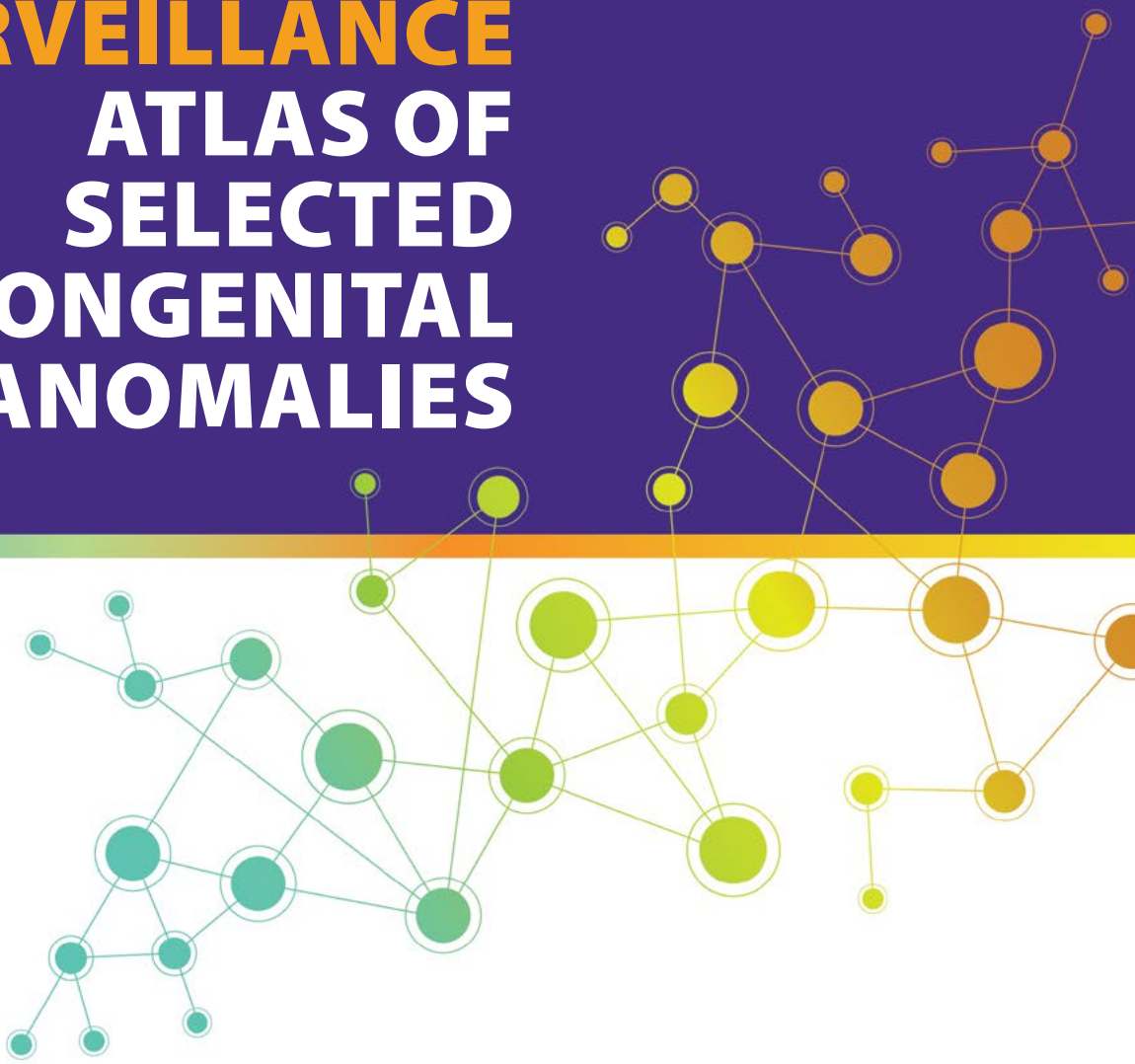
BIRTH DEFECTS SURVEILLANCE ATLAS OF SELECTED CONGENITAL ANOMALIES



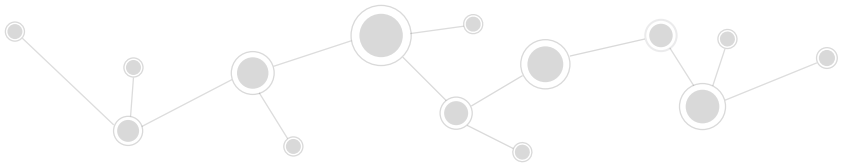
World Health Organization

CDC
Centers for Disease Control and Prevention

International Clearinghouse for Birth Defects Surveillance and Research

BIRTH DEFECTS SURVEILLANCE

ATLAS OF SELECTED CONGENITAL ANOMALIES



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Abbreviations

CDC	United States Centers for Disease Control and Prevention
ICBDSR	International Clearinghouse for Birth Defects Surveillance and Research
ICD-10	*International statistical classification of diseases and related health problems*, 10th revision
NCBDDD	National Center on Birth Defects and Developmental Disabilities
RCPCH	Royal College of Paediatrics and Child Health
USA	United States of America
WHO	World Health Organization

Objectives of the atlas

Congenital anomalies, also known as birth defects, are structural or functional abnormalities, including metabolic disorders, that are present from birth. Congenital anomalies are a diverse group of disorders of prenatal origin that can be caused by single gene defects, chromosomal disorders, multifactorial inheritance, environmental teratogens or micronutrient malnutrition.

This *Atlas of selected congenital anomalies* is a companion tool to *Birth defects surveillance: a manual for programme managers*, and is intended to help in the development, implementation and ongoing improvement of a surveillance programme for congenital anomalies, particularly in countries with limited human and financial resources.

This atlas uses the *International statistical classification of diseases and related health problems*, 10th revision (ICD-10) and the Royal College of Paediatrics and Child Health (RCPCH) extension for coding of congenital anomalies.

It provides selected illustrations and photographs of congenital anomalies that are severe enough to have a high probability of being captured during the first few days following birth. Also, because of their severity and frequency, these depicted conditions have significant public health impact, and for some there is a potential for primary prevention. When used in conjunction with the manual, the illustrations and photographs will help the reader to:

- identify an initial list of congenital anomalies to consider for monitoring;
- describe the tools needed to define and code identified cases;
- define specific congenital anomalies under surveillance.

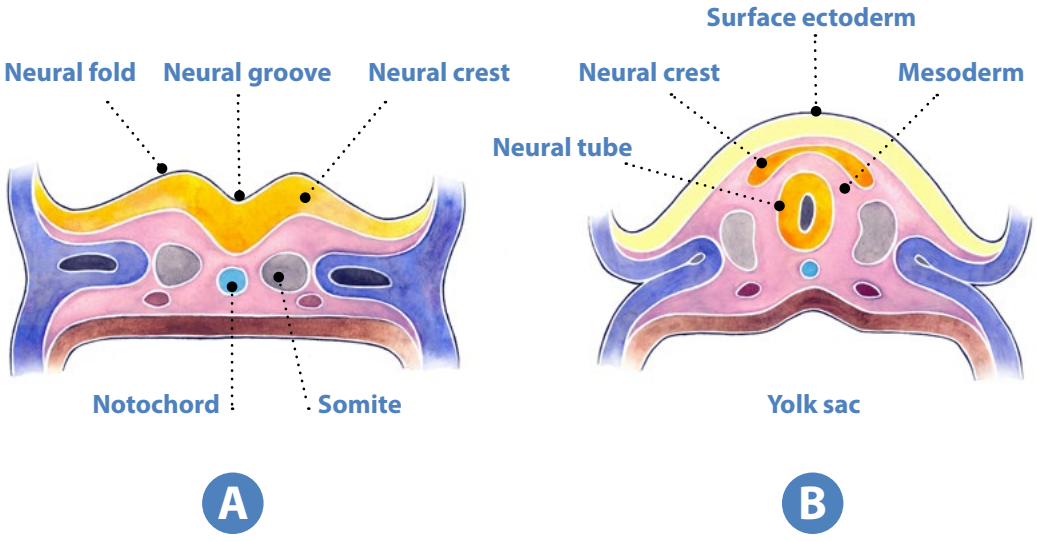
Congenital malformations of the nervous system: neural tube defects

Neural tube defects affect the brain and spinal cord, and are among the most common of the congenital anomalies (see Fig. 4.1). Panel A shows a cross section of the rostral end of the embryo at approximately three weeks after conception, showing the neural groove in the process of closing, overlying the notochord. The neural folds are the rising margins of the neural tube, topped by the neural crest, and demarcate the neural groove centrally. Panel B shows a cross section of the middle portion of the embryo after the neural tube has closed. The neural tube, which will ultimately develop into the spinal cord, is now covered by surface ectoderm (later, the skin). The intervening mesoderm will form the bony spine. The notochord is regressing. Panel C shows the developmental and clinical features of the main types of neural tube defects. The diagram in the centre is a dorsal view of a developing embryo, showing a neural tube that is closed in the centre but still open at the cranial and caudal ends. The dotted lines marked A and B refer to the cross sections shown in Panels A and B. Shaded bars point to the region of the neural tube relevant to each defect.

