BIRTH DEFECTS SURVEILLANCE, HARYANA

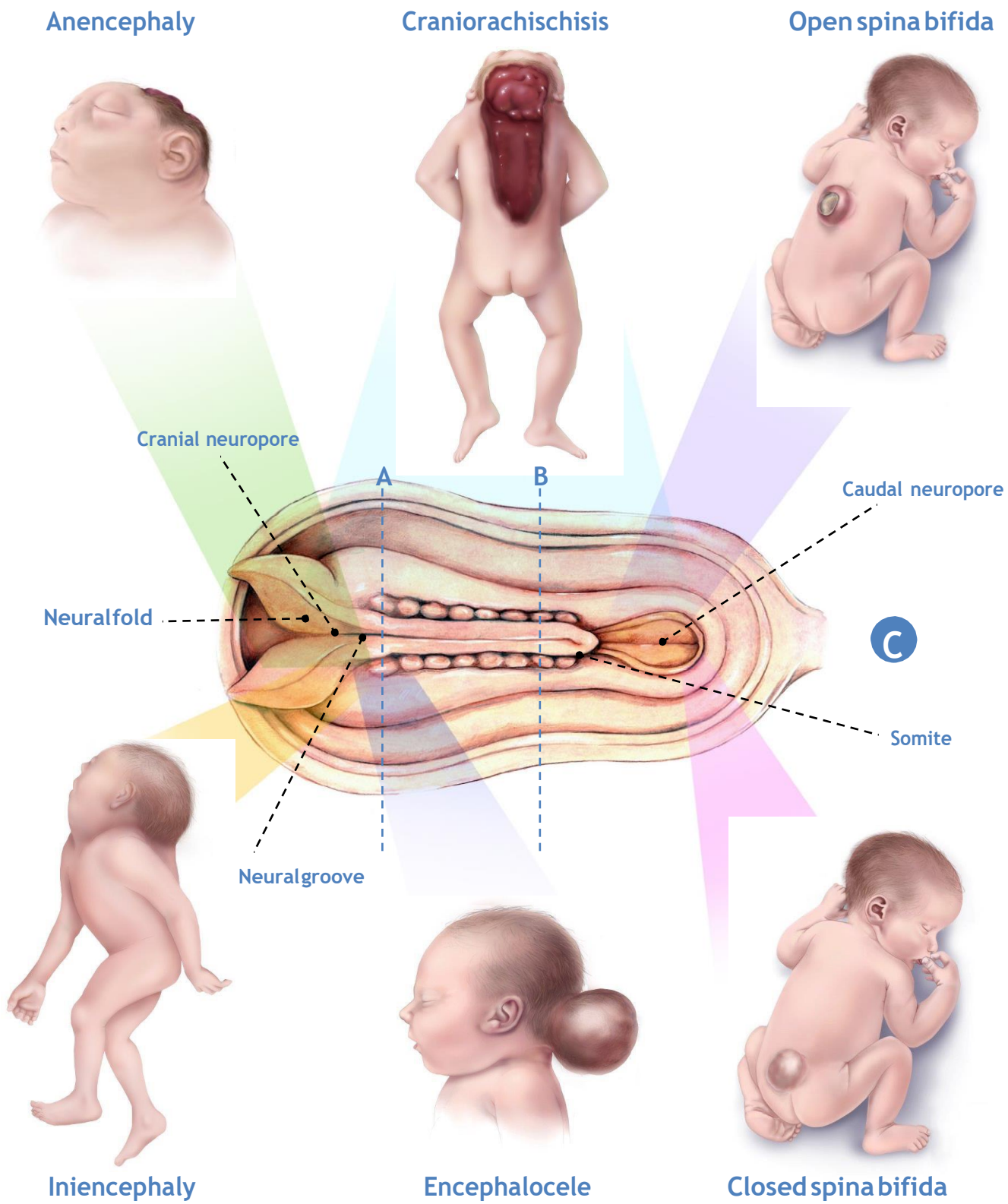
ATLAS OF SELECTED CONGENITAL ANOMALIES

I. Congenital malformations of the nervous system: neural tube defects

Neural tube defects affect the brain and spinal cord, and are among the most common of the congenital anomalies

- In anencephaly, the absence of the brain and calvaria can be total or partial. Craniorachischisis is characterized by anencephaly accompanied by a contiguous bony defect of the spine and exposure of neural tissue.
- In open spina bifida, a bony defect of the posterior vertebral arches (in this case, the lower thoracic vertebrae) is accompanied by herniation of neural tissue and meninges and is not covered by skin.
- In iniencephaly, dysraphia in the occipital region is accompanied by severe retroflexion of the neck and trunk.
- In encephalocele, the brain and meninges herniate through a defect in the calvaria.
- In closed spina bifida unlike open spina bifida, the bony defect of the posterior vertebral arches (in this case, the lumbar vertebrae), the herniated meninges, and neural tissue are covered by skin.
- The most prevalent types of neural tube defects are anencephaly, encephalocele and spina bifida.

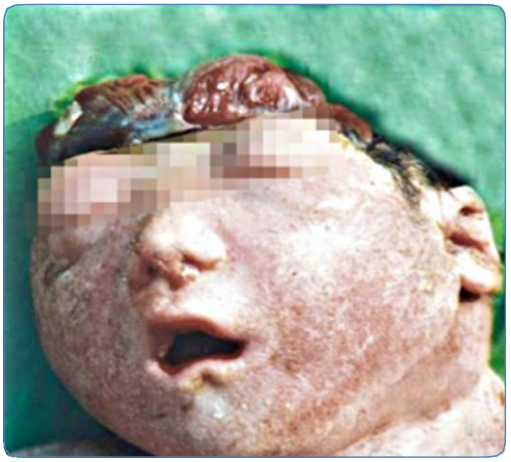
Neural Tube defects (continued)



Neural Tube defects (continued)

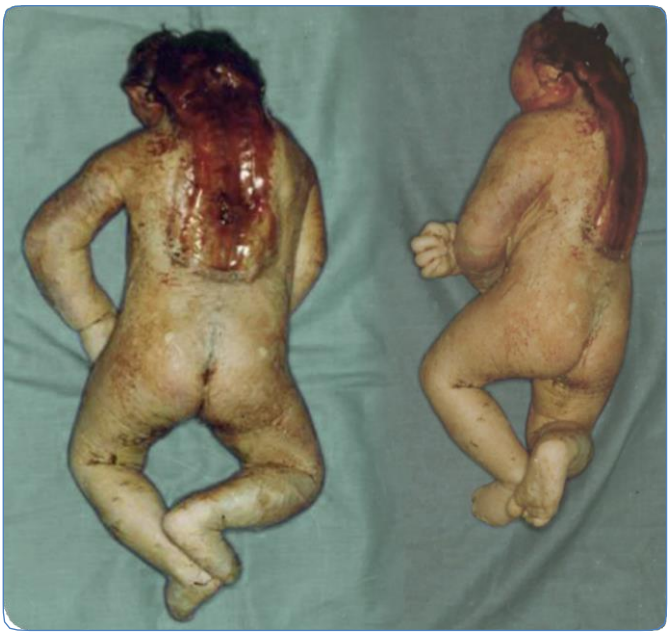
Anencephaly (Q00.0)

A total or partial absence of the brain, together with total or partial absence of the cranial vault and the covering skin.

