Coxsackie viruses, and West Nile virus [21,26]. When seen in conjunction with myoclonus, so called opsoclonus-myoclonus syndrome, it has been associated with neuroblastoma in 2% to 3% percent of cases [26]. Opsoclonus-myoclonus is often referred to as “Dancing Eyes–Dancing Feet Syndrome” because of its clinical presentation.

Ears

Cranial molding from birth can move the ears into various positions, so molding should be taken into account when examining the ears. One should examine the ears for their position, noting low-set ears, as this has been associated with several syndromes, and malformation of the auricle. Preauricular pits or skin tags are common, and often are hereditary. These are generally inconsequential and no further work-up is needed when they are seen unless there is indication of other anomalies that are present. Hereditary preauricular pits can be associated with deafness and this should be followed as the infant grows [2]. Additionally, ear anomalies can be associated with genitourinary anomalies. As such a renal ultrasound is generally indicated in these patients.

Mouth

Several variations of a normal exam present themselves in the mouth. These include natal teeth and Epstein’s pearls. In addition, one may see a cleft lip or palate. Natal teeth are defined as teeth that erupt at any point before 30 days of life. These occur in about 1 in every 3000 births and almost always involve the mandibular central incisors [27]. Structurally, they are similar to normal teeth; however, they generally lack a substantial root system. As such, they often fall out on their own at some point. There is no need to have them removed unless there is concern for aspiration of the tooth should it come loose on its own, or if the tooth prevents proper feeding [28].
Epstein’s pearls and Bohn nodules are common findings in neonates. They are often yellowish or white and slightly raised, giving the appearance of a pearl. Bohn nodules are located on the alveolar ridges, and Epstein pearls are those located near the midpalatal raphe at the junction of the hard and soft palates [28]. They are remnants of embryonic development of the dental lamina and will resolve without treatment.

Cleft lip and palate represent the most common anomaly of the head and neck in newborns. Individually and their various combinations are second only to club feet in regard to neonatal birth defects [29]. Cleft lip and palate are two distinct findings; however, the combination of the two is found more commonly than isolated cleft lip or palate with 46% representing combined cases, 21% isolated cleft lip, and 33% isolated cleft palate. Cleft lip carries an incidence that varies with ethnicity ranging from 0.41 per 1000 in African Americans to 2.1 per 1000 in Asians. White ethnicities show an incidence of 1:1000 [30]. The left lip is most commonly involved, followed by the right, then bilateral clefts. Surgical correction often produces very good cosmetic results and should be considered early in cases of poor nutritional intake and abnormal speech development.

While most clefts are quite obvious upon examination, mucosal and submucosal clefts can be less easy to identify visually. Generally, the clinician is able to palpate the cleft using a finger to examine the upper palate of the infant.

Neck

Congenital muscular torticollis is the third most common musculoskeletal anomaly of infants. It follows behind club foot and dysplasia of the hip and can have an incidence upwards of 1 in 250 live births [31]. The exact cause is unknown; however, it is known to be found more commonly in breech and forceps deliveries. In approximately two thirds of cases, a mass can be felt in the affected sternocleidomastoid muscle. It presents with the infant having its head flexed and the chin facing the direction opposite the affected muscle [32]. Occasionally, this contracture of the muscle can lead to plagiocephaly with abnormal placement of the eye and ear on the affected side. Physical therapy is generally therapeutic. Rarely, surgical release of the sternocleidomastoid muscle is needed [33]. If the torticollis is not corrected by age 1, plagiocephaly tends to be persistent [32].

The neck should be examined for any pits or other anomalies. These may represent branchial cleft anomalies, cysts, or sinuses. These branchial arch remnants will be found in the pinna of the ear, the preauricular area, or the lateral neck When present, these anomalies may be associated with systemic syndromes such as Goldenhar syndrome, Pierre-Robin Association, Treacher-Collins syndrome, and Hallerman-Streiff syndrome [14]. A midline neck lesion may represent a thyroglossal duct cyst or cervical cleft.
Chest

Clavicle fractures are one of the most common chest birth injuries, with an incidence of 0.2% to 3.5% [34]. They are identified by palpating crepitus in the clavicular region. Fractures can be confirmed by radiograph if necessary. No treatment is needed for clavicular fractures as they heal well, even in the presence of significant angulation.

