Physical examination of Newborn

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Swelling of the eyelids is common in newborns and resolves during the first few days of life.

Subconjunctival hemorrhages are common. They occur during delivery and resolve in 1 to 2 weeks.
The normally white **sclera** is evaluated for changes of color.

A bluish coloration, however, is present in premature infants and in other small babies because of their very thin sclera. Also presents in baby with osteogenesis imperfecta.
There may also be a mucoid discharge affecting the eyes, often called a “sticky eye,” in the first few days of life, which resolves spontaneously; if more prolonged, it is often from a blocked or incompletely canalized nasolacrimal duct.

The eyelids can be cleansed with sterile water.

This condition must be contrasted with the erythematous, swollen eyelids with purulent eye discharge seen in conjunctivitis.
Disconjugate Eye Movements

During the first few months of life, newborns will frequently have disconjugate eye movements, where the eyes appear to move independently. Eyes may transiently appear crossed or divergent. This phenomena is particularly noticeable when the infant is falling asleep or being woken from sleep. If the disconjugate movement is fixed (one eye is always out, or always in, relative to the other), a pediatric ophthalmologist should be consulted. But if the movements are transient, it is a normal finding and will spontaneously resolve.
ABNORMAL RED REFLEX

This is one of the most important abnormalities that requires immediate evaluation.
The term *leukocoria* is used to describe a white pupil seen by the naked eye or during the red reflex test.

Leukocoria is not a diagnosis but rather a description of an observation.

Because the infant sleeps much of the time and because the pupils are small, a white pupil often is not noticed until the infant becomes more alert and active.
False-positive red reflex test results are commonly due to small pupils, shifting gaze, limited patient cooperation, poor illumination from the ophthalmoscope, and examiner inexperience.

Regardless, these patients should be over-referred to the ophthalmologist to ascertain the true-positive results.

Causes of leukocoria in infants include opacities of the cornea, lens, and vitreous, as well as retinal diseases such as retinoblastoma, chorioretinal coloboma, persistent hyperplastic primary vitreous, endophthalmitis, Coats disease, congenital retinal fold, retinoschisis, or scarring from ROP.

Abnormal red reflexes can also result from misaligned eye or from high or asymmetric refractive errors.
Heterochromia (dissimilarity in pigmentation between the two eyes or within one eye) can indicate a normal hereditary pattern, congenital Horner syndrome, or one of several syndromes (e.g., Waardenburg). These syndromes might not become apparent until the end of the neonatal period or even later in life when the iris is fully pigmented.
Congenital glaucoma
These infants show marked proptosis and enlargement of the eye, 'buphthalmos', in association with congenital glaucoma.

The intraocular pressure is raised and blindness may result from effects on the optic nerve unless the pressure can be controlled. Congenital glaucoma is associated with numerous conditions, for example, Sturge-Weber syndrome and intra-ocular tumours.
Hypotelorism occurs when the eyes are unusually close together; hypertelorism occurs when the eyes are too far apart.

Clinically, hypotelorism and hypertelorism are defined by the interpupillary distance, which may be estimated in a relaxed patient by measuring between the midpoints of the pupils.
It is usually impossible to measure the interpupillary distance of a newborn; therefore, two other relevant and useful measurements that are easier to obtain are the inner canthal distance and the outer canthal distance.

Telecanthus is an increase in the inner canthal distance, and it may occur in the absence of hypertelorism, such as in Waardenburg syndrome type I.
There are other factors that may create an illusion of hypertelorism, such as epicanthal folds and a flat nasal bridge; therefore, a subjective impression should always be confirmed by measurement of all three distances, if possible.

Epicanthal folds are a feature of normal fetal development, and they may be present in normal infants. They are characteristic in trisomy 21, but they occur in many other malformation syndromes, especially in those that include a flat nasal bridge.
From the prognostic and diagnostic points of view, it is important to identify hypotelorism because often it is associated with holoprosencephaly, a major malformation of the central nervous system that usually is associated with severe disturbance of brain function and early death.

Holoprosencephaly can be isolated or can be part of trisomy 13
Normally, an imaginary line through the inner and outer canthi should be perpendicular to the sagittal plane of the face.