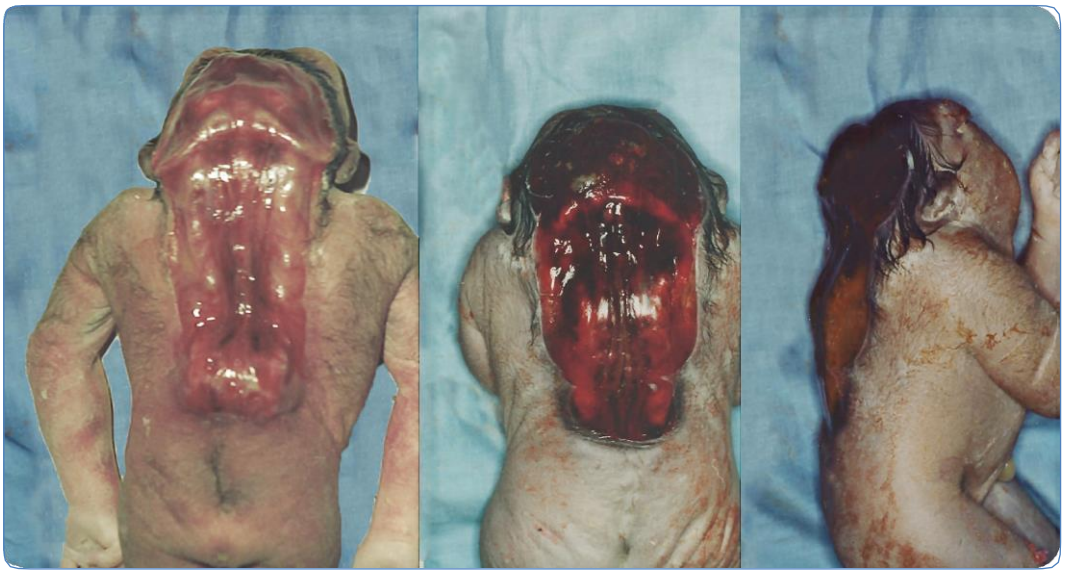


Craniorachischisis (Q00.1)

The presence of anencephaly with a contiguous spine defect without meninges covering the neural tissue (rachischisis).

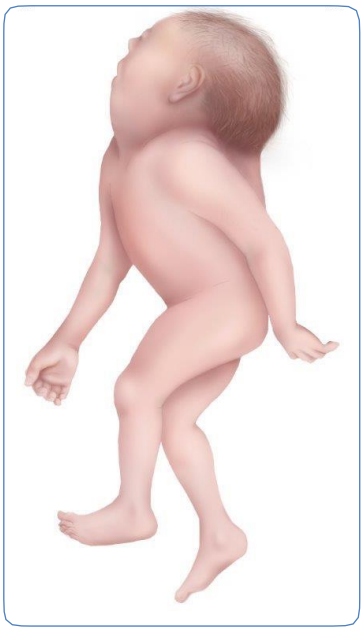


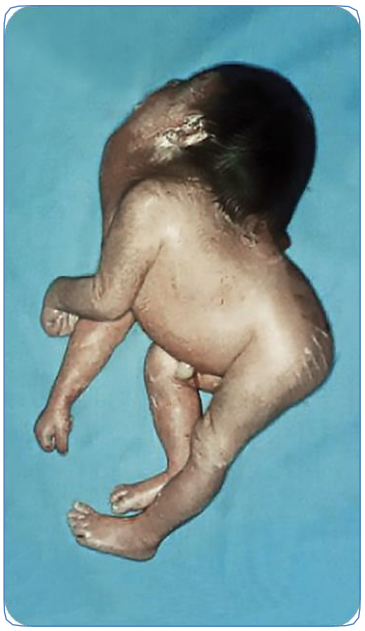
Neural Tube defects (continued)



Iniencephaly (Q00.2)

A rare and complex neural tube defect characterized by extreme retroflexion of the head, variably combined with rachischisis of the cervical and thoracic spine and, more rarely, with occipital encephalocele. In iniencephaly, the cranium is always closed.

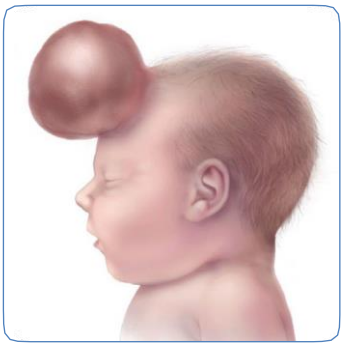
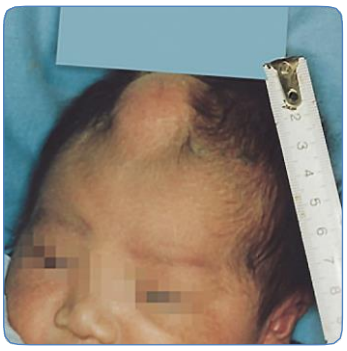
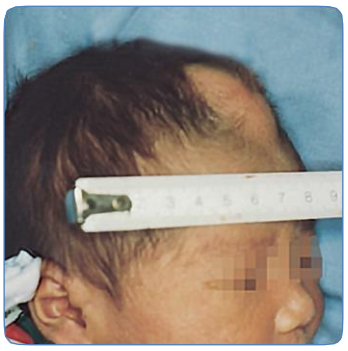




Neural Tube defects (continued)

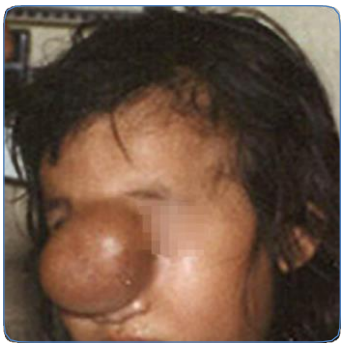
Frontal encephalocele (Q01.0)

Herniation of brain tissue, usually covered by meninges, through a defect in the frontal bone.

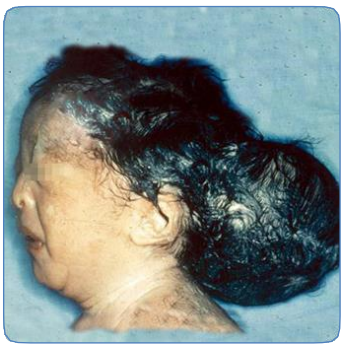
Nasofrontal encephalocele (Q01.1)

Herniation of brain tissue, usually covered by meninges, through an opening between the frontal bone and the nasal and ethmoid bones.

Occipital encephalocele (Q01.2)

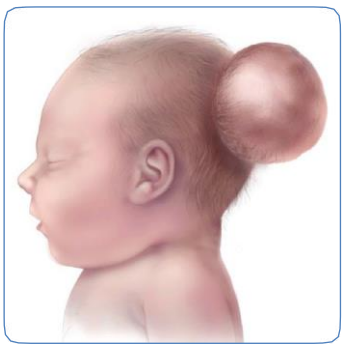
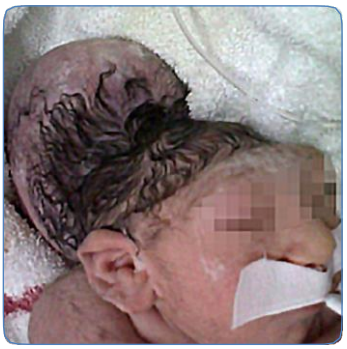
Herniation of brain tissue, usually covered by meninges, through an opening in the occipital bone.

Neural Tube defects (continued)

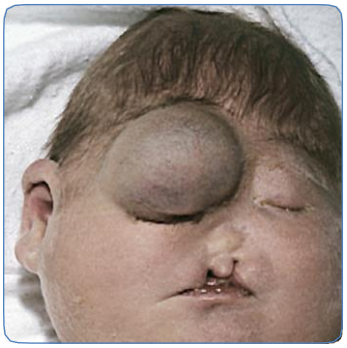
Parietal encephalocele (Q01.80)

Herniation of brain tissue, usually covered by meninges, through an opening in one of the parietal bones.

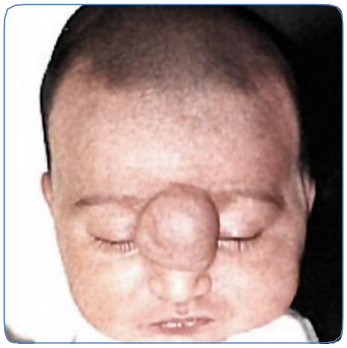
Orbital encephalocele (Q01.81)

Herniation of brain tissue, usually covered by meninges, through one of the orbits.