When examining the lungs of a newborn, one should assess for symmetry and equal chest excursion as well as adequacy of air exchange. Examination of the lungs includes assessment of the respiratory rate and other indicators of respiratory distress including nasal flaring, grunting, or retractions.

Newborns will frequently exhibit transient tachypnea of the newborn (TTN), especially following cesarean section deliveries. It affects approximately 0.3% to 0.5% of newborns and presents with tachypnea, increased oxygen requirements, and a lack of hypercapnia on blood gas [35]. It is thought to represent delayed absorption of fluid in the lungs. Recovery is the rule, although infants may need ventilatory assistance for a time period. Respiratory distress typically will present a few hours after birth and resolves in 24 to 72 hours [35].

Cardiovascular

The cardiovascular exam can be difficult in a newborn because of their often rapid heart rate. To briefly review neonatal physiology of the heart, one must have a basic understanding of fetal cardiac physiology. Before birth, the infant does not use the lungs for oxygenation. As such, pulmonary pressures are high and blood is preferentially shunted through the foramen ovale and the ductus arteriosus to return to the systemic side of blood circulation. At the time of birth, many transitions must be completed, including closure of the foramen ovale and a decrease in pulmonary pressures to allow blood to flow through the lungs. The ductus arteriosus typically closes 10 to 15 hours after birth. Full permanent fusion may take up to 3 weeks [36].

Auscultation of the heart sounds of a neonate typically reveals a single S1 and a single, or very slightly split, S2 because of the continued relatively high pulmonary pressures. It can take up to 4 to 6 weeks for pulmonary pressures to fully drop to their baseline [37]. As this occurs, the S2 becomes more physiologically split. Typically, S1 is louder near the apex of the heart and S2 is louder near the base. An S3 may be heard near the apex and is considered normal. S4 is never normal in a newborn. Because of rapid heart rates, it can be difficult to truly distinguish the location of a gallop sound. As such, they are often referred to as a summation gallop.

Palpation of peripheral pulses is important to perform in newborns. Weak femoral pulses in comparison with brachial pulses (especially of the right arm) can be an indicator of coarctation of the aorta.
Several murmurs can be heard at birth. These may or may not be innocent in nature. As the pressures in the pulmonary vasculature drop, a ductus that remains patent (PDA) may begin to shunt left to right, creating a murmur often heard best just below the left clavicle near the mid clavicular line [36]. This murmur is typically not heard at birth, as the pulmonary pressures have not yet dropped sufficiently to allow enough shunting to create a murmur.

An innocent flow murmur, termed a newborn murmur, is commonly heard at birth. It is characterized as a systolic ejection murmur located in the left lower sternal border area that is vibratory in nature. This murmur is likely to represent rapid blood flow and is not considered pathologic. Innocent flow murmurs typically do not radiate to other areas and are graded as a I–II/VI in strength.

One may hear a murmur from peripheral pulmonic stenosis (PPS) equally in the left upper sternal border area, back, and axilla [37]. Typically this murmur will be a soft, I–II/VI high-pitched systolic ejection murmur. PPS murmurs typically resolve by age 2 as the sharp turns in the pulmonary vasculature resolve.

Other commonly described innocent murmurs include Still’s murmurs, pulmonary ejection murmurs, and venous hums. These murmurs, in contrast to those previously described, are not typically heard in the newborn period.

Innocent murmurs tend to have a vibratory sound. More harsh-sounding murmurs should be evaluated to determine their etiology. Such harsh-sounding murmurs may represent valvular stenosis or regurgitation among other pathological causes.

