Physical examination of Newborn

By

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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.
The thoracic cage may be unusually small as part of a skeletal dysplasia, such as thanatophoric dysplasia or Jeune asphyxiating thoracic dystrophy.

The sternum itself may be unusually short, which is typical in trisomy 18.
it may be altered in shape, as is seen in **pectus excavatum**

pectus excavatum is most commonly an isolated congenital abnormality that results in no functional impairment.

When the infant is at rest, a small depression can be seen over the sternal area. The next photo shows how the appearance changes with inspiration while crying.
With deep inspiration, the sternum appears to almost collapse into the chest cavity. While connective tissue disorders, such as Marfan's, may be associated with this finding, pectus excavatum is more commonly a benign, isolated entity.
pectus carinatum are commonly seen in a variety of skeletal dysplasias and connective tissue disorders
The brown macule below the left nipple in this infant is a supernumerary nipple. Found along the "milk lines" they may be single or multiple, unilateral or bilateral. It is an insignificant finding.
The nipple area is a common location for small skin tags.

They are typically quite tiny and do not need to be removed.
This visible, firm lump in the midline of the chest is a frequently observed finding in newborns. It is simply a prominence of the xiphoid process and does not represent an abnormality. With time, this becomes less noticeable.
This newborn had an uncomplicated vaginal delivery, but on the initial examination, crepitus was appreciated over the left clavicle. Although a classic finding for clavicle fracture, crepitus is not always present.

The examiner should have a high index of suspicion for fracture whenever the clavicle cannot be palpated easily and distinctly along the entire length of the bone. No splinting or medications are recommended; babies are typically asymptomatic. Anticipatory guidance regarding the firm lump that often develops in this area over the next few weeks (as new bone development occurs) should be given.

This photo, taken 40 hours after delivery, already shows some asymmetry between the two sides.
One of the most important physical findings to be able to recognize in a newborn is the presence of retractions.

Sepsis, pulmonary pathology, cardiac disease, metabolic disorders, polycythemia, cold stress, and others can all cause retractions. It is a sign of a newborn in distress.

In this photo, taken during inspiration, the shadows between the ribs can be clearly seen. Retractions may or may not occur in combination with other signs of distress: nasal flaring, grunting, and tachypnea.
This infant demonstrates subcostal retractions. Here the shadow is seen at the lower margin of the rib cage only.
Before auscultating the chest, notice the shape and symmetry of the chest wall.

In this infant, the antero-posterior (AP) diameter appears greater than normal, and there was concern that the AP diameter of the left chest was greater than that on the right. (Notice the white lead on the right nipple and the gold lead over the midline)

This finding is suspicious for pneumothorax, which can occur spontaneously in well newborns. Congenital diaphragmatic hernia may present with an abdomen that appears flat relative to the chest (scaphoid abdomen), but in that case severe respiratory distress would be expected.
Breast enlargement can occur in newborns of either sex.

A small amount of milk ("witch’s milk") may be discharged
HEART
The normal heart rate is 110 to 150 beats per minute in term infants but can drop to 85 beats per minute during sleep.

The heart sounds should be loudest on the left side of the chest.

Heart murmurs can be heard in about 0.6% of infants at the routine examination of the newborn. Most are innocent and originate from the acute angle at the pulmonary artery bifurcation or are from a patent ductus arteriosus or tricuspid regurgitation.

The problem is to differentiate innocent murmurs from those caused by significant heart lesions. Clinical features may serve as a guide.
BOX 29-3  FEATURES OF A HEART MURMUR IN A NEONATE

FEATURES OF AN INNOCENT MURMUR

- Soft (grade 1/6 or 2/6) murmur at left sternal edge
- No audible clicks
- Normal pulses
- Otherwise normal clinical examination

FEATURES SUGGESTING A MURMUR IS SIGNIFICANT

- Pansystolic
- Loud (≥ grade 3/6)
- Harsh quality
- Best heard in the upper left sternal edge
- Abnormal second heart sound
- Femoral pulses difficult to feel
- Other abnormality on clinical examination
The usefulness of electrocardiograms and chest radiographs in helping to distinguish innocent from significant murmurs is controversial.