Nasal encephalocele (Q01.82)

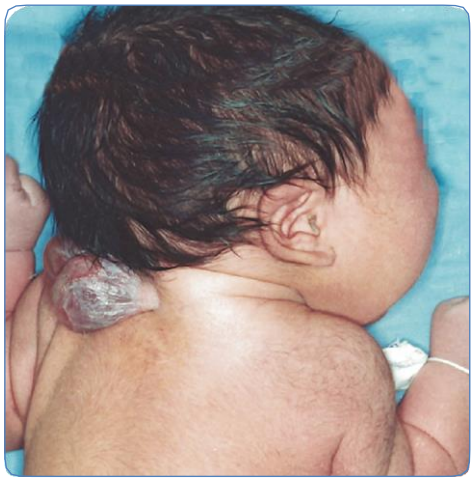
Herniation of brain tissue, usually covered by meninges, through an opening in the nasal region.

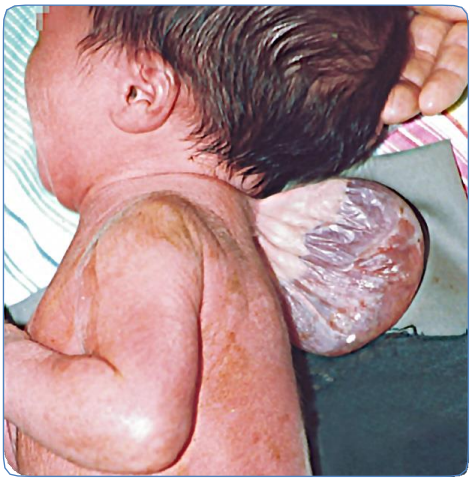



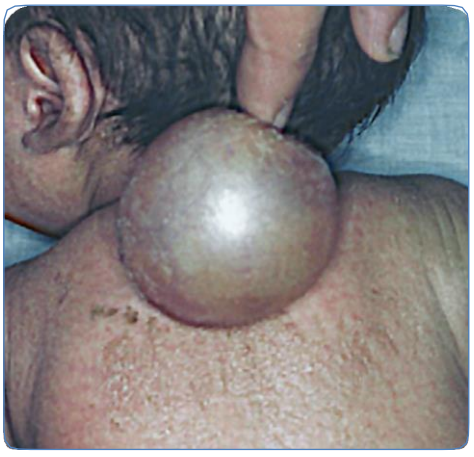
Neural Tube defects (continued)

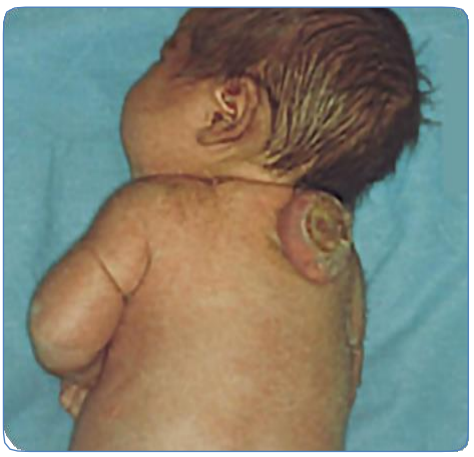
Cervical spina bifida

Most cases of cervical spina bifida will eventually develop hydrocephalus, although this may not be immediately obvious at birth; therefore, coding for cervical spina bifida with or without hydrocephalus could be difficult at birth. Close follow-up of these neonates is important for consideration of shunt surgery.

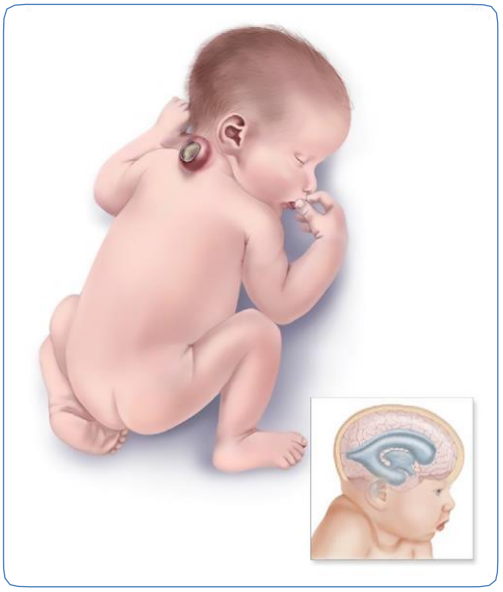






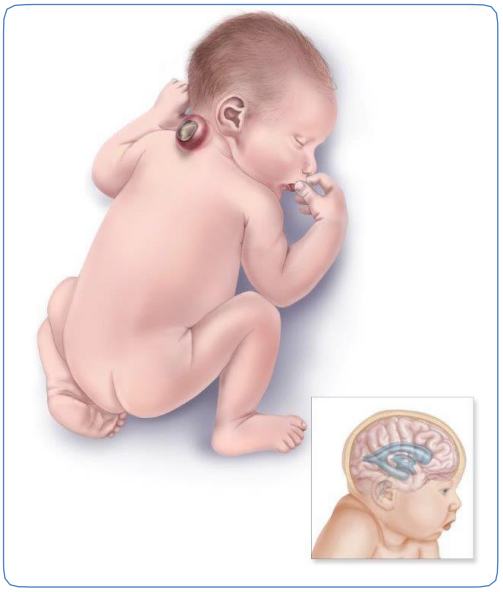


Neural Tube defects (continued)



Cervical spina bifida with hydrocephalus (Q05.0)

Protrusion of meninges and/or spinal cord through an opening in the cervical region of the spine, associated with hydrocephalus. It may be open, with or without membrane covering; or closed, covered by normal-appearing skin. Hydrocephalus is present as a result of obstruction to the flow of cerebrospinal fluid, secondary to the hindbrain herniation associated with Chiari II malformation.



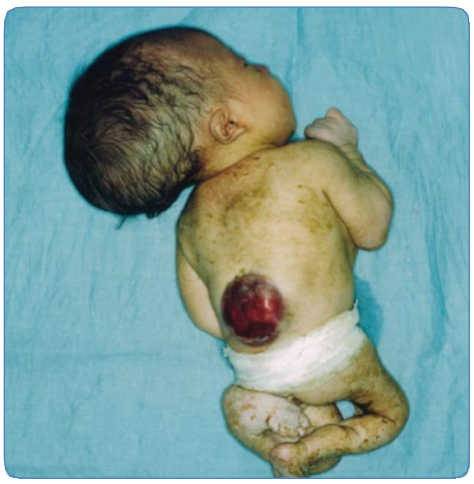
Cervical spina bifida without hydrocephalus (Q05.5)

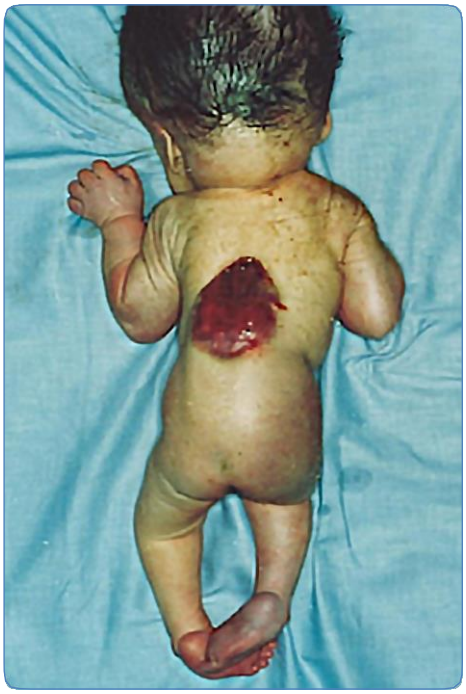
Protrusion of meninges and/or spinal cord through an opening in the cervical region of the vertebral column. It may be open, with or without membrane covering; or closed, covered by normal-appearing skin.

Neural Tube defects (continued)

Thoracic spina bifida

Most cases of thoracic spina bifida will eventually develop hydrocephalus, although this may not be immediately obvious at birth; therefore, coding for thoracic spina bifida with or without hydrocephalus could be difficult at birth. Close follow-up of these neonates is important for consideration of shunt surgery.