Holosystolic murmurs are never innocent. Most commonly they represent ventricular septal defects (VSD). They may also represent mitral or tricuspid regurgitation or more complex variations of VSDs, such as atrioventricular septal defects (AVSD). VSDs are the second most common congenital cardiac anomaly (falling only behind bicuspid aortic valves). They represent 15% to 20% of congenital cardiac defects [38]. A VSD murmur typically will be a harsh-sounding holosystolic murmur at the left lower sternal border. Smaller VSDs typically will have a louder sound as blood is forced across with a higher velocity. Importantly, VSD murmurs may not be present at birth because of elevated pulmonary pressures. As the pressures drop, the murmur becomes more pronounced.

Certain anomalies associated with murmurs are also associated with systemic syndromes. For example, approximately 30% of patients with a complete atrioventricular septal defect will have Down syndrome [38]. Noonan syndrome is associated with a rare cardiac defect—supravalvular pulmonic stenosis. The murmur of this anomaly is essentially identical to pulmonic stenosis, except that it is located slightly higher on the chest and an ejection click is not heard [37]. If a murmur is detected in the emergency department that is concerning for pathology, evaluation is warranted. This should
include a chest x-ray and an EKG. If abnormalities are found, the neonate should be admitted for further evaluation. If no abnormalities are detected and the child appears well, follow-up with a pediatric cardiologist as an outpatient is reasonable.

**Abdomen**

In examining the abdomen, the clinician should assess for typical abdominal components. This includes assessing for hepatomegaly, splenomegaly, and palpable enlarged kidneys. Abdominal palpation is best accomplished using the flats of the fingers rather than the tips, as they are more sensitive to masses and so forth. The liver is often felt approximately 1 cm below the costal margin. This degree of extrusion is considered normal.

In general unless it is enlarged the spleen should not be detected by palpation. An enlarged spleen in the setting of prolonged or severe jaundice should alert the clinician to the possibility of a hemolyzing state, as may be found in Rh or ABO incompatibility or other red cell morphological defects such as spherocytosis or elliptocytosis.

While attempting to palpate for an enlarged spleen, one can also palpate for enlarged or cystic-feeling kidneys. Autosomal recessive polycystic kidney disease can present with enlarged kidneys at birth [39]. Rarely, one can see congenital absence of the abdominal muscles in combination with cryptorchidism and urinary tract anomalies, termed Prune Belly Syndrome or the Eagle-Barrett triad [39]. Prune Belly syndrome has an incidence of 1 in 40,000 births, and 95% of cases are in males [40].

The umbilical cord should be inspected to ensure the presence of a single umbilical vein and two umbilical arteries. A single umbilical artery (SUA) is found in approximately 0.2% to 1% of newborns [41]. Studies have shown that infants with an SUA have a threefold higher incidence of severe renal anomalies and a sixfold higher incidence of any renal malformation when compared with the general population [42].

A scaphoid abdomen and respiratory distress at birth should raise concern for a congenital diaphragmatic hernia (CDH). When the possibility of CDH exists, the infant should be intubated in an effort to prevent the swallowing of air that occurs with spontaneous breathing or with bag-valve mask ventilation [2]. Air in the intestines will lead to worsening respiratory distress as the expanding intestines further compress the lungs and mediastinum. It should be noted that absence of a scaphoid abdomen does not rule out CDH.

The differential diagnosis for hepatomegaly in a neonate is quite extensive. First, it is important to recognize true hepatomegaly. Hepatic size can be assessed by various means, including percussion of liver span and palpation of the liver edge. When one palpates for the liver edge, it is recommended to start palpating in the lower quadrants to ensure that extreme hepatomegaly is not missed. In general, a liver edge palpable beyond 3.5 cm
below the costal margin is considered enlarged. At 1 week of age, a normal liver span is 4.5 to 5.0 cm [43]. An enlarged edge below the costal margin in the presence of a normal liver span may be caused by depression of the liver downward due to lung hyperinflation or other anatomical causes.