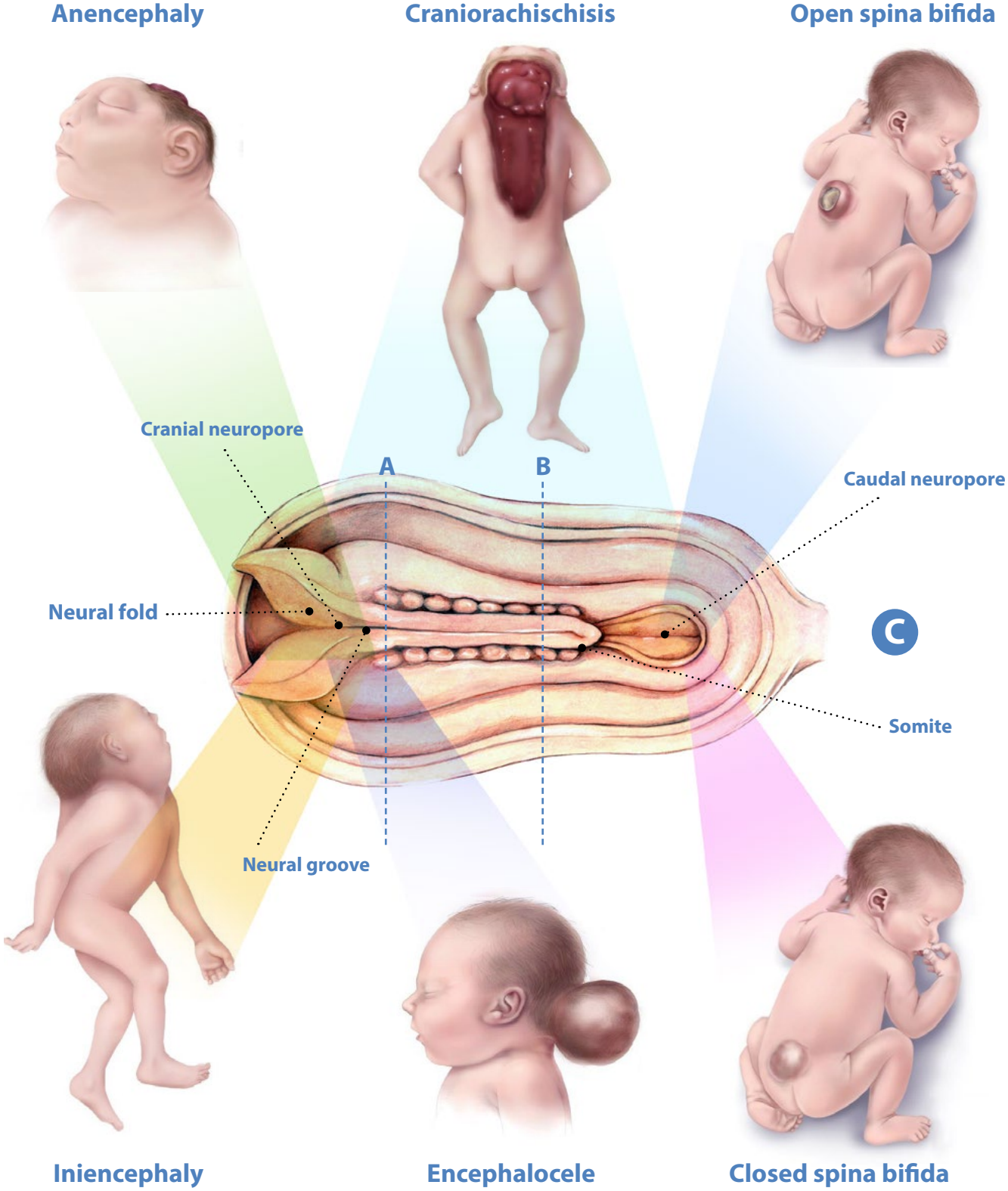
In anencephaly, the absence of the brain and calvaria can be total or partial. Craniorachischisis is characterized by anencephaly accompanied by a contiguous bony defect of the spine and exposure of neural tissue. In open spina bifida, a bony defect of the posterior vertebral arches (in this case, the 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.

Fig. 4.1. Neural tube defects

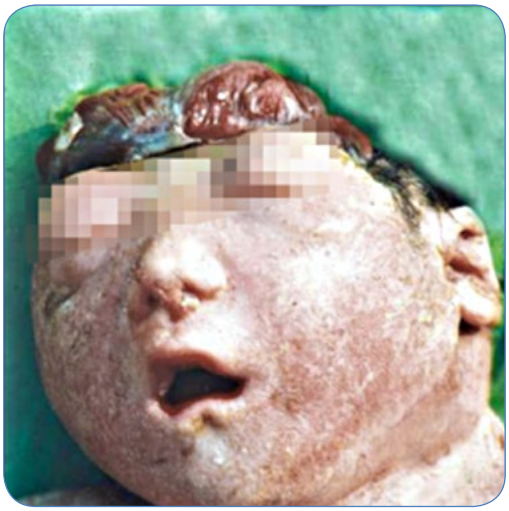


Source: adapted, with permsiion from the publisher, from Botto et al. N. Engl. J. Med. 1999;341:1509–19.


Anencephaly
Craniorachischisis
Open spina bifida
Cranial neuropore
A
B
Caudal neuropore
Neural fold
C
Somite
Neural groove
Iniencephaly
Encephalocele
Closed spina bifida

Anencephaly (Q00.0)

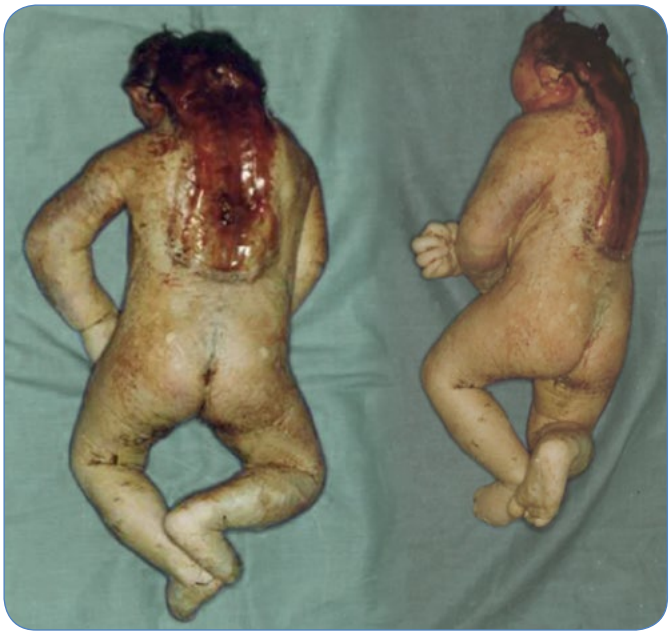
A total or partial absence of the brain, together with total or partial absence of the cranial vault and the covering skin.



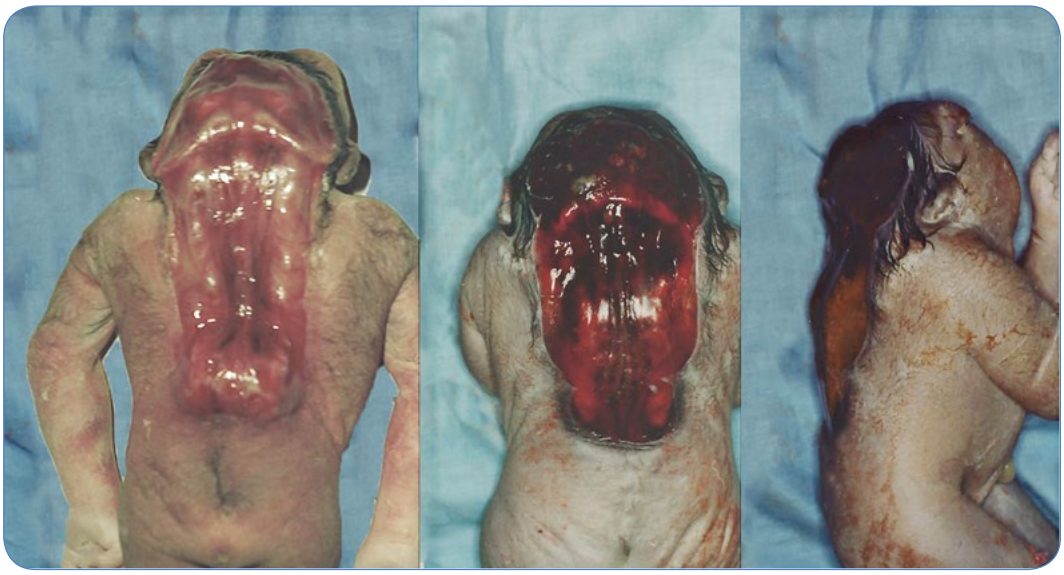
Photograph source: courtesy of CDC-Beijing Medical University collaborative project.