Neural Tube defects (continued)

Thoracic spina bifida with hydrocephalus (Q05.1)

Protrusion of meninges and/or spinal cord through an opening in the thoracic region of the vertebral column, associated with hydrocephalus. It may be open, with or without membrane covering; or closed, covered by normal-appearing skin. Hydrocephalus is present as a result of obstruction to the flow of cerebrospinal fluid, secondary to the hindbrain herniation associated with Chiari II malformation.

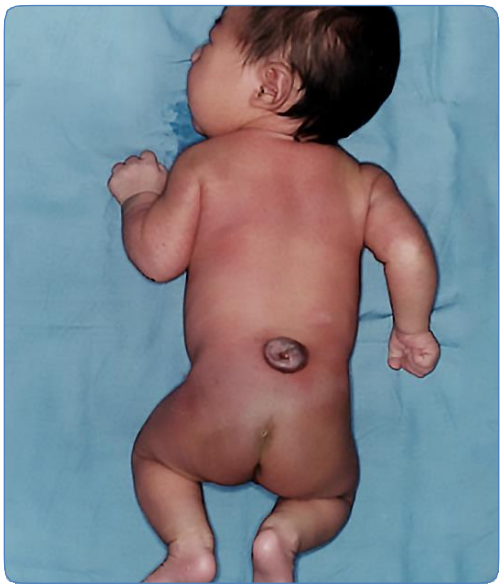
Thoracic spina bifida without hydrocephalus (Q05.6)

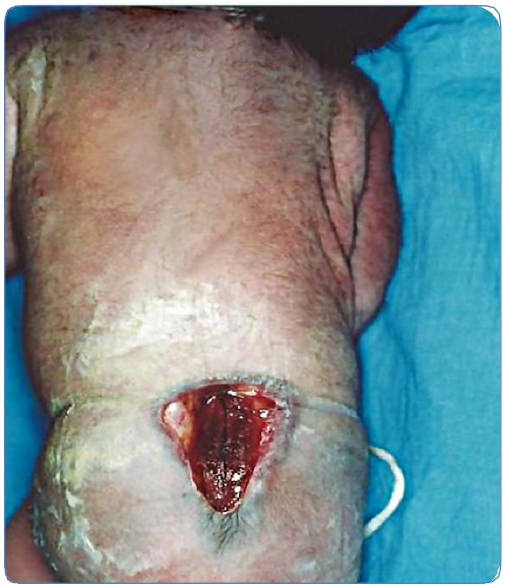
Protrusion of meninges and/or spinal cord through an opening in the thoracic region of the vertebral column. It may be open, with or without membrane covering; or closed, covered by normal-appearing skin.

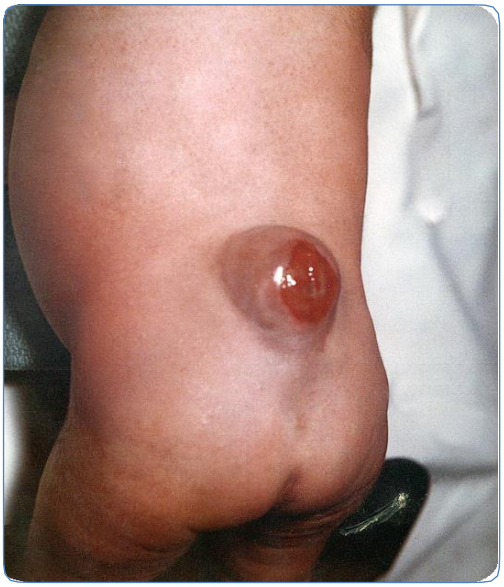
Neural Tube defects (continued)

Lumbar spina bífida

Some cases of lumbar spina bifida will eventually develop hydrocephalus, and this may not be immediately obvious at birth; therefore, coding for lumbar spina bifida with or without hydrocephalus could be difficult at birth. Close follow-up of these neonates is important for consideration of shunt surgery.



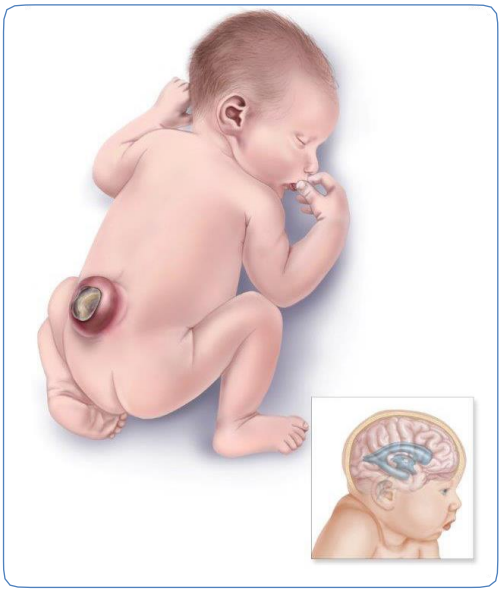




Neural Tube defects (continued)

Lumbar spina bifida with hydrocephalus (Q05.2)

Protrusion of meninges and/or spinal cord through an opening in the lumbar region of the vertebral column, associated with hydrocephalus. It may be open, with or without membrane covering; or closed, covered by normal-appearing skin. Hydrocephalus is present as a result of obstruction to the flow of cerebrospinal fluid, secondary to the hindbrain herniation associated with Chiari II malformation.



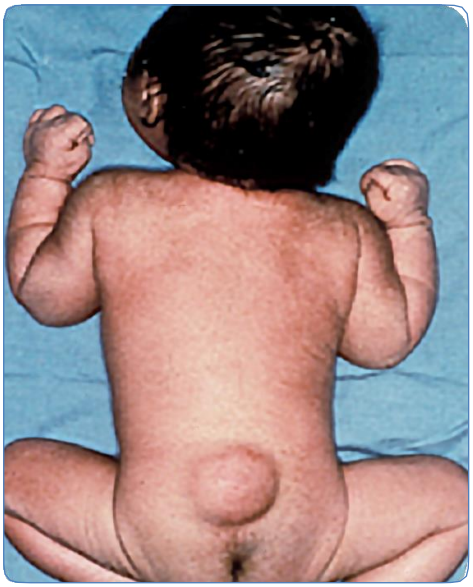
Lumbar spina bifida without hydrocephalus (Q05.7)

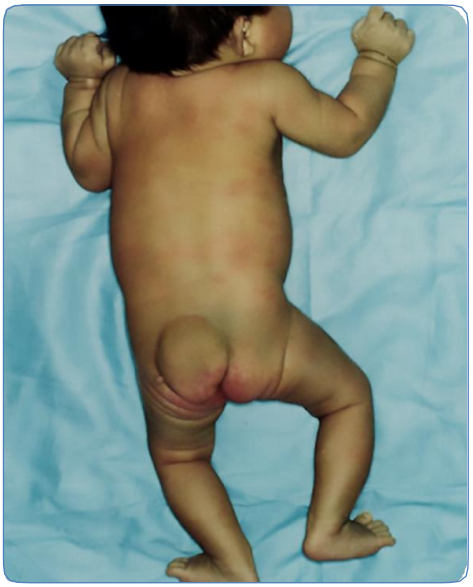
Protrusion of meninges and/or spinal cord through an opening in the lumbar region of the vertebral column. It may be open, with or without membrane covering; or closed, covered by normal-appearing skin.

Neural Tube defects (continued)

Sacral spina bifida

Few cases of sacral spina bifida will develop hydrocephalus, compared with the other types of spina bifida. The presence of hydrocephalus in infants with sacral spina bifida may not be immediately obvious at birth; therefore, coding for sacral spina bifida with or without hydrocephalus could be difficult at birth. Close follow-up of these neonates is important for consideration of shunt surgery.





Neural Tube defects (continued)

Sacral spina bifida with hydrocephalus (Q05.3)

Protrusion of meninges and/or spinal cord through an opening in the sacral region of the vertebral column, associated with hydrocephalus. It may be open, with or without membrane covering; or closed, covered by normal-appearing skin. Hydrocephalus is present as a result of obstruction to the flow of cerebrospinal fluid, secondary to the hindbrain herniation associated with Chiari II malformation.