The neonatal electrocardiogram and chest radiograph are difficult to interpret, and these tests have rarely been found to change decisions based on the clinical examination. Many centers have stopped performing them under these circumstances.

If a heart murmur is thought to be significant or cannot confidently be diagnosed as innocent, the infant should be referred directly for echocardiography.
The management of infants with an innocent murmur depends on the availability of echocardiography. If echocardiography is readily available, it can be performed directly and provides parents with a definitive diagnosis without delay.

If echocardiography is not readily available, a follow-up medical examination should be arranged soon after discharge and the parents warned to seek medical assistance if their infant becomes symptomatic with poor feeding, labored breathing, or cyanosis.

Most innocent murmurs disappear in the first year of life, most in the first 3 months. However, any mention of a heart murmur can create considerable parental anxiety, which sometimes continues for years. Attention must be paid to this to prevent parents from continuing to worry about their child’s heart even after the murmur has disappeared.
ABDOMEN

Observe for abdominal distention, asymmetry, and signs of umbilical inflammation.

For abdominal palpation, the infant must be relaxed.

The abdomen is palpated to identify any masses.

The liver is normally palpable 1 to 2 cm below the costal margin.

If palpation of the abdomen detects abnormally large renal masses or an enlarged bladder in a male infant, ultrasonography is required urgently to identify urinary outflow obstruction.
Most cases of urinary outflow obstruction are now detected on prenatal ultrasound screening, as are other major abnormalities of the kidneys and urinary tract.

Siblings of children with vesicoureteric reflux should be screened for this condition because up to 40% are also affected.

The American Urological Association has published clinical practice guidelines for screening siblings
Hypoplasia of the abdominal musculature may occur in association with intrauterine bladder outlet obstruction and other anomalies of the urogenital system. It results in a characteristic 

**prune-belly appearance** in Eagle Barrett syndrome.

An **omphalocele**, in which abdominal contents protrude through the umbilical opening, may be part of the Beckwith-Wiedemann syndrome or chromosomal abnormalities such as trisomy 13.
Gastroschisis, however, is usually an isolated disruption in which the abdominal contents protrude through the periumbilical abdominal wall.

Anomalies of a more minor nature, such as inguinal or umbilical hernias, occur in normal infants, but they are more frequent in various syndromes, particularly in connective tissue disorders.
Diastasis Recti
A vertical bulge down the midline of the abdomen can be seen in many newborns when intra-abdominal pressure increases.

In this photo, a shadow lateral to the bulge can be seen going up the left side of the abdomen, starting near the umbilical clamp. Diastasis recti is caused by a relative weakness of the fascia between the two rectus abdominus muscles. It is not a herniation and is not pathologic. With time, this will disappear.
The dark line down the midline in this baby is a linea nigra. It is a fairly common finding in newborns, especially those with more pigmented skin.

The presence of linea nigra seems to be impacted by hormonal levels, as this finding is often seen in pregnant women as well. It is completely benign.
Single umbilical artery occurs in about 0.3% of newborns. It is associated with an increased risk for chromosomal abnormalities and congenital malformations, particularly of the genitourinary system.

A single umbilical artery was associated with asymptomatic renal anomalies in 7% in one series. The yield is low from ultrasound screening of the kidneys and urinary tract when this is an isolated finding and most renal abnormalities identified are transient or mild. The yield is further reduced by routine prenatal ultrasound screening for congenital anomalies of the kidneys and urinary tract. It is probably best reserved for those who also have other anomalies on physical examination.
Excess Wharton's jelly
Cord hematoma
Excess skin
omphalocele
Umbilical granolomum
FEMORAL PULSES
Femoral pulses are palpated when the infant is quiet.

Pulse pressure is reduced if there is coarctation of the aorta.