Craniorachischisis (Q00.1)

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



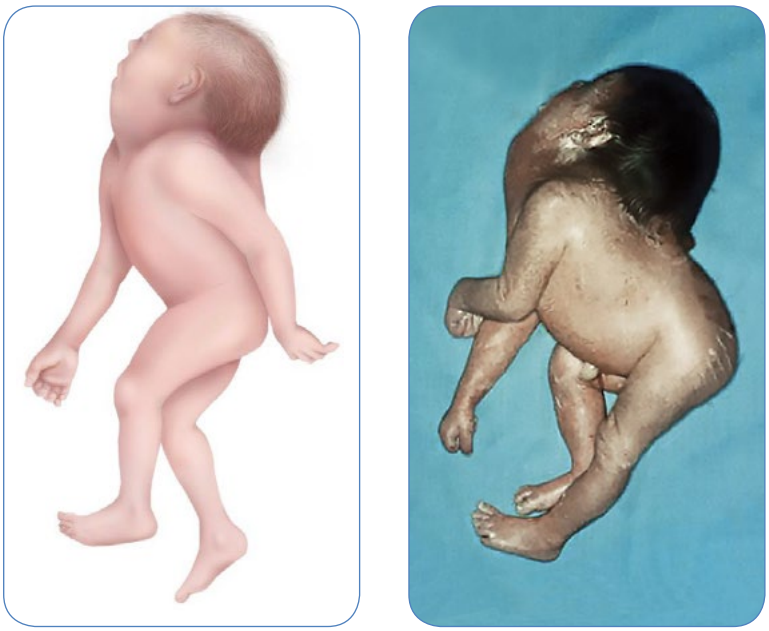
Photograph source: courtesy of CDC-Beijing Medical University collaborative project.



Photographs source: courtesy of CDC-Beijing Medical University collaborative project.

Iniencephaly (Q00.2)

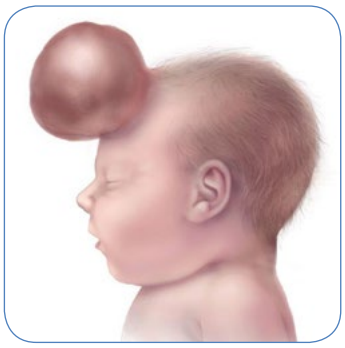
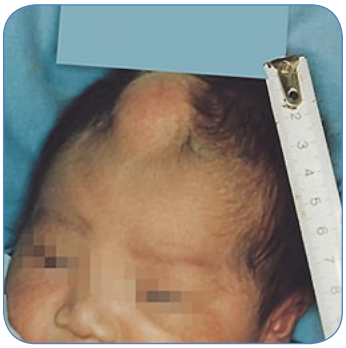
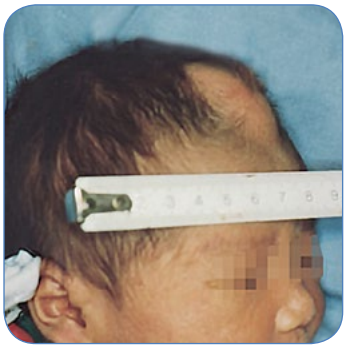
A rare and complex neural tube defect characterized by extreme retroflexion of the head, variably combined with rachischisis of the cervical and thoracic spine and, more rarely, with occipital encephalocele. In iniencephaly, the cranium is always closed.



Photograph source: courtesy of CDC-Beijing Medical University collaborative project.

Frontal encephalocele (Q01.0)

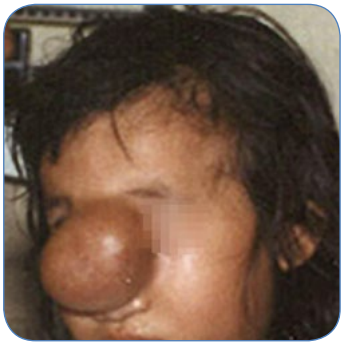
Herniation of brain tissue, usually covered by meninges, through a defect in the frontal bone.

Photographs source: courtesy of CDC-Beijing Medical University collaborative project.

Nasofrontal encephalocele (Q01.1)

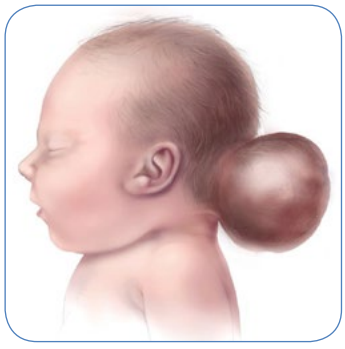
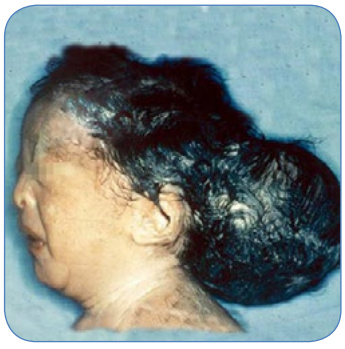
Herniation of brain tissue, usually covered by meninges, through an opening between the frontal bone and the nasal and ethmoid bones.

Photograph source: courtesy of Jaime Frías, MD, USA.

Occipital encephalocele (Q01.2)

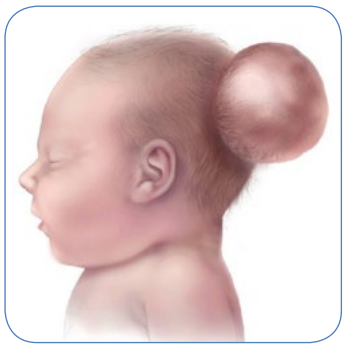
Herniation of brain tissue, usually covered by meninges, through an opening in the occipital bone.

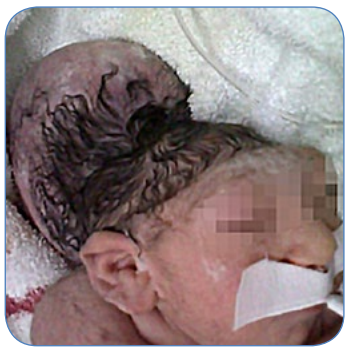



Photograph source: courtesy of CDC-Beijing Medical University collaborative project.

Parietal encephalocele (Q01.80)

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

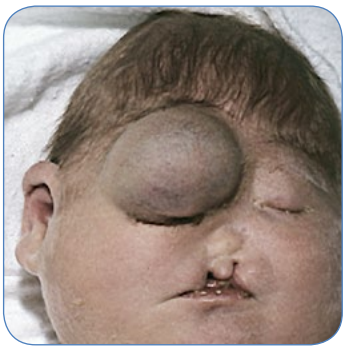




Photograph source: courtesy of Registro Nacional de Anomalías Congénitas Argentina (RENAC), Centro Nacional de Genética Médica, ANLIS, Ministerio de Salud de la Nación.

Orbital encephalocele (Q01.81)

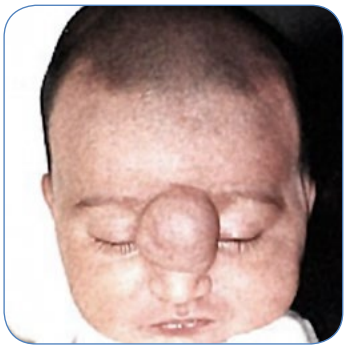
Herniation of brain tissue, usually covered by meninges, through one of the orbits.



Photograph source: courtesy of Jaime Frías, MD, USA.

Nasal encephalocele (Q01.82)

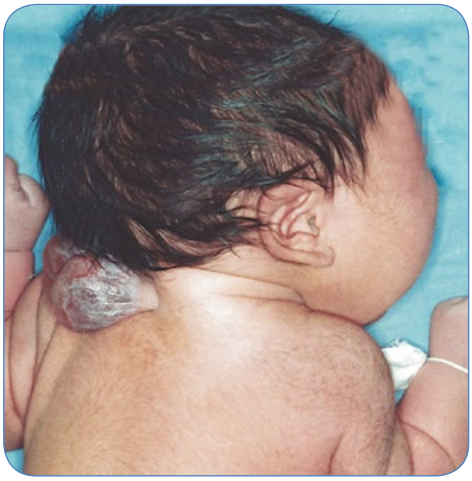
Herniation of brain tissue, usually covered by meninges, through an opening in the nasal region.