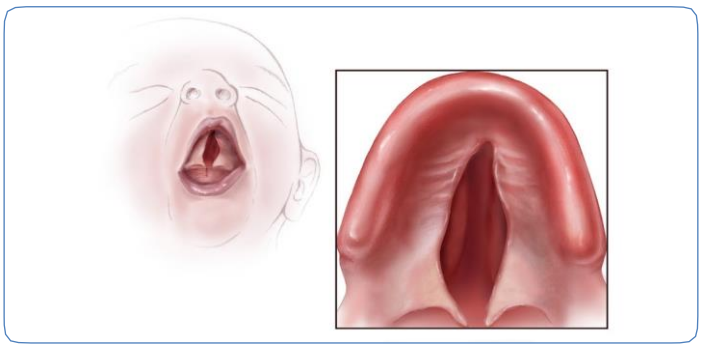
Sacral spina bifida without hydrocephalus (Q05.8)

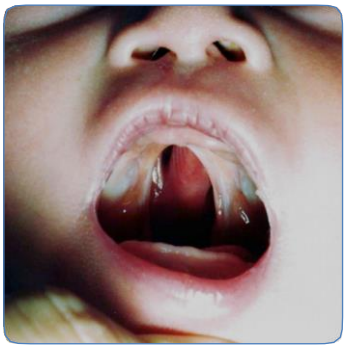
Protrusion of meninges and/or spinal cord through an opening in the sacral region of the vertebral column. It may be open, with or without membrane covering; or closed, covered by normal-appearing skin.

II. Cleft palate and cleft lip

Cleft palate (Q35.5)

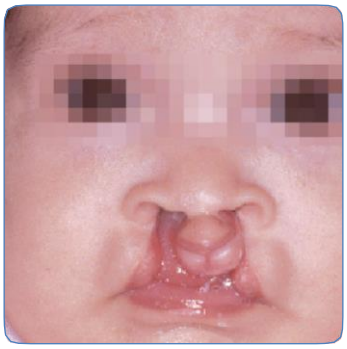
Fissure of the palate, which can affect the soft and hard palate, or only the soft palate.





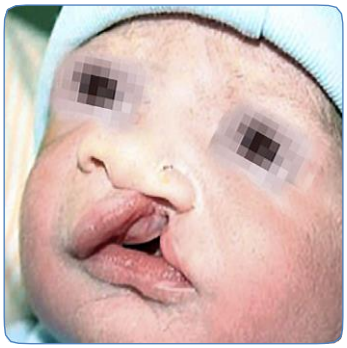
Cleft lip, bilateral (Q36.0)

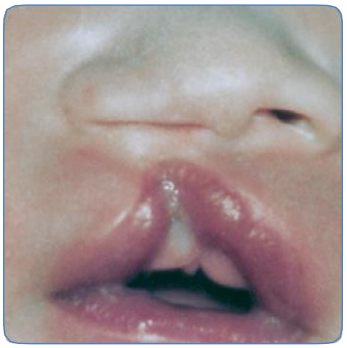
Partial or complete bilateral fissure of the upper lip that may be associated with a cleft of the gum.



Cleft lip, specified as unilateral (Q36.9, Q36.90)

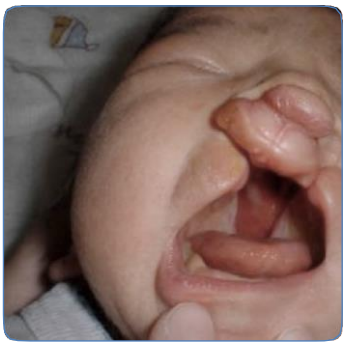
Partial or complete unilateral fissure of the upper lip that may be associated with a cleft of the gum.





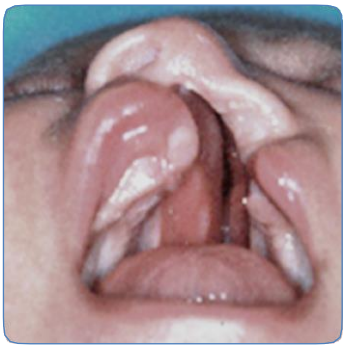
Cleft hard palate with bilateral cleft lip (Q37.0)

Partial or complete bilateral fissure of the upper lip, associated with a fissure of the palate.



Cleft hard palate with cleft lip, specified as unilateral (Q37.10)

Partial or complete unilateral fissure of the upper lip, associated with a fissure of the palate.



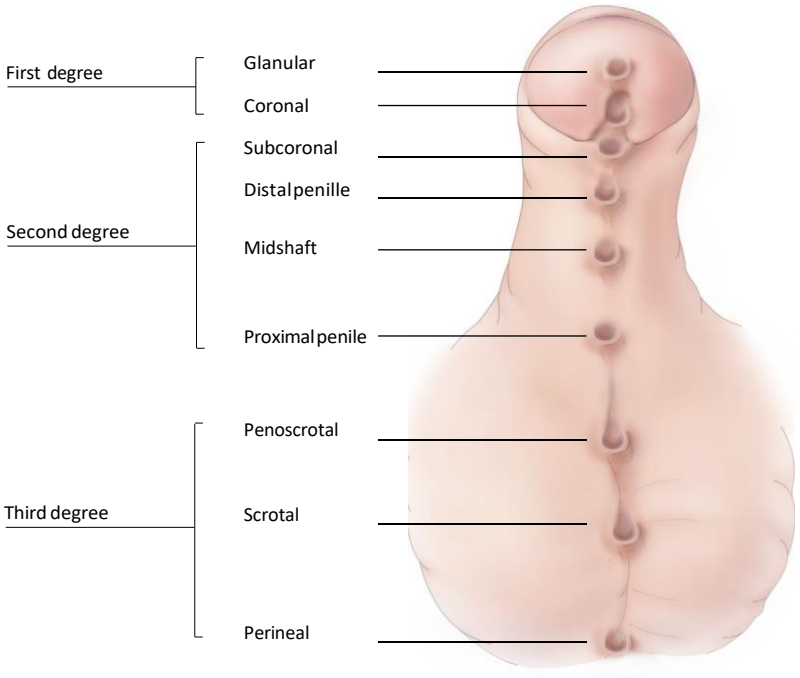
III. Congenital malformations of genital organs:

Hypospadias (Q54, Q54.0, Q54.1, Q54.2, Q54.3, Q54.8, Q54.9)

Displacement of the urethral meatus ventrally and proximally from the tip of the penis. It is classified according to the position of the meatus on the penis:

- First degree (Q54.0): the urethral meatus is located on either the glans (glanular hypospadias) or the corona (coronal hypospadias).
- Second degree (Q54.1): the urethral meatus is located in the balanopenile furrow or coronal sulcus (sub-coronal hypospadias) or in the shaft of the penis (distal penile, midshaft, and proximal penile hypospadias).
- Third degree (Q54.2, Q54.3): the urethral meatus is located in the junction of the penis and the scrotum (penoscrotal or scrotal hypospadias) or the perineum (perineoscrotal, perineal, or pseudovaginal hypospadias).

The shortening of the ventral side of the penis found in hypospadias can result in a penile curvature, known as chordee. This is present more commonly in severe cases, but also can occur independently of hypospadias.