If coarctation is suggested clinically, further evidence can be obtained by comparing the blood pressure in the arms and legs. A difference of more than 20 mm Hg is significant, and immediate treatment is indicated.

Pulse pressure is increased with a patent ductus arteriosus.
Among the most common congenital anomalies are the neural tube defects, which involve abnormalities of the central nervous system along with defects in the associated bony structures.

Minor external anomalies, particularly of the lower spine, include unusual pigmentary lesions, hair tufts, dimples, and sinuses.
Some of these changes, such as hair tufts and sinuses above the gluteal cleft, may be an indication of a more significant deeper anomaly and require further evaluation, such as magnetic resonance imaging.
Many newborns, especially those with increased skin pigmentation, will have an increased amount of hair over the lower back and sacrum. This appearance is entirely within normal limits and is not a marker for underlying spinal dysraphism.
Sacral dimples are a commonly encountered finding on the physical exam. Most of these dimples are "simple dimples" and require no further evaluation. A simple dimple is one that is located within 2.5 cm of the anus, has a base that can be visualized and is not associated with other abnormalities on exam. The dimple in this photo is the tiny dark brown shadow below the slate grey patch at the superior end of the gluteal crease.
When the skin lateral to the dimple is stretched, skin can be seen covering the entire dimpled area. If the base could not be seen, this would be called a coccygeal pit. Although fistulas above the gluteal cleft may be associated with spinal dysraphism, coccygeal pits are benign and do not need imaging.
Skin tags in the sacral area are also potential indicators of spinal dysraphism. In some cases, the "tag" may in actuality be a residual tail. This skin tag was quite small and appeared to be very superficial, but spinal ultrasound was still done as a screening measure. In this case, the ultrasound was normal and no further testing was needed.
Congenital midline vascular lesions usually raise questions about occult spinal dysraphism, but so far the literature is inconclusive.

Some authors suggest isolated midline port wine stains and medial telangiectatic vascular nevi ("butterfly marks") are benign and do not require further evaluation. Occult dysraphism is much more likely if two markers are present, so this appearance in combination with another finding should lead to imaging. As an isolated finding, the need for evaluation is debatable.
GENITALIA

Hypogenitalism can be seen in association with hypotonia in Prader-Willi syndrome or with low-set dysplastic ears, syndactyly of the toes, and thickened alveolar ridges in Smith-Lemli-Opitz syndrome.

Genital ambiguity is associated with renal anomalies and an increased risk for Wilms tumor in Denys-Drash syndrome.
Virilization may be associated with 21-hydroxylase deficiency, which requires urgent management of adrenal insufficiency.
GENITALIA

In boys, the penis is checked for length and the position of the urethral orifice.
The presence of testes in the scrotum is confirmed.

The penis may appear to be short but may be buried in suprapubic fat.
A penis less than 1 cm long is a micropenis suggesting congenital hypopituitarism.
In hypospadias, the urethral meatus is in an abnormal position, usually on or adjacent to the glans penis, but may be on the penile shaft or perineum.
The testis may feel enlarged on palpation; this is usually from a hydrocele, which can be confirmed on transillumination with a flashlight. Hydroceles are relatively common in boys and usually resolve spontaneously.

An undescended testis has failed to descend from its embryologic position from the urogenital ridge on the posterior abdominal wall through the inguinal canal to the scrotum.
Undescended testes may lie along the line of descent but may not have reached the scrotum or may be ectopic testes, which have deviated from the line of descent.

At birth, about 4% of full-term male infants have an undescended testis; by 3 months, about 1% are still undescended, with little reduction thereafter. An undescended testis should be reviewed at about 6 weeks of age. If still undescended, referral to a pediatric surgeon or urologist is indicated.

If both testes are undescended, the infant should be reviewed by a senior clinician. It needs to be considered if the infant is a virilized female with congenital adrenal hyperplasia.
Neonatal testicular torsion usually occurs before birth. The testis is usually hard, black, and nontender and is rarely salvageable because it has usually already undergone infarction.