Once hepatomegaly has been ascertained, other important factors to note are the presence or absence of jaundice, splenomegaly, and other physical anomalies or systemic symptoms. These will help guide the examiner’s work-up to determine underlying causes.

Genitourinary tract

Several anomalies can be found when evaluating the genitourinary tract. As such, a thorough examination is important to identify any such findings that may exist. Neither girls nor boys are exempt from anomalies.

Newborns should be examined for inguinal hernias, hydroceles, varicoceles, undescended testes, or signs of hypospadias and other urethral anomalies. Inguinal hernias have an incidence of 0.8% to 4.4% in boys, representing one of the most common surgical issues for newborns [44]. They can present as a bulge in the inguinal area or may have palpable bowel in the scrotum [45]. Inguinal hernias develop when the processus vaginalis fails to close, allowing enough of an opening to let bowel pass through. Approximately 60% involve the left side, 30% the right, and 10% will be bilateral [44]. When an inguinal hernia is identified, light, constant pressure should be applied to the herniated bowel to reduce the bowel back into the abdomen. Occasionally, the bowel may become trapped and nonreducible. This represents an incarcerated hernia and is a surgical emergency. Incarceration of an inguinal hernia may compromise blood flow to the intestines or the scrotal contents resulting in damage to the end-organ. Girls are susceptible to inguinal hernias as well, although the male:female ratio is 6:1 [44]. Physical examination generally reveals a lump in the inguinal area.

When the processus vaginalis fails to fully close but allows only fluid to pass into the scrotum a hydrocele results. Hydroceles are most commonly present at birth, although they rarely can arise later. Transillumination can be used to aid in the diagnosis of a hydrocele as they tend to easily and uniformly light up, although bowel can occasionally transilluminate as well, making the distinguishing of the two difficult at times. Hydroceles do not extend into the inguinal canal. Typically they resolve within 12 to 28 months, so surgical repair is generally deferred [44].

Varicoceles result from dilation of the pampiniform plexus and internal spermatic vein. Almost always, they occur on the left, and are often described as feeling like a “bag of worms.” Varicoceles are typically more easily seen with the patient upright, as this increases the hydrostatic pressure on the venous plexus. They should reduce easily or spontaneously with supine positioning. Any varicocele that does not reduce or is located on the right side should be evaluated by ultrasound, as failure to reduce is an indication
of a possible blockage in venous drainage. This may be the result of an abdominal mass [45]. With typical varicoceles, surgical correction can be undertaken but such a need is debated as most are asymptomatic [39]. If correction is desired, it typically is not done in the neonatal period.

At birth, full-term boys will experience an undescended testis (cryptorchidism) about 3% to 4% of the time. By age 1, only 0.3% persist [39]. If one fails to locate a testis in the scrotum, examination of the inguinal canal area should be performed. Often, testes are not truly undescended but are retractile. These testes can be brought fully into the scrotum, although it may be difficult. Retractile testes are the result of a hyperactive cremasteric reflex and will ultimately settle into the scrotum without intervention [45].

True undescended testes cannot be manually drawn into the scrotum on examination. These will require intervention to draw the testes down into the scrotum if they have not descended by 6 months of age. Typically this is achieved by surgical orchiopexy but treatment with hCG can be attempted (success with hormonal treatment can reach approximately 30% to 40% but more often failure results) [39]. Undescended testes should be brought into the scrotum by intervention as this allows for better examination for malignancy as they have a 4 to 10 times higher risk than descended testes. Seminomas are most commonly seen [39]. Approximately 10% of those affected with undescended testes will have bilateral involvement. When bilateral undescended tests are seen, there should be suspicion of an underlying congenital adrenal hyperplasia with virilization of a female infant. This is particularly true if hypospadias is also seen [38].