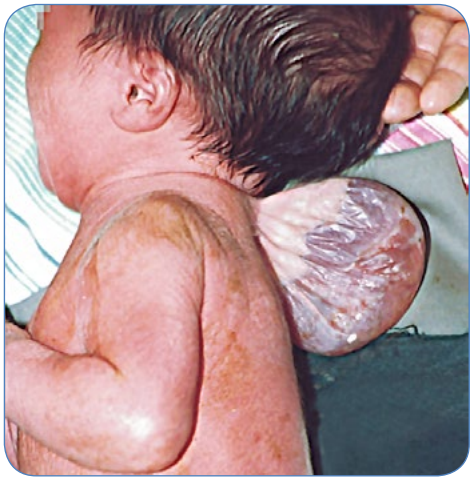


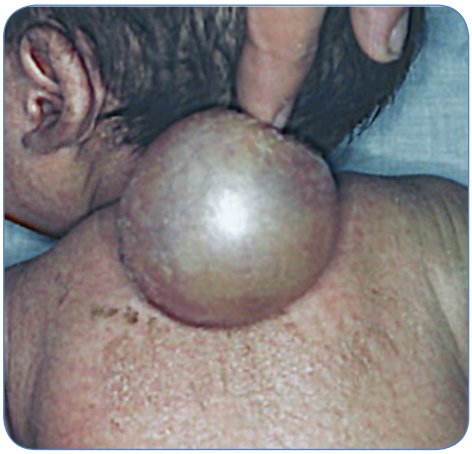
Photograph source: courtesy of Estudio Colaborativo Latino Americano de Malformaciones Congénitas (ECLAMC).

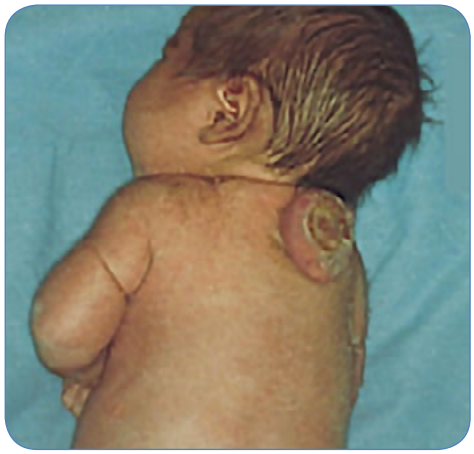
Cervical spina bifida

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.





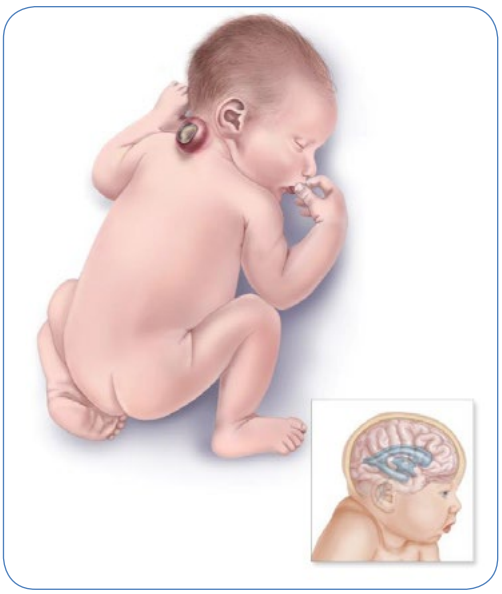




Photographs source: courtesy of CDC-Beijing Medical University collaborative project.

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.

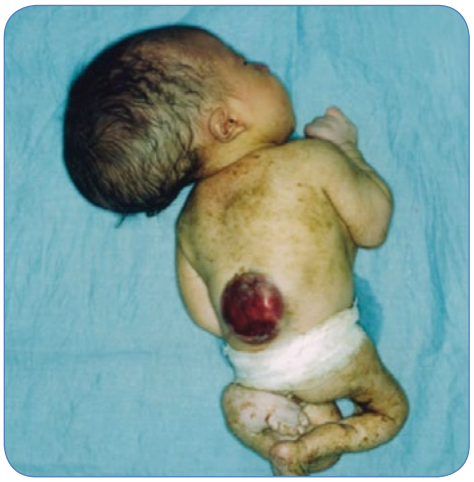


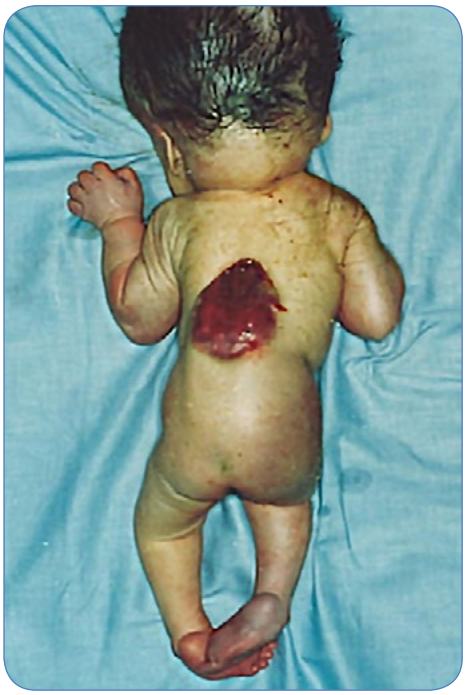
Cervical spina bifida without hydrocephalus (Q05.5)

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.

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.





Photographs source: courtesy of CDC-Beijing Medical University collaborative project.

Thoracic spina bifida with hydrocephalus (Q05.1)

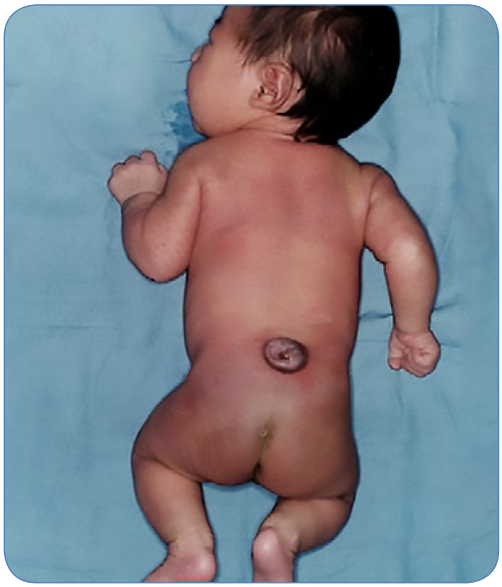
Protrusion of meninges and/or spinal cord through an opening in the thoracic region of the vertebral column, associated with hydrocephalus. It may be open, with or without membrane covering; or closed, covered by normal-appearing skin. Hydrocephalus is present as a result of obstruction to the flow of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

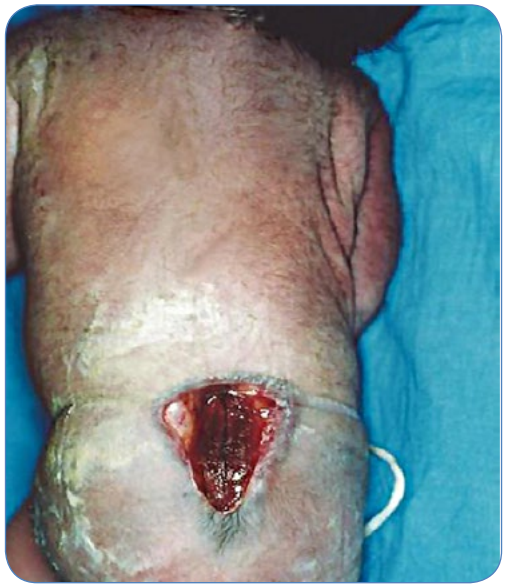
Thoracic spina bifida without hydrocephalus (Q05.6)

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.

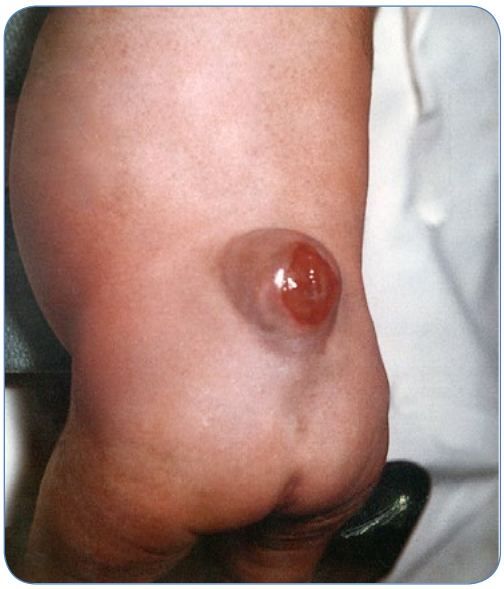
Lumbar spina bífida

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.





Photographs source: courtesy of CDC-Beijing Medical University collaborative project.



Photograph source: courtesy of Idalina Montes, MD and Rafael Longo, MD, FACS, Puerto Rico.