A pediatric surgeon should be consulted urgently to decide if the testis can be salvaged. Many pediatric urologists will perform surgical fixation of the contralateral testis.
In girls, the clitoris and labia minora are prominent if the infant is preterm but are covered by the labia majora at full term.

There may be a white vaginal discharge or small amount of bleeding from maternal hormone withdrawal.

There may also be prolapse of a ring of vaginal mucosa. These lesions resolve spontaneously.

Passage of urine and meconium should be monitored and recorded and occur within 24 hours of birth in most term newborns.
ANUS

Imperforate or displaced anus may be isolated or may occur as the mildest expression of a caudal regression sequence, in which other anomalies such as sacral dysgenesis are seen.

It is most commonly part of a constellation of anomalies, such as the VATER or VACTERL association.

It also can be seen in a number of chromosomal abnormalities or as part of diabetic embryopathy.
LIMBS AND HANDS AND FEET

The limbs and hands and feet are examined for general appearance, symmetry and posture.

Observe for spontaneous movement of all four limbs.

Examination of the hands and feet will identify extra digits (polydactyly), fused digits (syndactyly), or shortened digits (clinodactyly).

Extra digits are usually connected by a thin skin tag, but can be completely attached and contain bone. Extra digits should be removed by a plastic surgeon; many pediatricians tie off thin skin tags with a silk thread, but this may leave a stump of skin.
Most newborns have two major creases on the palm, neither of which completely extend from one side of the palm to the other.
common variant, found in approximately 5% of newborns, a transverse palmar crease is frequently inherited as a familial trait.

Although single palmar creases are also associated with Down's syndrome and other genetic disorders, the absence of other abnormalities on physical exam should reassure the examiner that no further evaluation is necessary. About 45% of infants with trisomy 21 have single palmar creases.

Unilateral single palmar transverse creases are also observed in about 4% of normal infants, but bilateral single palmar transverse creases occur in less than 1% of normal Caucasian infants.
Polydactyly may be familial, but it can also be caused by a dysmorphic syndrome.

Post-axial polydactyly is the most common variety. The extra digit may seem almost fully formed or may be attached only by a thin fleshy stalk. This is typically an isolated finding. A positive family history is often obtained.
In cases where the attachment of the extra digit is very thin, the appendage can be successfully tied off with a silk suture.

After allowing some time for hemostasis to occur under the ligature, we usually snip off the appendage with a sterile scissors.

The suture will either fall off spontaneously, or it may be removed in a few days.
This infant has polydactyly of the thumb as an isolated physical finding.

There was no family history and the finding was unilateral.

Preaxial polydactyly is much less common (and more likely to be related to an underlying medical condition) than the postaxial variety.

Surgical correction is generally delayed for several months to lessen the risk of general anesthesia.
Polydactyly refers to partial or complete supernumerary digits and is one of the most common limb malformations.

Postaxial polydactyly is more frequent than preaxial, particularly in African Americans.

As an isolated anomaly, polydactyly may be inherited as an autosomal dominant trait. It also may be a manifestation of a multiple malformation syndrome. Postaxial polydactyly may occur in a variety of syndromes, including trisomy 13, chondroectodermal dysplasia, Meckel-Gruber syndrome, and Bardet-Biedl syndrome.
Another example of postaxial polydactyly. In this case the polydactyly is bilateral and the extra digits are fully formed.
Preaxial polydactyly is characteristic of Carpenter syndrome and Majewski short rib—polydactyly syndrome.
Syndactyly refers to fusion of digits; it is usually cutaneous, but it may involve bone. Minimal syndactyly of the second and third toes is common in normal newborns.

More extensive syndactyly can be seen in trisomy 21 and Smith-Lemli-Opitz syndrome. As an isolated anomaly, different clinical types may be distinguished, but each of them is inherited as an autosomal dominant trait with variable expressivity and incomplete penetrance. Extensive syndactyly often is part of a syndrome, and typical examples include some of the craniosynostosis conditions, such as Apert and Pfeiffer syndromes.
Brachydactyly refers to shortening of one or more digits owing to anomalous development of any of the phalanges, metacarpals, or metatarsals.