Ambiguous genitalia result from virilization of females or undermasculinization of males. The range of phenotypes can run from almost normal female to almost normal male anatomy. In boys, ambiguous genitalia commonly result from problems in androgen synthesis (such as 17 alpha-hydroxylase deficiency) or end-organ resistance to these hormones (such as in 5 alpha-reductase deficiency or androgen insensitivity). In girls, congenital adrenal hyperplasia (CAH) is the leading cause of ambiguous genitalia. CAH occurs with an incidence of 0.06 to 0.08 per 1000 live births; 90% of these cases are a result of 21-hydroxylase deficiency [46].

During examination of the genitalia, the clinician should evaluate for clitoromegaly, micropenis, bifid scrotum, or fusion of the labia resulting in a scrotum-like appearance [47]. Increased pigmentation of the skin can be seen with CAH because of increased ACTH concentrations in an attempt to stimulate cortisol production.

**Extremities**

When examining the extremities, one should examine all four extremities for anomalies. Both hands and both feet should be examined for absent or supernumary digits. Polydactyly frequently is an isolated finding but may be associated with other malformations. While spontaneous occurrences are
frequent, a family history of polydactyly yields up to a 10-fold increase in occurrences. Polydactyly of the fingers involving the “pinky” finger side is the second most common anomaly of the hand with an incidence of 1 in 3000 [48]. Involvement can range from a simple skin tag that can be removed by tying a suture at the base to a fully formed digit including bony structures. These more complex digits require surgical removal. While they rarely produce a functional deficit, cosmetic reasons often lead to removal. Surgery is generally performed between 1 and 2 years of age [48]. Polydactyly of the toes commonly occurs as well. Surgical correction, if needed, for supernumerary toes generally occurs between 6 months and 1 year.

The palms of the hands should be examined for the presence of a single transverse palmar crease. While a single palmar crease is associated with several syndromes, most notably Down syndrome, approximately 4% of the healthy population will have a unilateral single palmar crease and 1% will have bilateral single transverse palmar creases.

Talipes equinovarus, commonly known a clubfoot, is a relatively common deformity with an incidence reported at 1 in 1000 live births. Bilateral involvement occurs in 30% to 50% of cases. Male to female ratios are 2:1 [48].

Clubfoot is a complex anomaly. It is composed of four main components: (1) the forefoot is inverted and adducted, (2) the heel and hindfoot are inverted, (3) limitation of extension at the ankle and subtalar joint, and finally (4) internal rotation of the leg [49]. Physical examination reveals inability to bring the foot fully to midline. Initial correction is attempted with serial casting. Progressive casting is successful in most patients; however, in some cases, surgery may be required [49]. If needed, surgery is generally performed between 6 and 9 months.

**Neurological**

An extensive neurological examination can be quite time consuming; however, all infants should receive at least a limited neurological examination including tone, primitive reflexes, and a gross assessment of muscular/sensory status and cranial nerves. A list of primitive reflexes and a short description of each can be found in Box 2. As in older children, a systematic approach is helpful. Often one can begin with a general assessment, followed by cranial nerves, motor function, sensory function, and reflexes. A good assessment of neurological status can often be obtained simply by careful observation of the infant. One should observe movements of the face for symmetry. Arms and legs should be watched to assess for movement or lack thereof. At birth, full-term infants show a flexed posture of the extremities and have the ability to raise their heads in the vertical midline plane, although they may still be quite unsteady [50]. Movements of the extremities should be generally smooth without a significant amount of jerking. The cry should be strong and vigorous. Often neurological problems
Box 2. Common Primitive Reflexes

Rooting Reflex
Touch newborn on either side of cheek and baby turns toward stimulus.

Walking Reflex
Hold baby up in vertical position. As feet touch ground, baby makes walking motion.

Tonic Neck (Fencing) Reflex
Rotate baby’s head leftward and the left arm stretches into extension and the right arm flexes up above head (opposite reaction if head is rotated rightward).