Lumbar spina bifida with hydrocephalus (Q05.2)

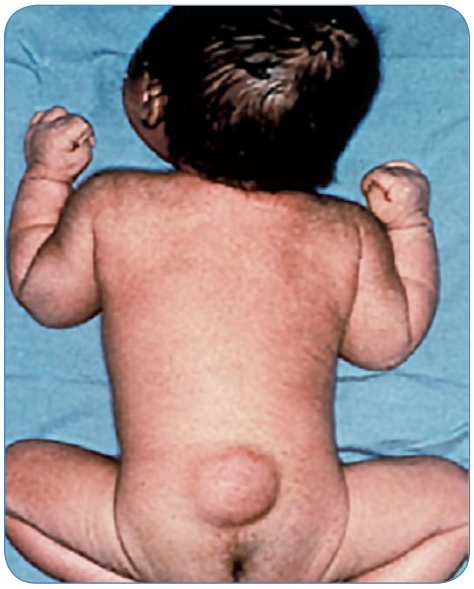
Protrusion of meninges and/or spinal cord through an opening in the lumbar region of the vertebral column, associated with hydrocephalus. It may be open, with or without membrane covering; or closed, covered by normal-appearing skin. Hydrocephalus is present as a result of obstruction to the flow of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

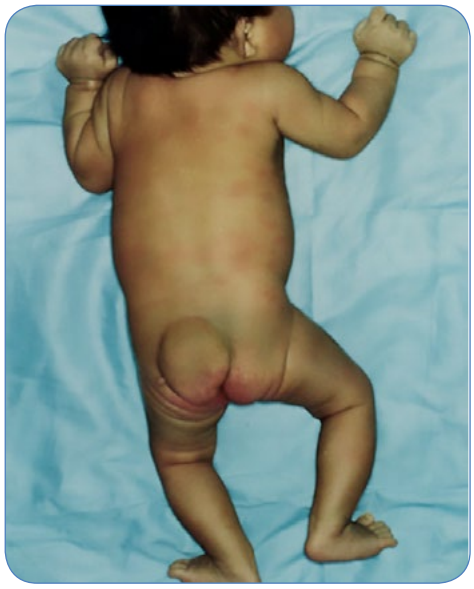
Lumbar spina bifida without hydrocephalus (Q05.7)

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.

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.





Photographs source: courtesy of CDC-Beijing Medical University collaborative project.

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.

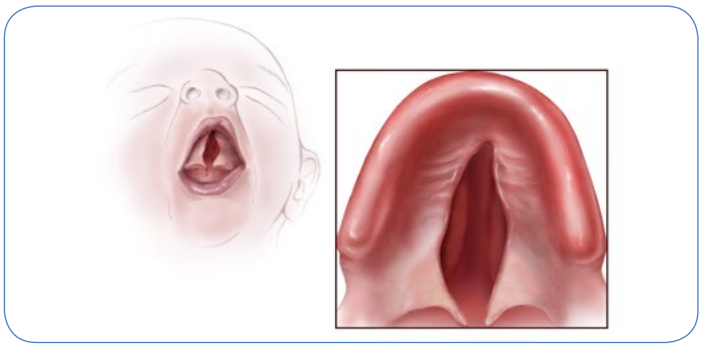
Sacral spina bifida without hydrocephalus (Q05.8)

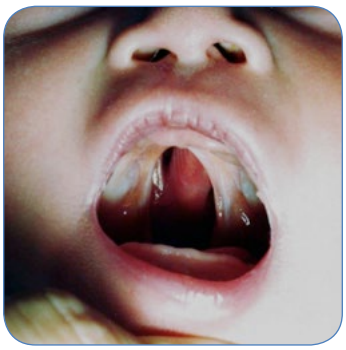
Protrusion of meninges and/or spinal cord through an opening in the sacral region of the vertebral column. It may be open, with or without membrane covering; or closed, covered by normal-appearing skin.

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.

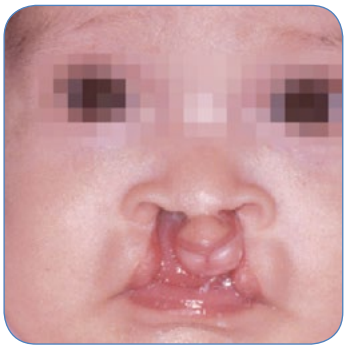




Photograph source: courtesy of CDC-Beijing Medical University collaborative project.

Cleft lip, bilateral (Q36.0)

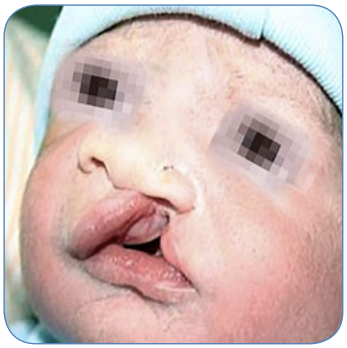
Partial or complete bilateral fissure of the upper lip that may be associated with a cleft of the gum.

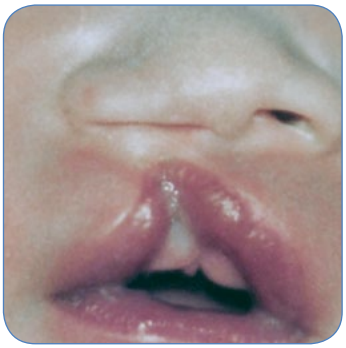


Photograph source: courtesy of Pedro Santiago, DMD and Miguel Yáñez, MD, FACS, USA.

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.

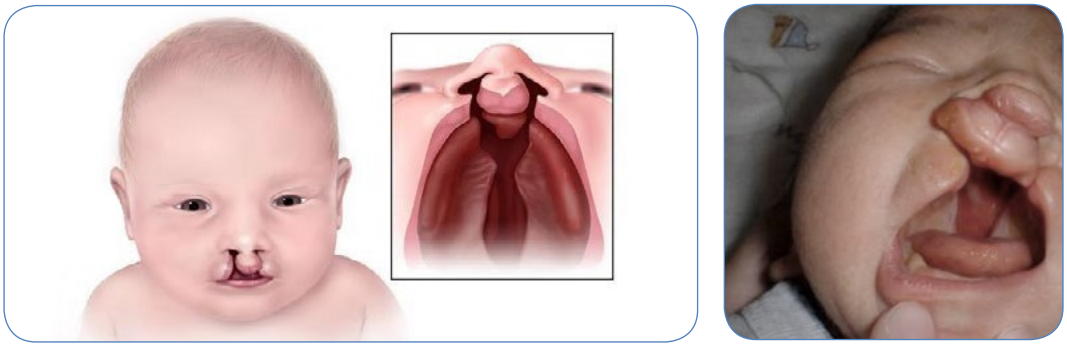




Photographs sources: courtesy of Jaime Frías, MD, USA (middle); CDC-Beijing Medical University collaborative project (right).

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

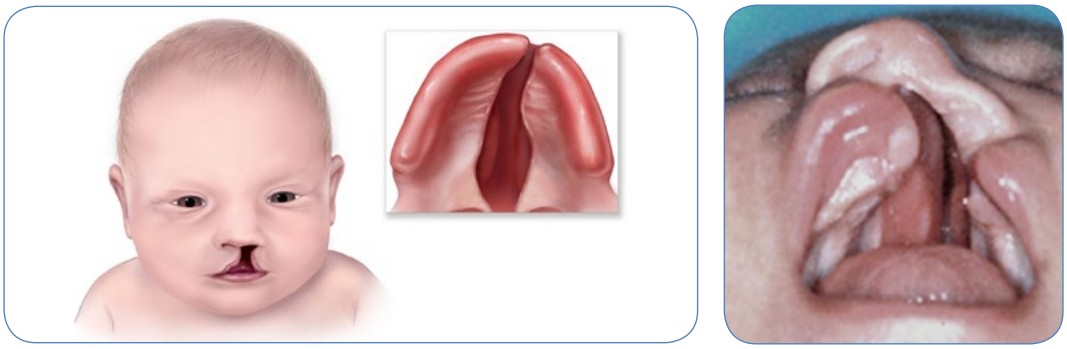
Partial or complete bilateral fissure of the upper lip, associated with a fissure of the palate.



Photograph source: courtesy of Estudio Colaborativo Latino Americano de Malformaciones Congénitas (ECLAMC).

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.



Photograph source: courtesy of Pedro Santiago, DMD and Miguel Yáñez, MD, FACS, USA.