An **upward slant** to the palpebral fissure is seen in **trisomy 21**

![Up-slanting palpebral fissures](image)

and a **downward slant** is seen in **mandibulofacial dysostosis** and **Noonan syndrome**.

Both types of slant can be part of a number of other syndromes.
A coloboma is a developmental defect in the normal continuity of a structure, and it often is used in reference to the eye.

Colobomas may involve the *eyelid margin*, as those seen in Treacher Collins syndrome or the *iris and retina*, as those seen in CHARGE syndrome.

Identification of a coloboma should lead to a formal ophthalmologic evaluation.
Synophrys, or fusion of the eyebrows in the midline, is common in hirsute infants, and it may also be familial in some instances.

The Cornelia de Lange syndrome is strongly associated with synophrys.
Normal Ear

Many variations in size and shape exist within the label of "normal ear", but in general, the normal ear is one in which all the structures (helix, antihelix, tragus, antitragus, scaphoid/triangular fossa, and external auditory canal) are all present and well formed.
Ear Tag

A single, small ear tag is an occasional finding on physical examination. It is often inherited as a familial trait.
Ear pits (preauricular pits) are often a rather subtle finding on physical exam. They are located at the superior attachment of the pinna to the face and may be unilateral or bilateral. Up to 10% of Asian infants will have pits — they are less common among caucasians and African Americans. There is a rare association between ear pits and Brachio-Oto-Renal Syndrome so audiologic testing of these infants is recommended, but otherwise this is considered a benign finding. With an isolated pit, renal ultrasounds are not generally recommended. Screening for associated renal anomalies becomes more important when other anomalies are present, when there is a positive family history of deafness, renal pathology or auricular malformations, or when the mother has a history of gestational diabetes.
Malformations of the ear may be associated with hearing loss. Skin tags anterior to the ear (preauricular tags) and accessory auricles should be removed by a plastic surgeon. Preauricular tags are usually isolated and benign, but infants warrant their hearing checked with the newborn hearing screen. They are sometimes associated with other dysmorphic features, a family history of deafness, maternal history of gestational diabetes, and increased risk for renal anomalies.
When a preauricular tag is associated with other abnormalities or risk factors, a renal ultrasound is recommended.

The need for a renal ultrasound examination for an isolated preauricular tag has been questioned; a meta-analysis showed similar risk of renal tract anomalies to that of the general population.
Stahl's Ear

Also known as Satyr ear, Spock ear, or Vulcan ear, this deformity of the pinna is characterized by a flat helix at the superior pole, a third crus extending into the helix, and a flattened scaphoid fossa. When this deformity is severe enough to be of cosmetic concern, molding or splinting can be initiated by a plastic surgeon (within the first week of life for the best cosmetic results). As an isolated finding, cosmesis is the only concern, but the appearance will not improve with time. Exact etiology is unknown.
Lop Ear

This pinna deformity, where the superior edge of the helix is folded down, is known as lop ear. Again, it is typically an isolated finding. This is another deformity that may be improved with splinting.
Cup Ear

This is the same ear as in the previous photo, viewed from the front. From this angle, the prominence of the auricle can be better seen. As an isolated finding, this ear shape does not require any further evaluation.
Microtia

In contrast to the previous photos, this pinna is not well-formed and is smaller than a normal ear. This is microtia, class II. Hearing evaluation is mandatory in these infants and referral to a pediatric ENT specialist is recommended. Fortunately, this infant was found to have normal hearing. Elective cosmetic repair was planned for a later date.
Hypoplastic Ear

Here is another infant with auricle malformation. The ear is hypoplastic and low set. Again, because genetic syndromes are frequently associated with abnormal ear shapes, a careful physical examination should be performed. Even in the absence of other findings, referral to a pediatric ENT specialist may be indicated, as an abnormal pinna may be an indicator of internal ear abnormalities.
Low-set ears are positioned so that the top of the pinna falls below a line drawn from the outer canthus of the eye at right angles to the face. Low-set or abnormal ears are characteristic of a number of syndromes.
NOSE

The nose, like the external ear, shows great individual variation, but certain alterations in shape are frequent in malformation syndromes involving the face.