Moro Reflex (Startle Reflex)
Hold supine infant by arms a few inches above bed and gently release infant back to elicit startle. Baby throws arms out in extension and baby grimaces.

Hand-to-Mouth (Babkin) Reflex
Stroke newborn’s cheek or put finger in baby’s palm and baby will bring his fist to mouth and suck a finger.

Swimmer’s (Gallant) Response
Hold baby prone while supporting belly with hand. Stroke along one side of spine and baby flexes entire torso toward the stroked side.

Palmar
Stroke inner palm/sole and toes/fingers curl around (“grasp”) examiner’s finger.

Plantar
Stroke outer sole (Babinski) and toes spread with great toe dorsiflexion.

Doll’s Eyes
Give one forefinger to each hand (baby grasps both) and pull baby to sitting with each forefinger. Eyes open on coming to sitting (like a doll’s).

Protective Reflex
Soft cloth is placed over the baby’s eyes and nose. Baby arches head, turns head side to side, and brings both hands to face to swipe cloth away.

Crawling Reflex
Newborn placed on abdomen and baby flexes legs under as if to crawl.
will produce an altered cry such as a weak cry or a particularly high-pitched cry.

Cranial nerve (CN) assessment can begin with the eyes. By 28 weeks an infant's neurological system has developed enough to produce a blink when a bright light is shone in the eyes (CN II and VII). Between 28 and 32 weeks the papillary reflex develops (CN II and III) [2]. CN V can be tested via corneal reflex or with light touch/pin prick to the trigeminal branch areas producing a withdrawal from the stimulus. Additionally, full-term infants can often fixate and track on large objects thus allowing for testing of CN II, III, IV, and VI. It should be noted that a dysconjugate gaze is not abnormal when a neonate is not fixing on an object [2]. Facial asymmetry may indicate a lesion involving the facial nerve. Loud noises should evoke a blink, testing CN VIII. The swallowing reflex tests CN IX and X. Fasciculations of the sternocleidomastoid muscle or tongue may represent a lesion involving CN XI or XII, respectively [2]. Detailed examination of the cranial nerves is generally not necessary unless there is suspicion of a problem.

Sensation testing is generally limited to light touch and pinprick testing. During the entire newborn exam, the clinician can be assessing the response of the baby to touch. For example, infants commonly will react to the stethoscope touching their chest.

Motor examination involves assessing spontaneous movement of the head and extremities, as well as muscular tone. Neonatal hypotonia is the most common abnormal neurological sign seen in neonates [50]. It is associated with a wide array of disorders. Hypotonia can be divided into central and peripheral origins. Central hypotonia tends to be associated with other central nervous system (CNS) findings. Failure to fixate or meet other milestones, or seizures, are associated with central hypotonia [51]. Several common syndromes are associated with hypotonia include Down Syndrome and Prader-Willi syndrome. Other important considerations are hypoglycemia, hypothyroidism, hypoxic-ischemic encephalopathy, infection, and metabolic disorders. Often, central causes of hypotonia become more classic of CNS lesions resulting in increased tone, increased reflexes, and so forth. Transection of the spinal cord should also be considered, especially when there is a report of possible neck hyperextension in utero or during delivery. These infants may present severely with respiratory failure. Lower spinal level lesions may present with hypotonia that often becomes hypertonia with hyperreflexia characteristic of central CNS lesions [51].

Peripheral hypotonia, in contrast with central causes, often will present as hypotonia without any specific central signs. Eyes are able to fix well. Infants often are responsive to sounds and stimuli. Examples of this include spinal muscular atrophy, neonatal myasthenia gravis, and infantile botulism.

Infant spinal muscular atrophy, also known as Werdnig-Hoffman syndrome or SMA type 1, can present with hypotonia at birth (in 60% of
cases), weakness, poor suck, and absent reflexes. This is due to anterior horn cell degeneration. Werdnig-Hoffman typically presents between 0 and 6 months [52]. In contrast to cerebral damage hypotonia, these infants are typically alert and have facial reactions.