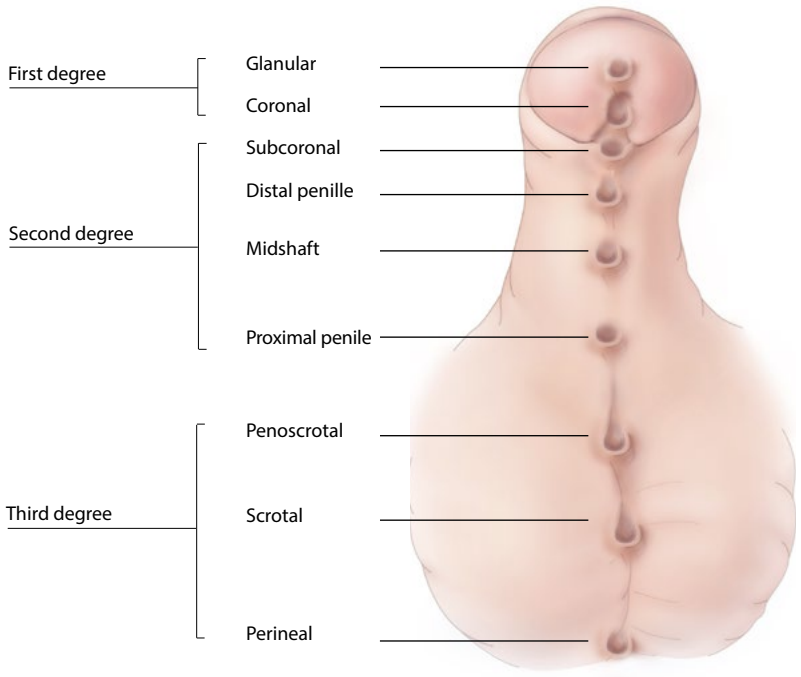
Congenital malformations of genital organs

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 (subcoronal 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**)

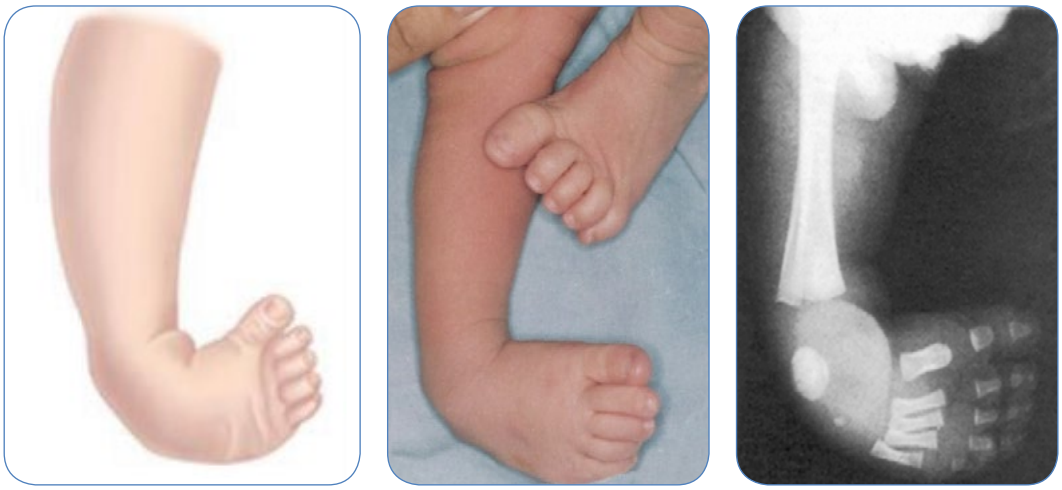
Q54.9 Hypospadias, unspecified

Note: illustration indicates all possible locations for the malformation, but one case will not have all.

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).

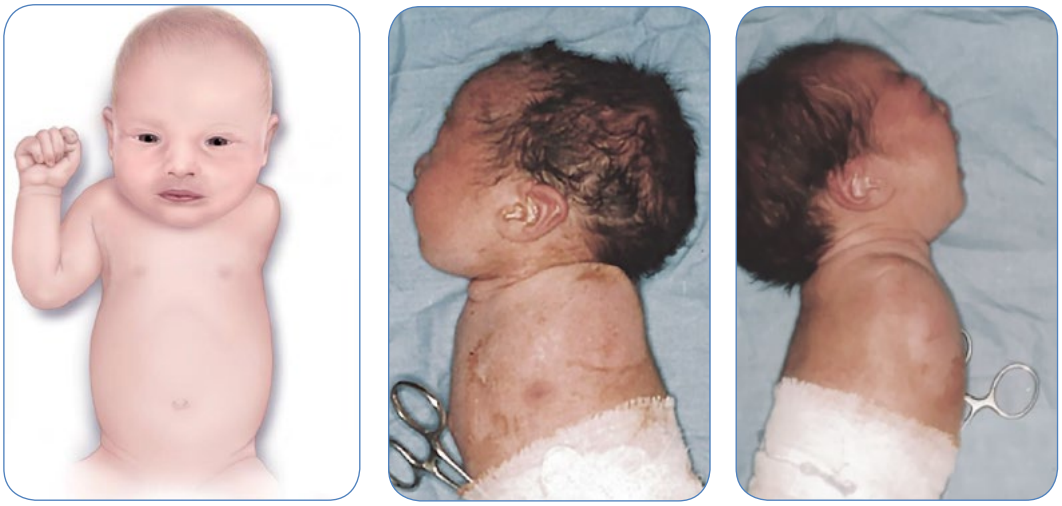


Photograph and X-ray source: courtesy of Idalina Montes, MD and Rafael Longo, MD, FACS, Puerto Rico.

Reduction defects of upper and lower limbs

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

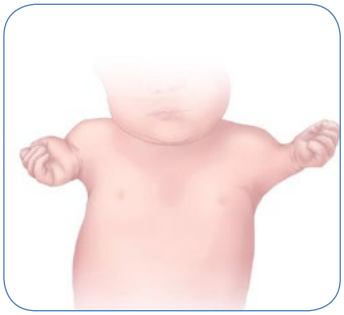
Complete absence of one or both upper limbs.

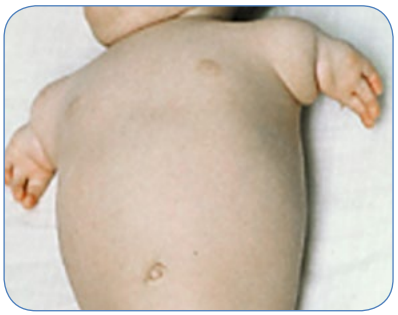


Photographs source: courtesy of CDC-Beijing Medical University collaborative project.

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.

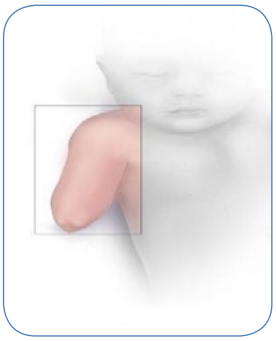


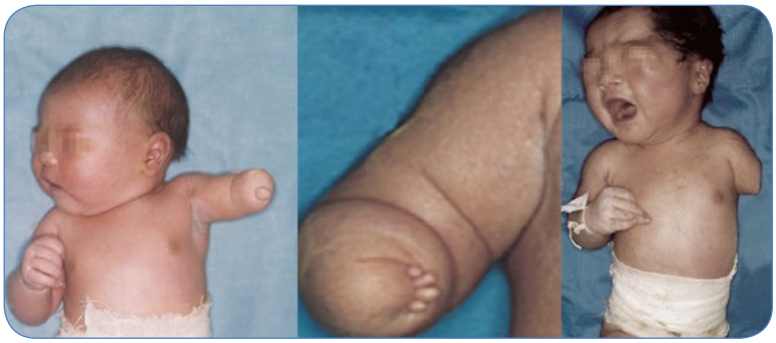


Photograph source: courtesy of Jaime Frías, MD, USA.

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

Complete or partial absence of both the forearm and hand.

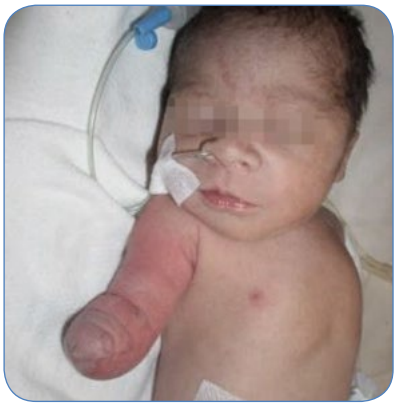




Photographs source: CDC-Beijing Medical University collaborative project.

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

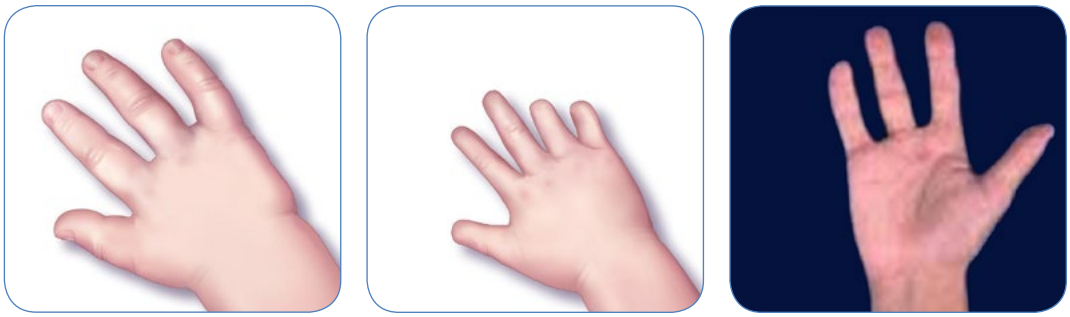
Complete or partial absence of the hand and finger(s).