The nose may be unusually thin with hypoplastic alae nasi, as in Hallermann-Streiff syndrome,

or it may be unusually broad, as in frontonasal dysplasia.
A depressed nasal bridge with an upturned nose occurs in many skeletal dysplasias, such as achondroplasia.

When the depression is severe, the nostrils may appear to be anteverted, and the nose may appear to be shortened.

A hypoplastic nose is often syndromic.
and a nose with a single nostril is highly suggestive of holoprosencephaly
Positional Nasal Deformity

Viewed from below, one can more clearly appreciate the asymmetry of the nares and the angulation of the septum.
Positional Nasal Deformity

With compression of the nasal tip, the septum becomes even more angled, raising concern for septal dislocation, but the base is still centrally located. In this case, there was enough doubt about the possibility of dislocation that ENT was consulted, but with more complete evaluation, dislocation was ruled out. By the following morning, the appearance of the nose was significantly improved and the compression test was less ambiguous -- the septum remained straight and midline with pressure.
Dislocated Nasal Septum

From this view, the deformity is more easily seen. The septum is not straight and the whole bottom segment of the nose appears to be shifted to the baby’s left (look towards the bridge of the nose to appreciate this). Compressing the tip of the nose gently can also help distinguish this condition from a positional problem, as the next photo demonstrates.
Dislocated Nasa Septum

When the tip of the nose is compressed gently in the presence of a dislocated septum, the deformity appears to worsen. In this photo, the tip of the nose is actually beside the base of the septum. With positional deformity, the septum would stay straight, even if the nares appear uneven. Although some authors estimate the incidence of dislocated septum to be as high as 4%, in our experience this condition is quite rare. An experienced pediatric otolaryngologist can relocate the septum fairly easily, without surgery, if consulted early (within the first few days of life), so recognition of this condition and timely referral is important to prevent permanent deformity.
Figure 28–8. Result of finger compression (A) when nasal septum is dislocated. Normal septal relationship (B) results in no nasal deviation with pressure. (Modified from Daily W, et al: Nasal septal dislocation in the newborn, Mo Med 74:381, 1977.)
MOUTH AND PALATE

The mouth is observed for size, position, and asymmetry.

The palate must be inspected, including posteriorly, to exclude a posterior cleft palate. It should also be palpated to detect an indentation of the posterior palate from a submucous cleft or a posterior cleft palate.

Micrognathia describes a small mandible and may be associated with glossoptosis and a posterior cleft palate (Pierre Robin sequence) and may cause upper airway obstruction and feeding difficulties.
Here is a third example of natal teeth. When palpated, these teeth could be moved slightly, but they were placed low enough in the gum that they do not currently present an aspiration risk. They were not removed.
Natal teeth are observed in approximately 1 in 2,000 newborn infants usually in the position of the mandibular central incisors.

Natal teeth are present at birth, whereas neonatal teeth erupt in the 1st mo of life. Attachment of natal and neonatal teeth is generally limited to the gingival margin, with little root formation or bony support.

They may be a supernumerary or a prematurely erupted primary tooth. A radiograph can easily differentiate between the two conditions.

Natal teeth are associated with cleft palate, Pierre Robin syndrome, Ellis-van Creveld syndrome, Hallermann-Streiff syndrome, pachyonychia congenita, and other anomalies. A family history of natal teeth or premature eruption is present in 15-20% of affected children.
Natal or neonatal teeth occasionally result in pain and refusal to feed and can produce maternal discomfort because of abrasion or biting of the nipple during nursing.

If the tooth is mobile there is a danger of detachment, with aspiration of the tooth.

Because the tongue lies between the alveolar processes during birth, it can become lacerated (Riga-Fede disease).

Decisions regarding extraction of prematurely erupted primary teeth must be made on an individual basis.