Neonatal myasthenia gravis occurs in 10% to 15% of infants born to mothers with myasthenia gravis. The acetylcholine receptor antibodies readily cross the placenta and can attach to the infant’s receptors producing a myasthenic phenotype. Signs of bulbar weakness including a weak suck and cry, hypotonia, absence of a moro reflex, and respiratory insufficiency can develop up to 72 hours after delivery. Treatment is with acetylcholinesterase inhibitors as in adults. Symptoms generally resolve by about 12 weeks of age as the maternal antibodies are cleared.

During delivery, particularly with difficult or breech deliveries, the brachial plexus may be injured causing a spectrum of resulting outcomes. The brachial plexus is formed from the C5-T1 nerve roots. Palsies involving this plexus are divided into three main types. Erb palsy involves the C5, C6, and occasionally C7. Klumpke palsy involves C8 and T1. The third type involves the complete plexus. Erb palsies present with the affected upper extremity in the classic “waiter’s tip” position. Klumpke palsies yield a paralyzed hand with full function of the elbow and shoulder. This palsy is rare, comprising only 0.5% of all brachial plexus injuries [2].

Total plexus injuries yield a complete paralysis of the upper extremity. Injuries may be transient in mild cases to permanent in more severe instances. The range of resolution relates to the actual injury to the nerve roots. Mild cases generally are the result of the brachial plexus nerve roots being stretched, termed neurapraxia. In these cases, prognosis is excellent. The most severe cases result when the nerve root is avulsed from the spinal cord, known as axonotmesis. These cases have no chance for spontaneous resolution and surgical correction often does not yield good outcomes [2]. Fortunately, 80% to 95% of cases (primarily Erb palsies) will resolve spontaneously over the course of a few weeks to months.

If, on physical examination, there is difficulty determining if the infant does not move the area because of pain (as a result of a clavicular fracture, for example) or a palsy, the Moro reflex can be used. In cases of palsies, the Moro reflex will result in asymmetric movement.

Skin

Newborn babies exhibit a plethora of skin findings. Often, these findings can be alarming to parents and result in numerous clinic and emergency department visits. Often, visual diagnosis is quite easy and can alleviate parental concerns.

Newborns often experience some degree of vasomotor instability that can present in several ways, including cutis marmorata, harlequin color changes, and acrocyanosis. Cutis marmorata is a condition resulting from uneven
distribution of capillary blood flow. It gives a mottled appearance to the skin and often can be induced by cold exposure.

Harlequin color changes are precipitated by turning the baby on the side. When this is done, the lower half of the baby becomes erythematous or dusky while the upper half becomes pale. Often there is a sharp demarcation between zones. This typically resolves within a few seconds after returning to the supine position but can persist for up to 20 minutes [53]. Harlequin color changes are more commonly seen in low-birth-weight infants and often resolve after the first few weeks of life.

Acrocyanosis results from venous blood pooling in the extremities giving a bluish color to the hands, feet, and occasionally around the lips. This condition is exaggerated with cool temperatures and resolves with warming. Acrocyanosis is not pathological and will resolve as vasomotor stability improves.

Erythema toxicum is extremely common, affecting up to 50% of full-term infants [53]. It typically presents in the first 2 to 3 days of life and typically resolves in 5 to 7 days. Lesions classically are described as having a central flesh-colored papule with surrounding erythema at the base. Numbers can range from a few lesions to over 100. With many lesions, the erythema can become confluent making the appearance less classic, but examination of the margins often reveals individual papules with the typical appearance. Biopsy is generally not needed but would show the presence of eosinophils. Its etiology is not well known at this point.

Less common is neonatal pustular melanosis, which affects approximately 5% of black infants and less than 1% of white infants [2]. The pustules are always present at birth or form within the first 24 hours of life. Following pustule formation, the pustules rupture leaving the classic collarettes of scaly skin and hyperpigmentation. It is possible for the pustules to rupture in utero leaving only the collarettes and hyperpigmentation at birth. Pustules may be present on the face, neck, hands, and feet (including the palms and soles). Atypical presentations should be evaluated very closely as some infectious processes, such as impetigo, can mimic pustular melanosis and require rapid treatment [2].