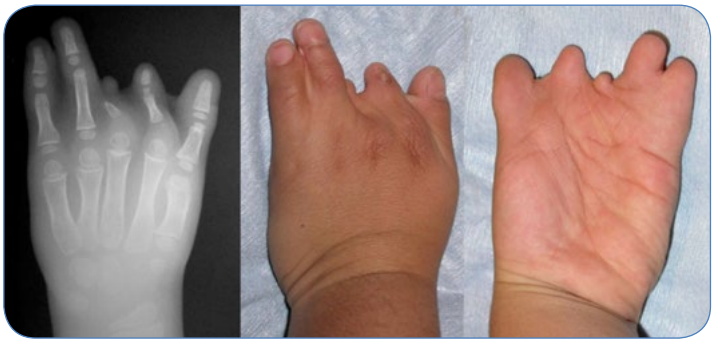
Photograph source: courtesy of Estudio Colaborativo Latino Americano de Malformaciones Congénitas (ECLAMC).

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

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



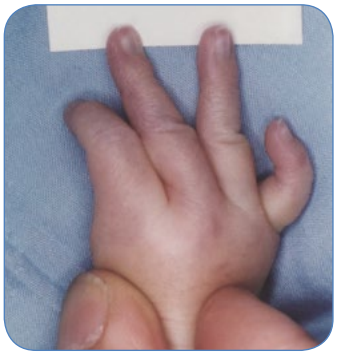
Photograph source: courtesy of John Wiley and Sons ©2009. Biesecker LG et al. Am. J. Med. Genet. A. 2009;149A:93–127.



Photographs and X-ray source: courtesy of Dr E Gene Deune, MD; Associate Professor, Johns Hopkins Department of Orthopedic Surgery, Division of Hand Surgery, Baltimore, MD, USA.

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

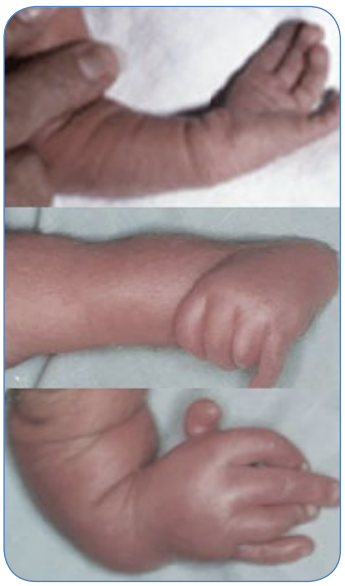
Complete or partial absence or hypoplasia of the thumb.



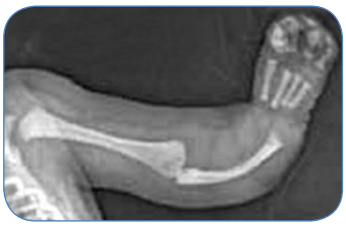
Photograph source: CDC-Beijing Medical University collaborative project.

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.



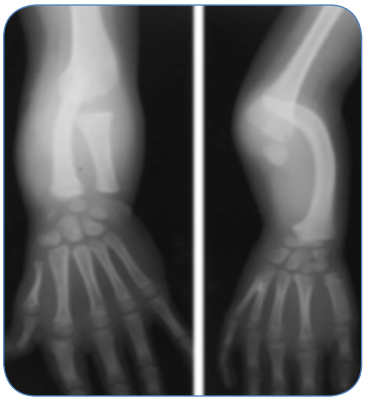
Photographs source: courtesy of CDC-Beijing Medical University collaborative project.



X-ray source: courtesy of John Wiley and Sons ©2011. Umaña LA et al. Am. J. Med. Genet. A. 2011;155A:3071–4.

Longitudinal reduction defect of ulna (Q71.5)

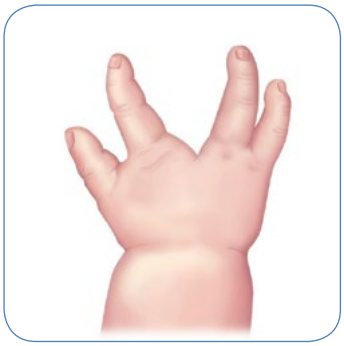
Complete or partial absence of the ulna.

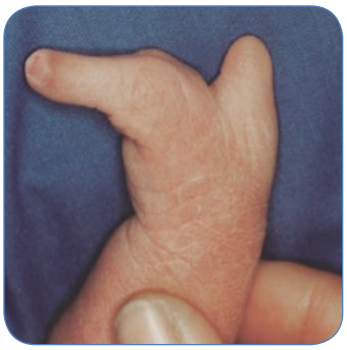


X-ray source: courtesy of John Wiley and Sons ©1997. Kumar D et al. Am. J. Med. Genet. A. 1997;70A:107–13.

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.

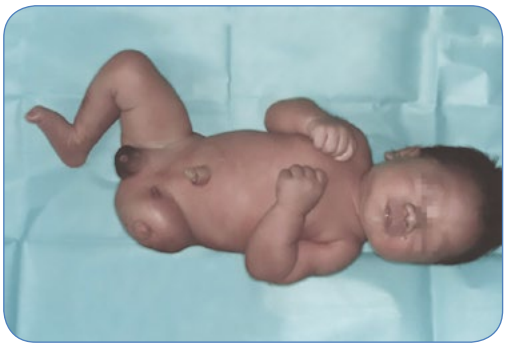




Photographs source: CDC-Beijing Medical University collaborative project.

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

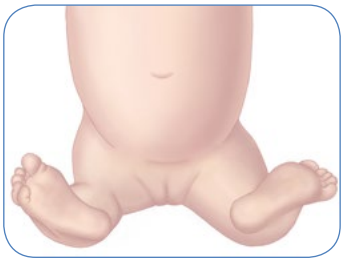
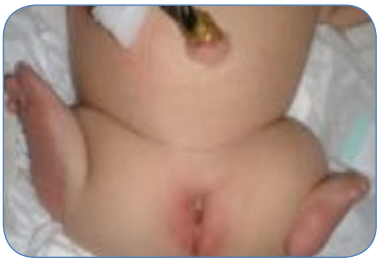
Complete absence of one or both lower limb(s).

Photograph source: CDC-Beijing Medical University collaborative project.

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

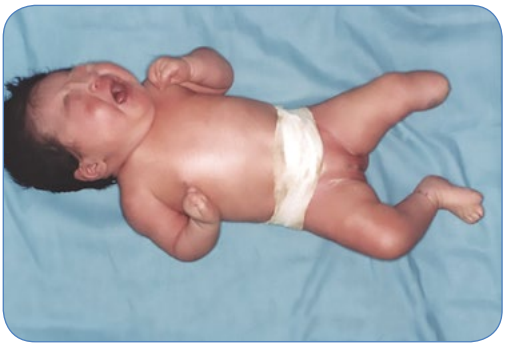
Complete or partial absence of the thigh and lower leg but with the foot present.

Photograph source: courtesy of Estudio Colaborativo Latino Americano de Malformaciones Congénitas (ECLAMC).

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

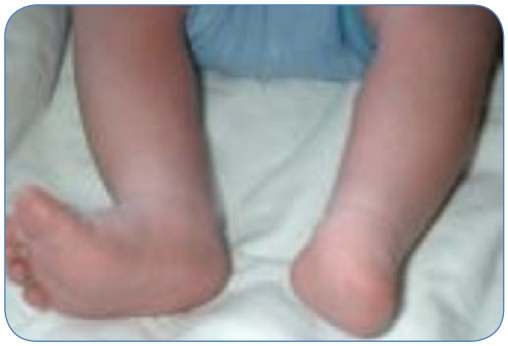
Complete or partial absence of both the lower leg and foot.



Photograph source: CDC-Beijing Medical University collaborative project.

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

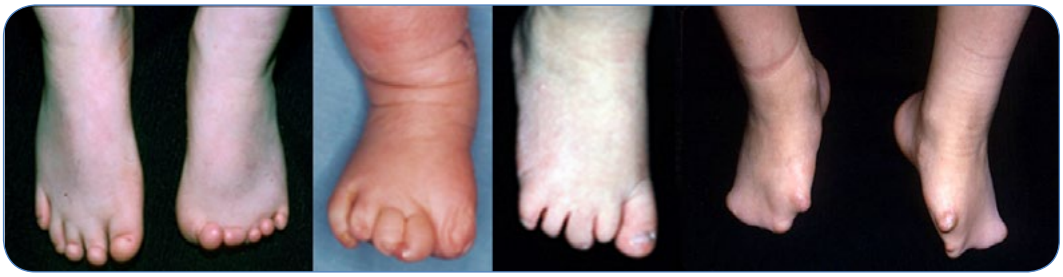
Complete or partial absence of the foot and toe(s).



Photograph source: courtesy of Estudio Colaborativo Latino Americano de Malformaciones Congénitas (ECLAMC).