Exfoliation failure occurs when a primary tooth is not shed before the eruption of its permanent successor. Most often the primary tooth exfoliates eventually, but in some cases, the primary tooth needs to be extracted. This occurs most commonly in the mandibular incisor region.
Tongue-tie occurs in approximately 4% of newborns. Many babies with this condition can breastfeed without difficulty, but in some cases, a tight frenulum makes latching on difficult. In those cases, frenotomy may be indicated. The Hazeltaker Assessment Tool for Lingual Frenulum Function is one tool that may be used to grade the severity of the tongue-tie objectively. (Video clip of assessment being done). There are no prospective trials on the outcome of speech in those infants identified at birth, so this is not currently an evidence based reason to clip the frenulum in the nursery. The impact on breastfeeding, however, is well documented.
Ankyloglossia

This is the same infant as in the previous photo, immediately after frenotomy. There is no bleeding, and the tongue is more mobile. Latching was significantly improved.
These vesicular-looking lesions were first noted by the mother as she was getting the baby latched on for feeding. These are minor salivary glands that are just more prominent than usual. No evaluation is needed.
The mouth is a complex structure with component parts that each require separate evaluation.

The size and shape of the mouth may be altered. A small mouth, or microstomia, should be noted; it occurs in trisomy 18.

Macrostomia, a large mouth, should be noted as well; it may be present in such conditions as mandibulofacial dysostosis.
The corners of the mouth may be downturned, as in Prader-Willi syndrome and other conditions with hypotonia.

An asymmetric face during crying occurs with congenital deficiency in the depressor anguli oris muscle on one side, and this may be associated with other abnormalities, such as hemifacial microsomia and velocardiofacial/DiGeorge syndrome.
Prominent, full lips occur in various syndromes, including Williams syndrome.

A thin upper lip may be seen in Cornelia de Lange syndrome and in fetal alcohol syndrome.
A cleft upper lip is usually *lateral*, as in the common multifactorial cleft lip (or palate) anomaly, occurring in the position of one of the philtral ridges.

The presence of *pits in the lower lip* associated with a cleft lip or palate, however, is suggestive of *Van der Woude syndrome*, which is inherited in an autosomal dominant manner.
A median cleft lip is very suggestive of holoprosencephaly.

In fact, there are many diverse syndromes with cleft lip or palate that are important to identify because they may have other associated malformations and relatively high genetic risks for recurrence.

Therefore, it is particularly important to evaluate the infant with cleft lip or palate carefully for evidence of other malformations to give accurate recurrence risk and prognostic information to the family.
Isolated cleft palate is different genetically from cleft lip. Mild forms of cleft palate are represented by submucosal clefts, pharyngeal incompetence with nasal speech (velopharyngeal insufficiency), and bifid uvula.

A high arched palate may occur normally, but it is also a feature of many syndromes, especially if hypotonia or another long-standing neurologic abnormality is present.
Hypertrophied alveolar ridges are apparent in the palate along the inner margin of the teeth, and they are suggestive of Smith-Lemli-Opitz syndrome if seen in an infant.

Macroglossia may be relative, as in the Pierre Robin malformation sequence, in which the primary abnormality is mandibular hypoplasia.
In other cases, such as hypothyroidism, Beckwith-Wiedemann syndrome, and Down syndrome, the tongue protrudes and is enlarged.

A cleft or irregular tongue or oral frenula occurs in various syndromes such as the orofaciiodigital syndromes.
An excessively small mandible is termed micrognathia, which is a feature of many syndromes. It is a characteristic of the Pierre Robin sequence, which consists of the triad of micrognathia, glossoptosis, and a U-shaped cleft palate, as opposed to the common V-shaped cleft. The Pierre Robin sequence may be part of a syndrome, such as Stickler syndrome (hereditary arthro-ophthalmopathy), and thus other anomalies and a family history must be sought.
In other syndromes (like Treacher Collins syndrome), the maxilla likewise may be hypoplastic, decreasing the prominence of the upper cheeks (malar hypoplasia).
NECK

The neck may be short, and limitation of rotation should raise the suspicion of fusion of cervical vertebrae, as in a Klippel-Feil anomaly.

Excessive skinfolds are characteristic of Turner syndrome, Noonan syndrome, and Down syndrome.

In these examples, the excess nuchal skin often represents resolution of a cystic hygroma that was present prenatally.
A webbed neck is a feature of Turner syndrome, which may also be associated with lymphedema of the feet.
Cystic hygromas are soft, fluctuant swellings usually in the posterior triangle that transilluminate.