Forty percent of newborns will have milia, most commonly found on the nose [53]. They represent small cysts filled with keratinocytes and sebaceous debris. They may also be seen on other areas of the body. Generally, these papules are self-resolving over the course of a few weeks. Epstein’s pearls represent the same process in the oral cavity.

Neonatal acne is occasionally seen shortly after birth but typically develops between the second and fourth week of life. It peaks around 8 to 12 weeks of life, under the influence of maternal and fetal androgens, then subsequently resolves. Lesions are characteristically comedonal or pustular in appearance. Occasionally, nodulocystic acne can occur. In these cases, medical management may be indicated to prevent scarring. Persistent acne may be an indication of excess androgen production. If there are concerns
about this, growth should be followed closely and a bone age can be checked for accelerated bone growth. Normal bone growth makes excess androgen production unlikely [2].

Nevus simplex is a blanching macule that is typically pink or red in color. When located on the glabella they are commonly referred to as angel kisses, and when located on the posterior neck they are often termed stork bites. While these two sites are the most common, they may be found anywhere along the midline from the eyes to the nape of the neck. Often they fade with time, although they may not completely.

Nevus simplex should be distinguished from a nevus flammeus or “port-wine stain.” Port-wine stains do not tend to resolve much spontaneously. Laser treatment can help reduce their appearance. They are important to recognize, as they are associated with some syndromes. In particular, a nevus flammeus that involves the ophthalmic and maxillary regions of the trigeminal nerve (unilaterally or bilaterally) may be associated with Sturge-Weber syndrome that can have CNS involvement and lead to seizures. Port-wine stains that involve the face and/or upper extremity with hypertrophy of the affected side may have a condition termed Klippel-Trenaunay [2].

Dermal melanoses, formerly referred to as Mongolian spots, are bluish-black areas of hyperpigmentation often found on the buttocks, back, and shoulders. They are extremely common in darker skinned babies (upwards of 90% of Native American, Asian, and African American babies will have them) [53]. These spots can be confused with bruising and have resulted in evaluation for abuse in some cases. For this reason it is important to recognize them. Typically they will fade with age but may persist into adulthood.

Another commonly seen hyperpigmented macule is the café au lait spot. Typically these macules are light brown in color. A few café au lait macules are normal; however, the presence of many macules or a very large macule may indicate the presence of a systemic disease such as neurofibromatosis type 1 or McCune-Albright syndrome, respectively. The following list summarizes the National Institutes of Health criteria for the diagnosis of neurofibromatosis type 1 [54]. Any two or more clinical features are required for diagnosis.

- Six or more café-au-lait macules over 5 mm in greatest diameter in prepubertal individuals and over 15 mm in greatest diameter in postpubertal individuals
- Two or more neurofibromas of any type or one plexiform neurofibroma
- Freckling in the axillary or inguinal regions
- Optic glioma
- Two or more Lisch nodules (iris hamartomas)
- A distinctive osseous lesion such as sphenoid dysplasia or thinning of the long bone cortex with or without pseudarthrosis
- A first-degree relative (parent, sibling, or offspring) with neurofibromatosis type 1 by the above criteria
Summary

Despite the broad technologic advancements of medicine, screening for illness in infants is highly reliant on a complete physical examination. For this reason it is critical that the examining physician not only have a thorough understanding of abnormal findings but also the normal findings and their variants. The vast majority of infants are healthy and findings predictive of future health problems are subtle and infrequent. Yet, outcomes can be devastating. Therefore it is critical for the physician to remain diligent during screening. It is our hope that this article will assist the physician in this task and allow for more accurate and timely diagnosis that prevents or minimizes long-term health problems in children.

References