Subtypes:

Q54 Hypospadias (avoid using this general code if more specific information is available)

Q54.0 Hypospadias, balanic coronal glanular

Q54.1 Hypospadias, penile (subcoronal hypospadias)

Q54.2 Hypospadias, penoscrotal

Q54.3 Hypospadias, perineal

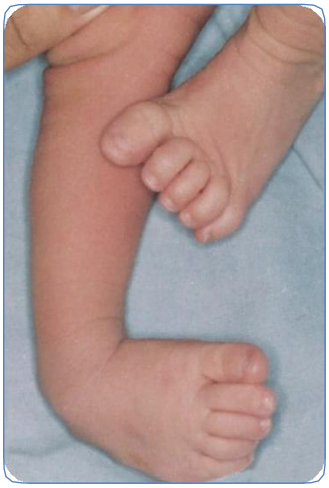
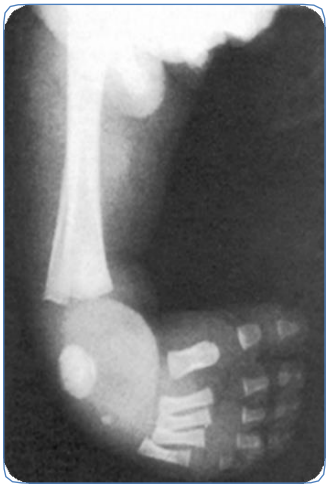
Q54.8 Other hypospadias, excludes: female hypospadias (**Q52.81**)

Q54.9 Hypospadias, unspecified

IV. Congenital malformations and deformations of the musculoskeletal system:

Talipes equinovarus (Q66.0)

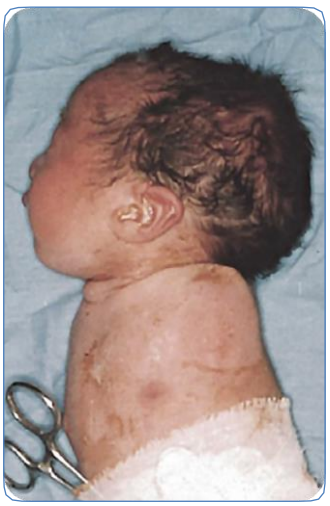
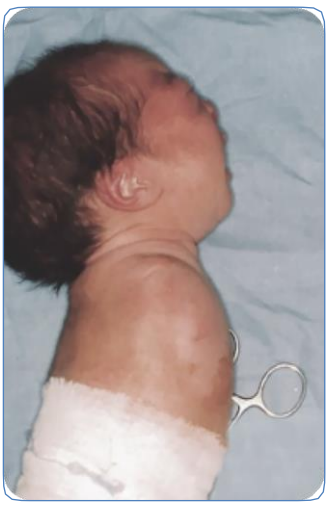
Combination of forefoot and hindfoot in equinus (plantar flexed) and in varus (rotated toward the midline). In other words, the foot points downward and inward and is rotated outward axially. Other anomalies of the foot and ankle include talipes calcaneovalgus (in which the ankle joint is dorsiflexed and the forefoot deviated outwards) and talipes calcaneovarus (in which the ankle joint is dorsiflexed and the forefoot deviated inwards).

Reduction defects of upper and lower limbs

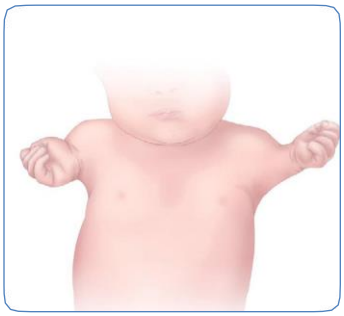
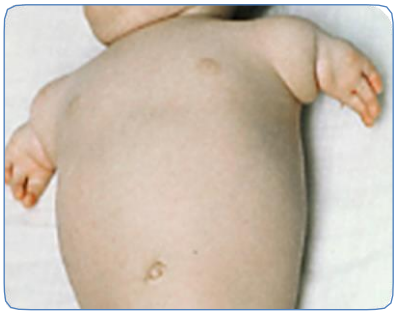
Congenital complete absence of upper limb(s); amelia of upper limb (Q71.0)

Complete absence of one or both upper limbs.

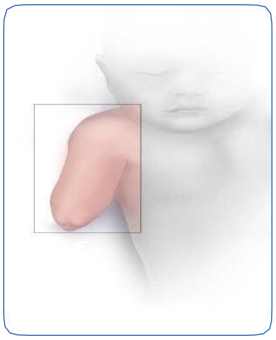
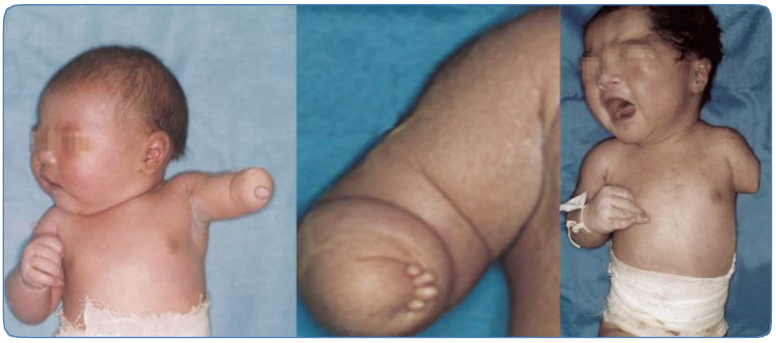
Congenital absence of upper arm and forearm with hand present; phocomelia of upper limb (Q71.1)

Complete or partial absence of the upper arm and forearm but with the hand present.

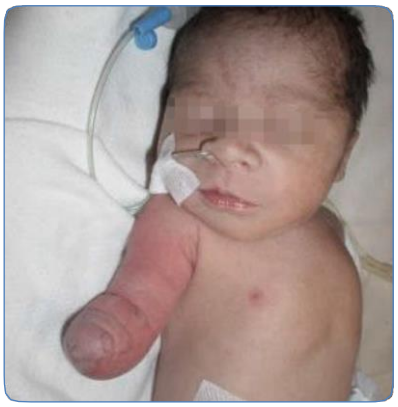
Congenital absence of both forearm and hand (Q71.2)

Complete or partial absence of both the forearm and hand.

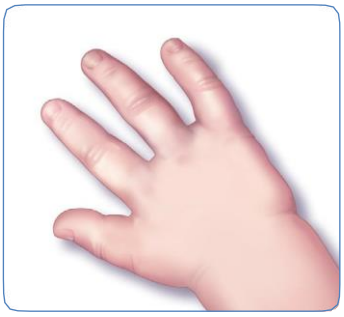
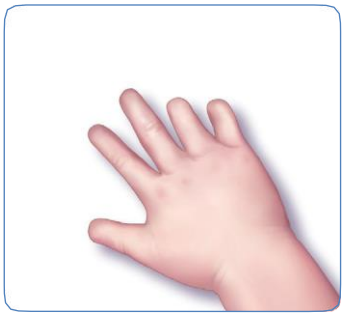
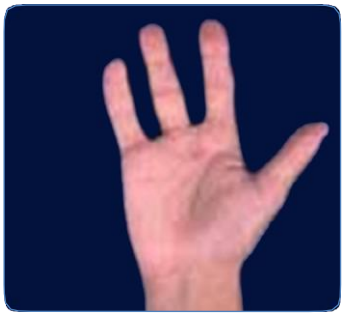
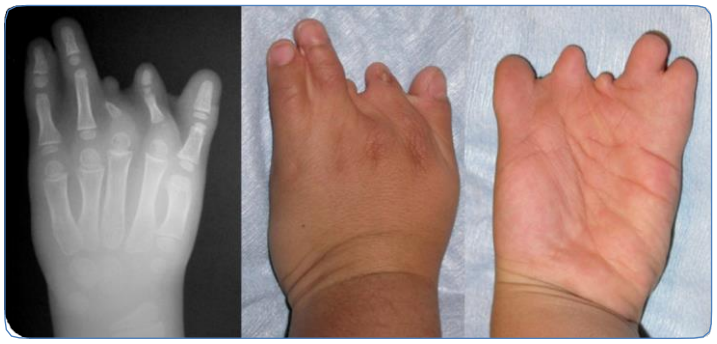
Congenital absence of hand and finger(s) (Q71.3)

Complete or partial absence of the hand and finger(s).

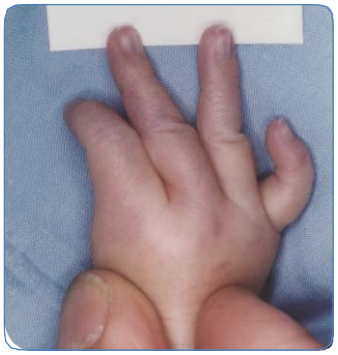
Congenital absence of finger(s) (remainder of hand intact) (Q71.30)

Complete or partial absence of fingers, with the remainder of the hand intact.