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

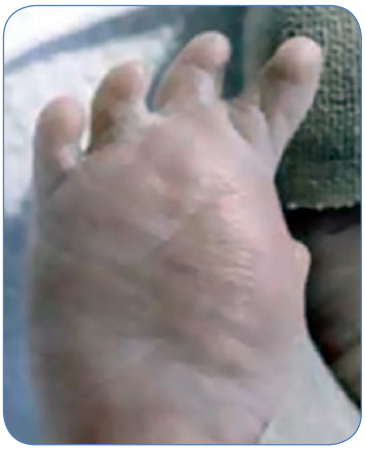
Complete absence or hypoplasia of toe(s) with the remainder of the foot intact.



Photographs source: courtesy of John Wiley and Sons ©2009. Biesecker LG et al. Am. J. Med. Genet. A. 2009;149A:93–127.

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

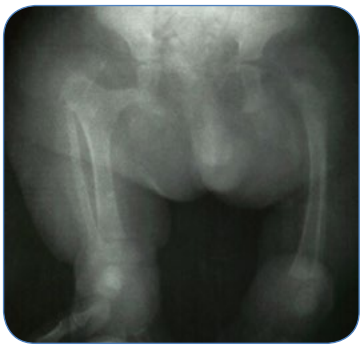
Complete absence or hypoplasia of the first (great) toe with other digits present.



Photograph source: courtesy of John Wiley and Sons ©2009. Biesecker LG et al. Am. J. Med. Genet. A. 2009;149A:93–127.

Longitudinal reduction defect of femur (Q72.4)

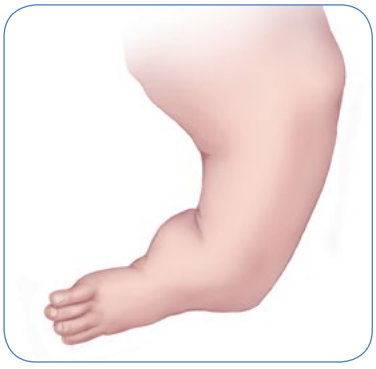
Complete or partial absence of the femur.

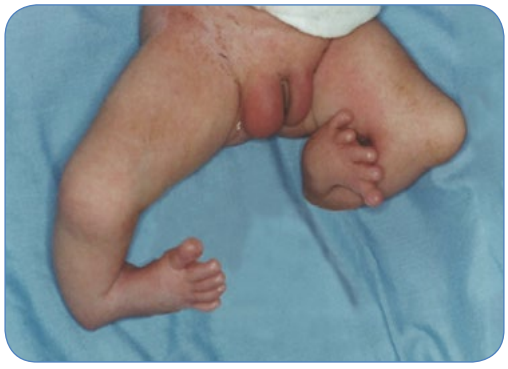


X-ray source: courtesy of Jaime Frías, MD, USA.

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.

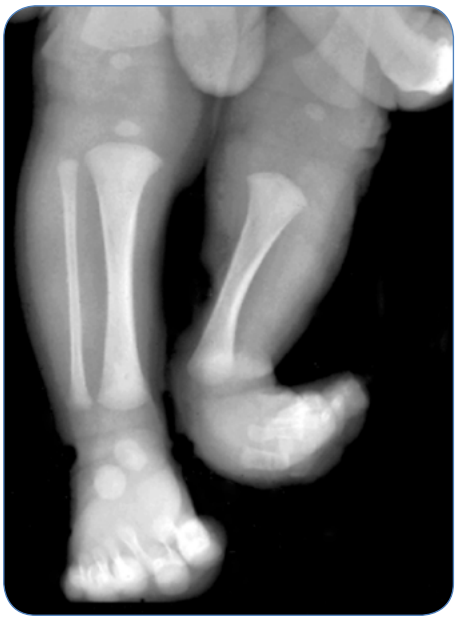




Photograph source: CDC-Beijing Medical University collaborative project.

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

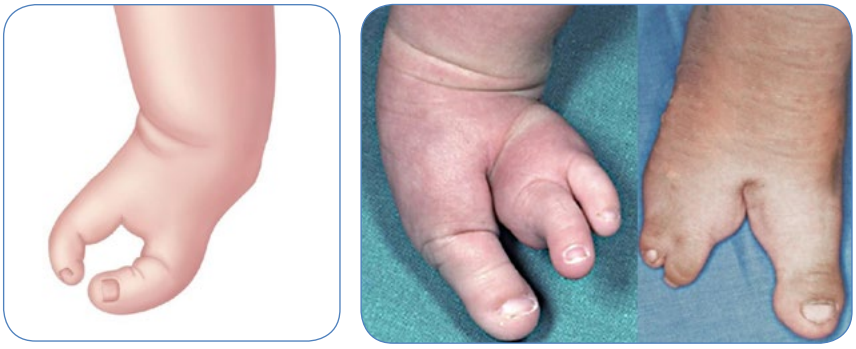
Complete or partial absence of the fibula.



X-ray source: image reprinted with permission from Holmstrom MC et al. Medscape 2013 (http://emedicine.medscape.com/article/1251558-overview).

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

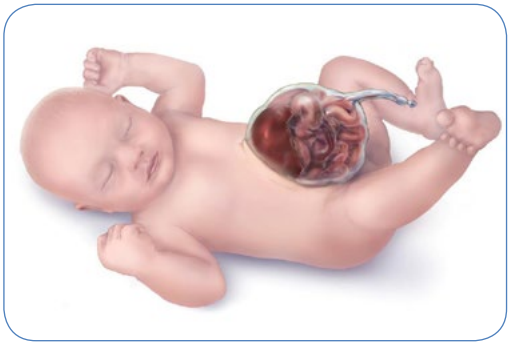
Complete or partial absence of central toes and metatarsals. The term ectrodactyly, used by some, should be discouraged.

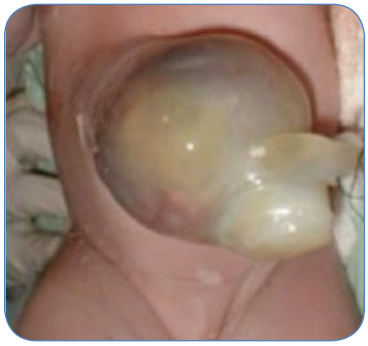


Photographs source: CDC-Beijing Medical University collaborative project.

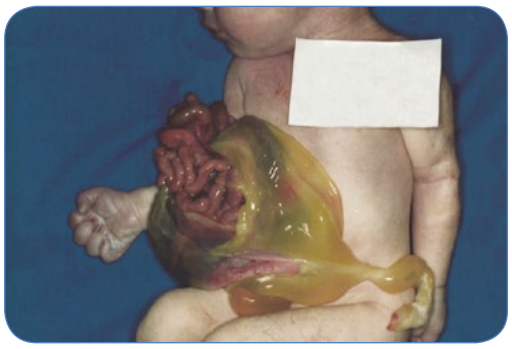
Exomphalos/omphalocele (Q79.2)

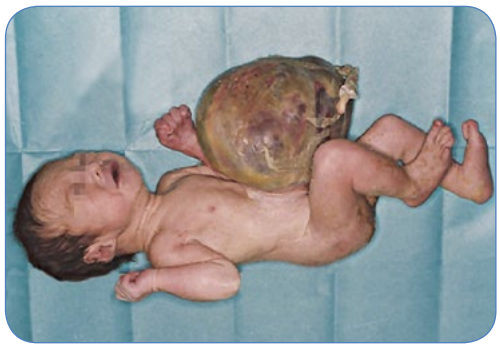
Congenital anomaly of the anterior abdominal wall, in which the abdominal contents (gut, but at times also other abdominal organs) are herniated in the midline through an enlarged umbilical ring. The umbilical cord is inserted in the distal part of the membrane covering the anomaly. The herniated organs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





Photograph source: courtesy of Estudio Colaborativo Latino Americano de Malformaciones Congénitas (ECLAMC).



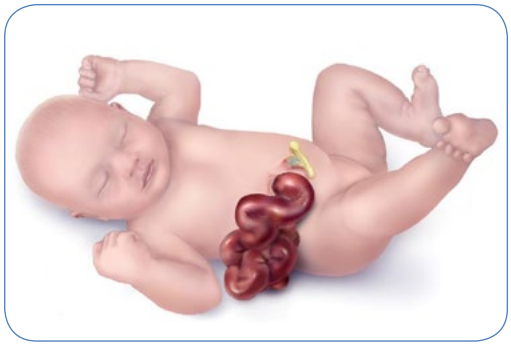


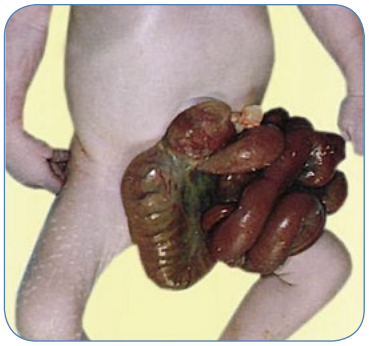
Photographs source: CDC-Beijing Medical University collaborative project.

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.





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