Various clinical types may be distinguished, but most isolated forms of brachydactyly are inherited in an autosomal dominant fashion.

Brachydactyly is also a component of numerous disorders, including skeletal dysplasias such as achondroplasia and syndromes such as Albright hereditary osteodystrophy and Down syndrome.
**Arachnodactyly** refers to unusually long, spider-like digits, and it is characteristic but not invariable in **Marfan syndrome** and homocystinuria.

![Hand Image]

The appearance of brachydactyly and arachnodactyly can be confirmed by measuring and determining the ratio of middle finger to total hand length, which is normally about 0.43 in the newborn.
Extremities may be relatively long, as occur in Marfan syndrome or homocystinuria or unusually short, as occur in a diverse group of skeletal dysplasias, the most common being achondroplasia.
In normal newborns, the ratio of the upper segment to the lower segment is about 1.7, and the ratio decreases with age to about 1.0 in the adult.

A high ratio suggests relative shortening of the extremities, and a low ratio implies either unusually long extremities or a foreshortened trunk, as may occur in spondyloepiphyseal dysplasia.
Hypertrophy of limbs may be a manifestation of Beckwith-Wiedemann syndrome or Klippel-Trenaunay-Weber syndrome.

Isolated hemiatrophy may occur with long-standing corticospinal tract damage as well as in Russell-Silver syndrome.

It is important to identify hemihypertrophy because individuals with this finding are at increased risk for intra-abdominal tumors, such as Wilms tumor, and thus require close monitoring throughout infancy and childhood.
In foot increased separation of the first and second toes and a prominent interdigital furrow in trisomy 21.
The hands and feet may be enlarged as a result of lymphedema. This is characteristic of infants with Turner or Noonan syndrome, in which the dorsum of the hands and feet may have a puffy appearance.

Congenital lymphedema can also be an autosomal dominantly inherited condition with variable expressivity.
Rocker-bottom feet is a term used to describe a prominent heel and a loss of the normal concave longitudinal arch of the sole. This feature is common in trisomy 18 and other syndromes.
Rhizomelia denotes proximal shortening of the limbs, such as those in achondroplasia.

Mesomelia refers to shortening of the middle segment, and acromelia refers to relative shortening of the hands or feet.
Congenital absence of an entire hand is termed **acheiria**

absence of both hands and feet is **acheiropodia**

**Ectrodactyly** refers to a partial or total absence of the distal segments of a hand or foot, with the proximal segments of the limbs more or less normal.
All such anomalies are examples of terminal transverse defects and may occur sporadically or as part of a syndrome.

The term ectrodactyly is frequently misused for the lobster-claw anomaly, which is best described as split hand/split foot. In this anomaly, the central rays are deficient, and there is often fusion of the remaining digits. Split hand/split foot may be seen in isolation, when it is of autosomal dominant origin, or it may be seen with other anomalies.
Absent radius or ulna

It is useful to determine whether the defects involve primarily the radial, or preaxial, side of the limb or the ulnar, or postaxial, side.

For example, blood dyscrasias such as Fanconi anemia and the thrombocytopenia-absent radius syndrome commonly involve radial deficiency.
A slight *medial incurvation of the fifth finger* is a frequent finding on the newborn exam. It is often inherited as a familial trait. Although it can be associated with several genetic disorders, including Down's syndrome, the absence of other physical findings will confirm that it is a benign, isolated finding. In this case, no further evaluation is necessary.
A variety of congenital joint deformities involving the limbs may occur.

**Arthrogryposis**, multiple congenital contractures, is most often sporadic and may be associated with oligohydramnios or may be the result of some underlying neuromuscular abnormality, such as spinal muscular atrophy.
Talipes equinovarus or calcaneovalgus deformities of the ankle are common isolated joint contractures.