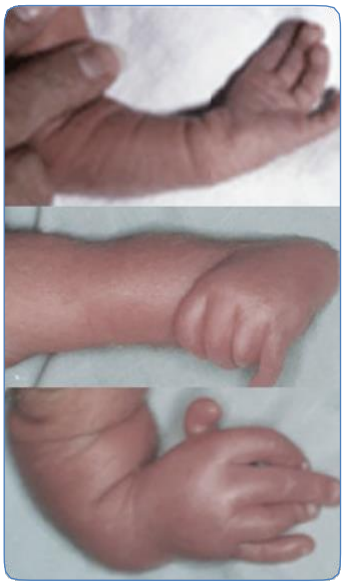
Absence or hypoplasia of thumb (other digits intact) (Q71.31)

Complete or partial absence or hypoplasia of the thumb.



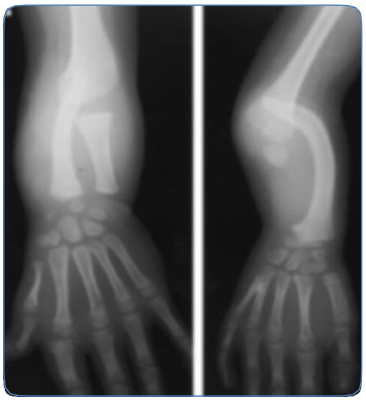
Longitudinal reduction defect of radius: clubhand (congenital), radial clubhand, absence of radius (Q71.4)

Complete or partial radial aplasia/hypoplasia. Usually accompanied by complete or partial absence or hypoplasia of the thumb.



Longitudinal reduction defect of ulna (Q71.5)

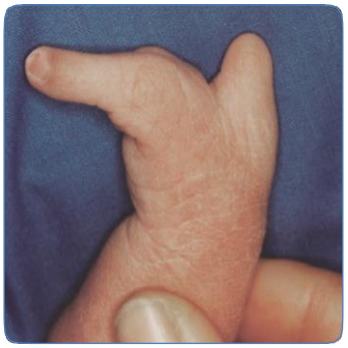
Complete or partial absence of the ulna.



Split hand (congenital cleft hand) (Q71.6)

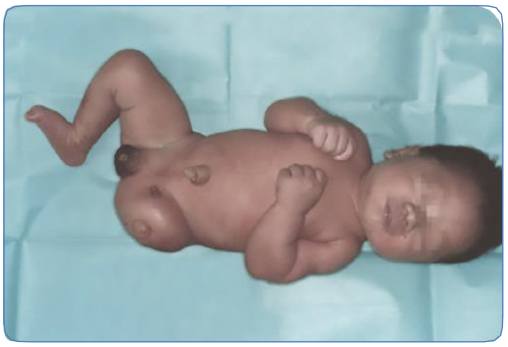
Complete or partial absence of central fingers and metacarpals. The terms lobster claw and ectrodactyly, used by some, should be discouraged.





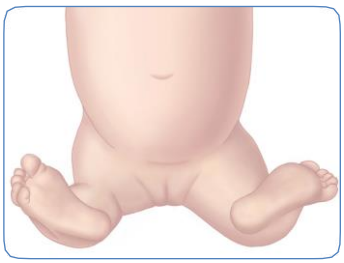
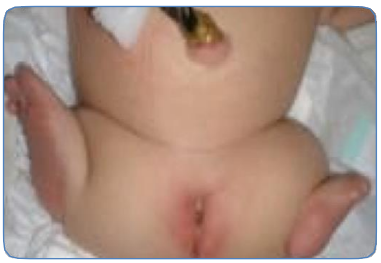
Congenital complete absence of lower limb(s); amelia of lower limb (Q72.0)

Complete absence of one or both lower limb(s).

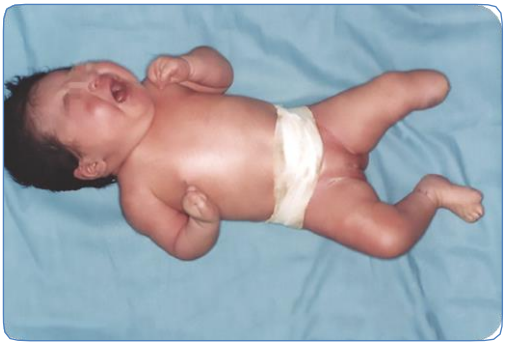
Congenital absence of thigh and lower leg with foot present; phocomelia of lower limb (Q72.1)

Complete or partial absence of the thigh and lower leg but with the foot present.

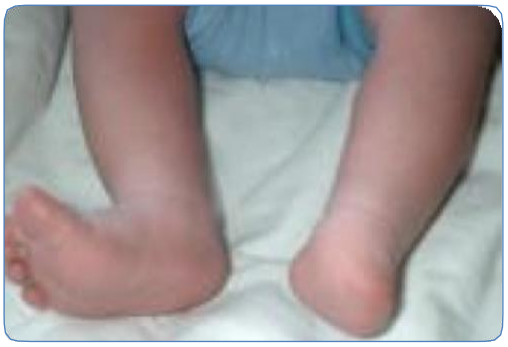
Congenital absence of both lower leg and foot (Q72.2)

Complete or partial absence of both the lower leg and foot.



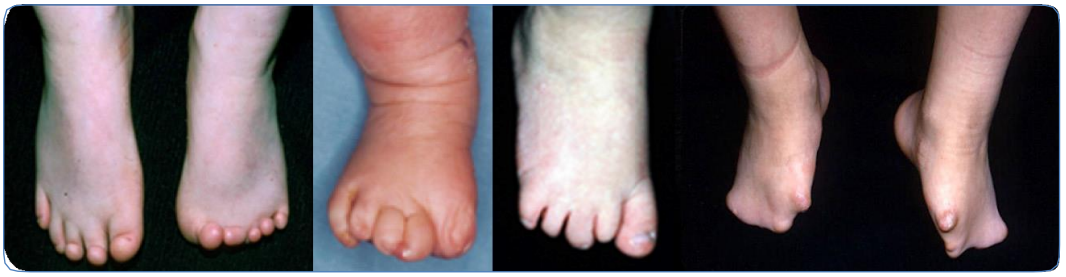
Congenital absence of foot and toe(s) (Q72.3)

Complete or partial absence of the foot and toe(s).



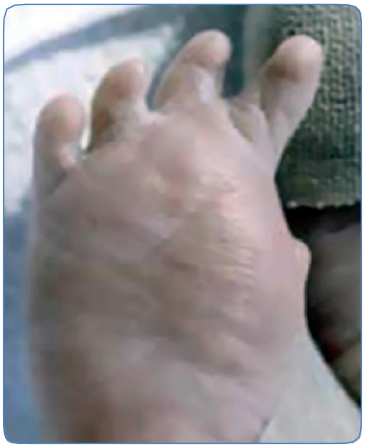
Congenital absence or hypoplasia of toe(s) with remainder of foot intact (Q72.30)

Complete absence or hypoplasia of toe(s) with the remainder of the foot intact.



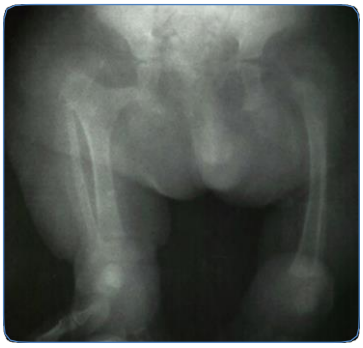
Absence or hypoplasia of first toe with other digits present (Q72.31)

Complete absence or hypoplasia of the first (great) toe with other digits present.