Joint hypermobility is frequent in various connective tissue disorders, such as Marfan and Ehlers-Danlos syndromes, and can also be seen in a number of multiple anomaly syndromes such as Kabuki syndrome.
With talipes equinovarus, the affected foot is shorter than normal. The entire foot is inverted and supinated, and the forefoot is adducted. The heel is rotated inward and in plantar flexion. The position of the foot is fixed and cannot be corrected completely. It may be secondary to oligohydramnios during pregnancy, a feature of a malformation syndrome, or of a neuromuscular disorder such as spina bifida. The infant must be referred directly to a pediatric orthopedic surgeon.
Positional deformity
A closer view shows how the sole appears to be everted
With gentle pressure on the medial malleolus and the lateral aspect of the sole, the foot can be easily brought into a neutral position.
Another example of a positional deformity. In this case, the foot is dorsiflexed to such a degree that it is almost flat against the lower leg.
Here again, the foot can easily be brought back to a neutral position. A concavity due to the unusual position can be seen near the ankle, but this will resolve as the infant has full range of motion of the foot. In some cases, physical therapy that involves stretching and range of motion exercises can help speed progress.
This infant has more marked overlapping of the toes. This child did have an underlying genetic diagnosis, but there were other abnormal physical findings present.
Some newborns will have toes that seem to overlap one another.

At times, this is related to clinodactyly of one of the digits; at times, it is a positional deformity. Either way, it is considered a minor deformity. As an isolated finding, this is of no significance.
Brachial plexus lesions cause lack of active movement of the affected limb; passive movement is not painful or restricted.

The most common is Erb palsy from an upper root palsy (C5, C6, and sometimes C7).
The arm is held internally rotated and pronated in the “waiter’s tip” position.
Although most brachial plexus injuries resolve, a significant proportion have not recovered fully at 6 months of age.
Those that do not recover steadily during the first 2 months of life or are severe should be seen by a specialist because surgical repair may be indicated.
Accompanying respiratory symptoms may be secondary to damage of phrenic nerve roots.
Child's age

Birth
Diagnosis of injury

Radiographic studies

Neurologic evaluation

Cervical
Chest
Shoulder
Arm

1 month

Physical medicine evaluation
(every month)

No significant improvement

Significant improvement

No further improvement

Continued improvement

3-4 months

Brachial plexus exploration

Physical therapy implementation

3-9 months

Improvement

Lack of improvement in certain muscle groups

>18 months

Nerve, tendon, muscle transfer

Physical therapy
Figure 107-9 The Barlow test permits the early diagnosis of hip instability. It is a provocation test for dislocatability. The pelvis is steadied with one hand while the leg to be tested is grasped with the other. The thumb of the examiner's hand should lie over the lesser trochanter and the tip of the middle finger over the greater trochanter. With the hip and knee in flexion, gentle pressure is exerted with the thumb over the lesser trochanter. Dislocatability is manifested by a sudden shift of the proximal femur. Dislocation may be reduced with the Ortolani test.
Figure 107-10 In the Ortolani test, the pelvis is held steady with one hand while the limb to be examined is grasped with the other. The hip and knee are flexed. While gently pulling the femur forward, the examiner abducts the limb under examination, using the greater trochanter as a fulcrum. Reduction of dislocation is manifested by a sudden shift in the position of the proximal femur, often accompanied by a palpable or audible clunk. There must be sufficient ligamentous laxity to permit relocation of the proximal femur into the acetabulum; therefore, the test is useful primarily in the neonatal period.
Physical exam positive
  + Refer to orthopedist
  + Do not use triple diapers

Physical exam inconclusive
  + Follow-up examination at 2 weeks

Physical exam normal but with risk factors
  + Female or Family history + male
  + Recheck at periodic intervals
  + Optional imaging:
    - Ultrasound <5 months old
    - X-ray >4 months old

  + Family history + female or Breech + male
  + Imaging (see above)

  + Breech + female