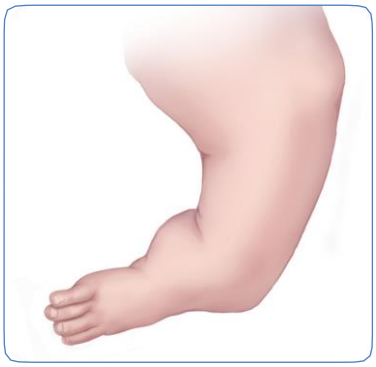
Longitudinal reduction defect of femur (Q72.4)

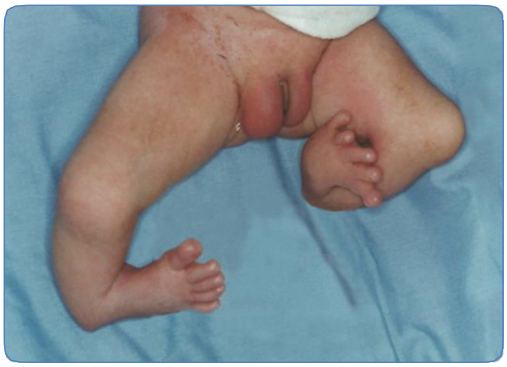
Complete or partial absence of the femur.



Longitudinal reduction defect of tibia (Q72.5)

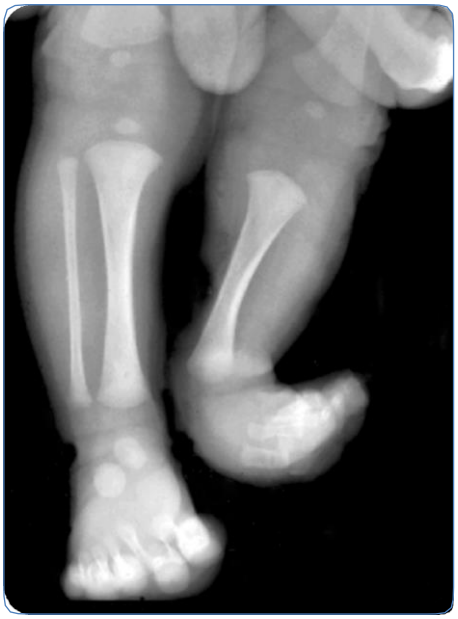
Complete or partial absence of the tibia. Usually accompanied by complete or partial absence or hypoplasia of the first (great) toe.





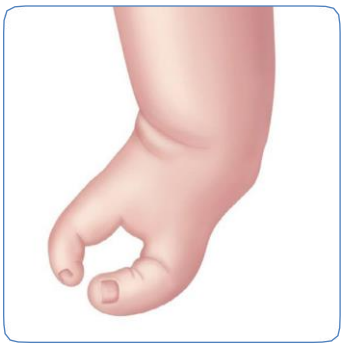
Longitudinal reduction defect of fibula; fibular aplasia/hypoplasia (Q72.6)

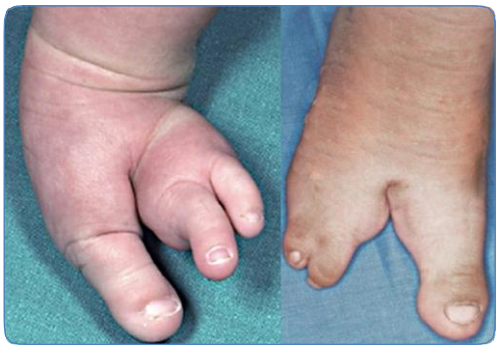
Complete or partial absence of the fibula.



Split foot (congenital cleft foot) (Q72.7)

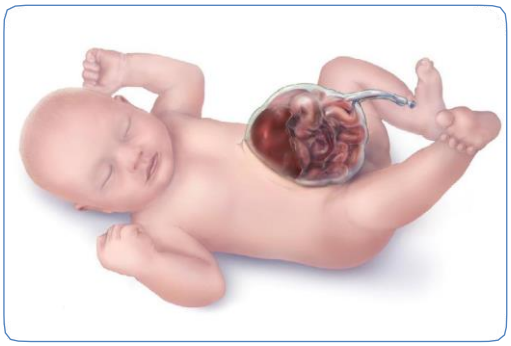
Complete or partial absence of central toes and metatarsals. The term ectrodactyly, used by some, should be discouraged.

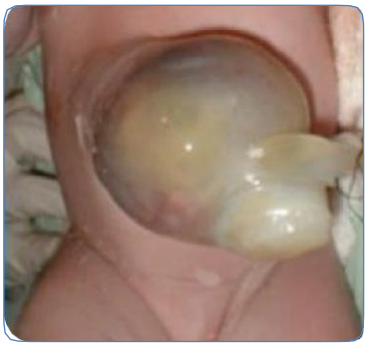


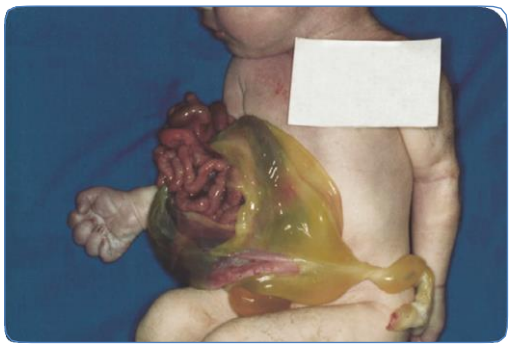


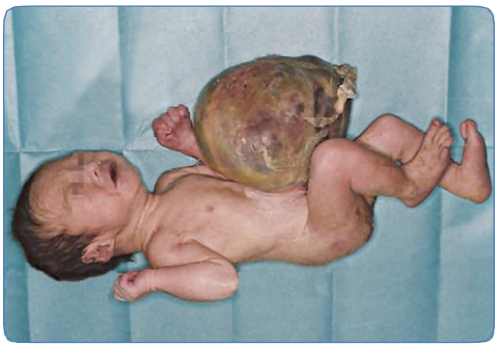
Exomphalos/omphalocele (Q79.2)

Congenital anomaly of the anterior abdominal wall, in which the abdominal contents (gut, but at times also other abdominal organs) are herniated in the midline through an enlarged umbilical ring. The umbilical cord is inserted in the distal part of the membrane covering the anomaly. The herniated organs are covered by a membrane consisting of the peritoneum and amnion (but this membrane can be ruptured).





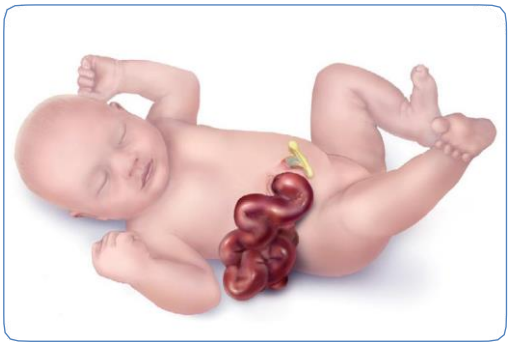


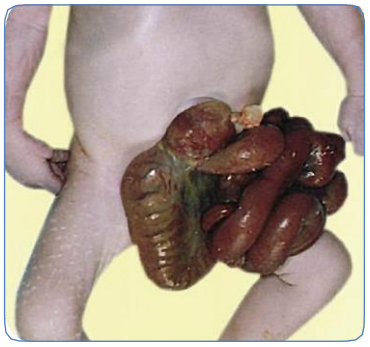


Gastroschisis and omphalocele can be confused with one another when the membrane covering the omphalocele has ruptured. In that case, the location of the abdominal opening is decisive: it is midline in omphalocele, and lateral to the umbilicus in gastroschisis.

Gastroschisis (Q79.3)

Gastroschisis is a congenital anomaly of the anterior abdominal wall, accompanied by herniation of the gut and occasionally other abdominal organs. The opening in the abdominal wall is lateral to the umbilicus, and the herniated organs lack a protective membrane. Note that the extruded abdominal contents can be matted and covered by a thick fibrous material, but this membrane does not resemble skin.





Gastroschisis and omphalocele can be confused with one another when the membrane covering the omphalocele has ruptured. In that case, the location of the abdominal opening is decisive: it is midline in omphalocele, and lateral to the umbilicus in gastroschisis.

Adapted from: Birth Defects Surveillance Atlas of Congenital Anomalies developed by World Health Organization (WHO), the National Center on Birth Defects and Developmental Disabilities (NCBDDD) from the US Centers for Disease Control and Prevention (CDC), and the International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR).