

Chapter XVII, (Q00-Q99) (version 23 June 2008)

Congenital malformations, deformations and chromosomal abnormalities

Excludes: inborn errors of metabolism (E70-E90)

This chapter contains the following blocks:

Q00-Q07	Congenital malformations of the nervous system
Q10-Q18	Congenital malformations of eye, ear, face and neck
Q20-Q28	Congenital malformations of the circulatory system
Q30-Q34	Congenital malformations of the respiratory system
Q35-Q37	Cleft lip and palate
Q38-Q45	Other congenital malformations of the digestive system
Q50-Q56	Congenital malformations of genital organs
Q60-Q64	Congenital malformations of the urinary system
Q65-Q79	Congenital malformations and deformations of the musculoskeletal system
Q80-Q89	Other congenital malformations
Q90-Q99	Chromosomal abnormalities, not elsewhere classified

Q00-Q07 Congenital malformations of the nervous system

Q00 Anencephaly and similar malformations

Q00.00 Anencephaly, NOS
Acephaly
Acrania
Amyelencephaly
Excludes: hydranencephaly (Q04.35)

Q00.01 Incomplete anencephaly
Hemianencephaly
Hemicephaly

Q00.1 Craniorachischisis
Rachischisis:
 . craniospinal
 . complete
 . total

Q00.2 Iniencephaly
Q00.20 Iniencephaly, open
Q00.21 Iniencephaly, closed

Q01

Includes:

Encephalocele

encephalomyelocele
hydroencephalocele
hydromeningocele, cranial
meningocele, cerebral
meningoencephalocele

Note:

cranial hydromeningocele and cerebral meningocele are not considered to be encephaloceles as they do not contain brain tissue but have been included here in ICD-10

Excludes: Meckel-Gruber syndrome (Q61.9)

Q01.0 Frontal encephalocele
Q01.1 Nasofrontal encephalocele
Q01.2 Occipital encephalocele

Q01.8 Encephalocele of other sites
Q01.80 Parietal encephalocele
Q01.81 Orbital encephalocele
Q01.82 Nasal encephalocele
Q01.83 Nasopharyngeal encephalocele

Q01.9 Encephalocele, unspecified

Q02

Microcephaly

Hydromicrocephaly
Micrencephalon

Excludes:

Meckel-Gruber syndrome (Q61.9)
microcephaly due to:
 . congenital infection (P35-P37)
 . exposure to ionising radiation (Q86.85)

Q03 Congenital hydrocephalus*Includes:* hydrocephalus in newborn*Excludes:* Arnold-Chiari syndrome (Q07.0)
hydrocephalus: . acquired (G91.-)
. due to congenital toxoplasmosis (P37.1)
. with spina bifida (Q05.0-Q05.4)Q03.0 Malformations of aqueduct of Sylvius
Aqueduct of Sylvius: . anomaly
. obstruction, congenital
. stenosisQ03.1 Atresia of foramina of Magendie and Luschka
Dandy-Walker syndrome

Q03.8 Other congenital hydrocephalus

Q03.80 Clover leaf skull
Kleeblattschaedel deformity syndrome

Q03.9 Congenital hydrocephalus, unspecified

Q04 Other congenital malformations of brain*Excludes:* cyclopia (Q87.03)
macrocephaly (Q75.3)

Q04.0 Congenital malformations of corpus callosum

Q04.00 Agenesis of corpus callosum

Q04.1 Arhinencephaly

Q04.2 Holoprosencephaly

Q04.3 Other reduction deformities of brain

Absence }

Agenesis }

Aplasia } of part of brain

Hypoplasia }

Excludes: congenital malformations of corpus callosum (Q04.0)

Q04.30 Reduction anomalies of cerebrum

Q04.31 Reduction anomalies of hypothalamus

Q04.32 Reduction anomalies of cerebellum

Q04.33 Agyria or lissencephaly

Q04.34 Microgyria or pachygyria

Polygyria

Micropolygyria

Q04.35 Hydranencephaly

Q04.4 Septo-optic dysplasia

Q04.5 Megalencephaly

Q04.6 Congenital cerebral cysts

Porencephaly

Schizencephaly

Excludes: acquired porencephalic cysts (G93.0)

Q04.60 Multiple congenital cerebral cysts

Q04.61 Single congenital cerebral cyst

Q04.8 Other specified congenital malformations of brain

Macrogyria

Walnut brain

Congenital haematocephalus

Congenital malformation of cerebral meninges

Q04.9 Congenital malformation of brain, unspecified
 Congenital: . anomaly }
 . deformity }
 . disease or lesion } NOS of brain
 . multiple anomalies }

Q05 Spina bifida

Includes: hydromeningocele (spinal)
 meningocele (spinal)
 meningomyelocele
 myelocele
 myelomeningocele
 spinal rachischisis
 spina bifida (aperta)(cystica)
 syringomyelocele

Excludes: Arnold-Chiari syndrome (Q07.0)
 spina bifida occulta (Q76.0)
 rachischisis (Q00.1): . cranial
 . craniospinal

Note: For Spina bifida Q05.0-Q05.8 the following fifth-character subdivision can be used if desired-

- 1 open, aperta, not covered with skin or membrane
- 2 closed, cystica, covered with skin or membrane
- 9 if not known whether lesion is open or closed

Q05.0 Cervical spina bifida with hydrocephalus

Q05.1 Thoracic spina bifida with hydrocephalus
 Spina bifida: . dorsal }
 . thoracolumbar } with hydrocephalus
 . dorsolumbar }

Q05.2 Lumbar spina bifida with hydrocephalus
 Lumbosacral spina bifida with hydrocephalus

Q05.3 Sacral spina bifida with hydrocephalus

Q05.4 Unspecified spina bifida with hydrocephalus
 Site unspecified

Q05.5 Cervical spina bifida without hydrocephalus

Q05.6 Thoracic spina bifida without hydrocephalus
 Spina bifida: . dorsal NOS
 . thoracolumbar NOS
 . dorsolumbar NOS

Q05.7 Lumbar spina bifida without hydrocephalus
 Lumbosacral spina bifida NOS

Q05.8 Sacral spina bifida without hydrocephalus

Q05.9 Spina bifida, unspecified

Q06 Other congenital malformations of spinal cord

Excludes: syringomyelia and syringobulbia (G95.0)

Q06.0 Amyelia

Q06.1 Hypoplasia and dysplasia of spinal cord
 Atelomyelia
 Myelatelasia
 Myelodysplasia of spinal cord

Q06.2 Diastematomyelia

Q06.3 Other congenital cauda equina malformations

Q06.4 Hydromyelia
 Hydrorachis

Q06.8 Other specified congenital malformations of spinal cord

Q06.9 Congenital malformations of spinal cord, unspecified

Congenital: . anomaly }
 . deformity } NOS of spinal cord
 . disease or lesion } or meninges

Q07 Other congenital malformations of nervous system

Excludes: familial dysautonomia [Riley-Day] (G90.1)
 neurofibromatosis (nonmalignant) (Q85.0)

Q07.0 Arnold-Chiari syndrome

Q07.8	Other specified congenital malformations of nervous system Agenesis of nerve, NOS Cayler syndrome Congenital facial diplegia Displacement of brachial plexus Nuclear agenesis <i>Excludes:</i> Moebius syndrome (Q87.06) Duane syndrome (H50.8)
Q07.80	Jaw-winking syndrome Marcus Gunn's syndrome
Q07.81	Optic nerve hypoplasia Congenital optic atrophy
Q07.82	Crocodile tears
Q07.9	Congenital malformations of nervous system, unspecified Congenital malformation of meninges, unspecified Congenital: . anomaly } . deformity } NOS of nervous . disease or lesion } system

Q10-Q18 **Congenital malformations of eye, ear, face and neck**

Excludes: cleft lip and cleft palate (Q35-37)
congenital malformations of:
 . cervical spine (Q05.0, Q05.5, Q67.5, Q76.0-Q76.4)
 . larynx (Q31.-)
 . lip NEC (Q38.0)
 . nose (Q30.-)
 . parathyroid gland (Q89.2)
 . thyroid gland (Q89.2)
 retinoblastoma (C69.2)

Q10	Congenital malformations of eyelid, lacrimal apparatus and orbit
<i>Excludes:</i>	cryptophthalmos: . NOS (Q11.2) . syndrome (Q87.02) Goldenhar syndrome [oculo-auriculo-vertebral syndrome] (Q87.04)
Q10.0	Congenital ptosis Blepharophimosis-ptosis syndrome
Q10.1	Congenital ectropion
Q10.2	Congenital entropion
Q10.3	Other congenital malformations of eyelid Ablepharon (absence of eyelids) Absence or agenesis of: . cilia (eyelashes) . eyelid Accessory: . eyelid . eye muscle Blepharophimosis, congenital [fused eyelids] Congenital symblepharon Coloboma of eyelid Mongoloid slant (of palpebral fissure) Antimongoloid slant (of palpebral fissure) Congenital malformation of eyelid NOS
Q10.4	Absence and agenesis of lacrimal apparatus Absence of punctum lacrimale
Q10.5	Congenital stenosis and stricture of lacrimal duct
Q10.6	Other congenital malformations of lacrimal apparatus Congenital malformations of lacrimal apparatus NOS
Q10.7	Congenital malformations of orbit
Q11	Anophthalmos, microphthalmos and macropthalmos
Q11.0	Cystic eyeball
Q11.1	Other anophthalmos Agenesis } Aplasia } of eye <i>Excludes:</i> cryptophthalmos syndrome (Q87.02)

Q11.2	Microphthalmos Cryptophthalmos NOS Dysplasia of eye Fraser syndrome Hypoplasia of eye Lenz' microphthalmus syndrome Rudimentary eye <i>Excludes:</i> cryptophthalmos syndrome (Q87.02)	Q13.3	Congenital corneal opacity
		Q13.4	Other congenital corneal malformations Congenital malformation of cornea NOS Microcornea Peter's anomaly
		Q13.5	Blue sclera
Q11.3	Macrophthalmos <i>Excludes:</i> macrophthalmos in congenital glaucoma (Q15.0)	Q13.8	Other congenital malformations of anterior segment of eye Rieger's anomaly Iridogoniodysgenesis with somatic anomalies
		Q13.9	Congenital malformations of anterior segment of eye, unspecified
Q12	Congenital lens malformations	Q14	Congenital malformations of posterior segment of eye
Q12.0	Congenital cataract	Q14.0	Congenital malformation of vitreous humour Congenital vitreous opacity
Q12.1	Congenital displaced lens		
Q12.2	Coloboma of lens	Q14.1	Congenital malformation of retina Congenital retinal aneurysm Coloboma of retina
Q12.3	Congenital aphakia		
Q12.4	Spherophakia		
		Q14.10	Congenital retinoschisis
Q12.8	Other congenital lens malformations		
Q12.80	Microphakia		
		Q14.2	Congenital malformation of optic disc Coloboma of optic disc
Q12.9	Congenital lens malformation, unspecified	Q14.3	Congenital malformation of choroid
Q13	Congenital malformations of anterior segment of eye		
Q13.0	Coloboma of iris Coloboma NOS	Q14.8	Other congenital malformations of posterior segment of eye Coloboma of the fundus
Q13.1	Absence of iris Aniridia See also nephroblastoma [Wilms' tumour] (C64)	Q14.9	Congenital malformation of posterior segment of eye, unspecified
Q13.2	Other congenital malformations of iris Anisocoria, congenital Atresia of pupil Congenital malformation of iris NOS Corectopia Polycoria <i>Excludes:</i> ectopic pupil (H21.5)		

Q15 Other congenital malformations of eye

Excludes: congenital nystagmus (H55)
ocular albinism (E70.3)
retinitis pigmentosa (H35.5)

Q15.0 Congenital glaucoma
Buphthalmos
Glaucoma of newborn Hydrophthalmos
Macrophthalmos in congenital glaucoma

Q15.00 Congenital keratoglobus
Enlarged cornea
Megalocornea

Q15.8 Other specified congenital malformations of eye

Q15.9 Congenital malformation of eye, unspecified
Congenital: . anomaly }
. deformity } NOS of eye

Q16 Congenital malformations of ear causing hearing impairment

Excludes: congenital deafness (H90.-)

Q16.0 Congenital absence of (ear) auricle
Anotia
Congenital absence of ear lobe

Q16.1 Congenital absence, atresia and stricture of auditory canal (external)
Atresia, stenosis or stricture of osseous meatus

Q16.2 Absence of Eustachian tube

Q16.3 Congenital malformation of ear ossicles
Fusion of ear ossicles

Q16.4 Other congenital malformations of middle ear
Congenital malformations of middle ear NOS

Q16.5 Congenital malformation of inner ear
Anomaly of: . membranous labyrinth
. organ of Corti

Q16.9 Congenital malformation of ear causing impairment of hearing, unspecified
Congenital absence of ear NOS

Q17 Other congenital malformations of ear

Excludes: preauricular sinus (Q18.1)

Q17.0 Accessory auricle
Accessory tragus
Polyotia
Preauricular appendage or tag
Supernumary: . ear
. lobule

Excludes: Goldenhar syndrome
[oculo-auriculo-vertebral syndrome] (Q87.04)

Q17.1 Macrotia

Q17.2 Microtia

Q17.3 Other misshapen ear
Pointed ear
Vulcan ear
Simple ear

Q17.4 Misplaced ear
Low set ears
Excludes: cervical auricle (Q18.2)

Q17.5 Prominent ear
Bat ear

Q17.8 Other specified congenital malformations of ear
Darwin's tubercle
Branchio-oro-renal syndrome
Melnick-Fraser syndrome

Q17.9 Congenital malformation of ear, unspecified
Congenital anomaly of ear NOS

Q18 Other congenital malformations of face and neck**Excludes:**

cleft lip and cleft palate (Q35-37)
 conditions classified to Q67.0-Q67.4
 congenital malformations of skull and face bones (Q75.-)
 cyclopia (Q87.03)
 dentofacial anomalies [including malocclusion] (K07.-)
 malformation syndromes affecting facial appearance (Q87.0-)
 persistent thyroglossal duct (Q89.2)

Q18.0 Sinus, fistula and cyst of branchial cleft

Branchial vestige

Q18.1 Preauricular sinus and cyst

Fistula : . of auricle, congenital
 . cervicoaural

Q18.2 Other branchial cleft malformations

Branchial cleft malformations NOS

Cervical auricle

Otocephaly

Q18.3 Webbing of neck

Pterygium colli

Q18.4 Macrostomia

Q18.5 Microstomia

Q18.6 Macrocheilia

Hypertrophy of lip, congenital

Q18.7 Microcheilia

Q18.8 Other specified congenital malformations of face and neck

Medial: . cyst }
 . fistula } of face and neck
 . sinus }

Q18.80 Synophrys

Q18.9 Congenital malformation of face and neck, unspecified

Congenital anomaly NOS of face and neck

Q20-Q28 Congenital malformations of the circulatory system**Q20****Congenital malformations of cardiac chambers and connections****Excludes:**

dextrocardia with situs inversus (Q89.3)
 mirror image atrial arrangement with situs inversus (Q89.3)

Q20.0 Common arterial trunk

Persistent truncus arteriosus

Q20.1 Double outlet right ventricle

Taussig-Bing syndrome

Q20.2 Double outlet left ventricle

Q20.3 Discordant ventriculoarterial connection

Dextrotransposition of aorta

Transposition of great vessels (complete)

Q20.4 Double inlet ventricle

Common ventricle

Cor triloculare biatriatum

Single ventricle

Q20.5 Discordant atrioventricular connection

Corrected transposition

Levotransposition

Ventricular inversion

Q20.6 Isomerism of atrial appendages

Isomerism of atrial appendages with asplenia or polysplenia

Ivemark syndrome

Q20.8 Other congenital malformations of cardiac chambers and connections

Cor biloculare

Q20.9 Congenital malformation of cardiac chambers and connections, unspecified

Q21**Congenital malformations of cardiac septa****Excludes:**

acquired cardiac septal defect (I51.0)

Q21.0 Ventricular septal defect

Roger's disease [Maladie de Roger]

Small VSD with no significant haemodynamic effects

Q21.1	Atrial septal defect, ASD
Q21.10	Ostium secundum atrial septal defect (type II)
Q21.11	Patent or persistent foramen ovale
Q21.12	Sinus venosus defect
Q21.13	Coronary sinus defect
Q21.14	Lutembacher's syndrome (ASD plus mitral stenosis)
Q21.15	Common atrium
	Cor triloculare biventriculare
Q21.18	Other specified atrial septal defect
	<i>Excludes:</i> ostium primum atrial septal defect (type I) Q21.20
Q21.2	Atrioventricular septal defect
Q21.20	Ostium primum atrial septal defect (type I)
Q21.21	Common atrioventricular canal
Q21.28	Other specified atrioventricular septal defect
	Endocardial cushion defect NOS
Q21.3	Tetralogy of Fallot
	Ventricular septal defect with pulmonary stenosis or atresia, dextroposition of aorta and hypertrophy of right ventricle.
Q21.4	Aortopulmonary septal defect
	Aortic septal defect
	Aortopulmonary window
Q21.8	Other congenital malformations of cardiac septa
Q21.80	Left ventricle to right atrial communication
	Gerbode defect
Q21.81	Eisenmenger's syndrome
Q21.82	Pentalogy of Fallot
	Fallot's tetralogy plus atrial septal defect
Q21.9	Congenital malformation of cardiac septum, unspecified
	Septal heart defect, NOS

Q22	Congenital malformations of pulmonary and tricuspid valves
Q22.0	Pulmonary valve atresia
Q22.1	Congenital pulmonary valve stenosis
Q22.2	Congenital pulmonary valve insufficiency
	Congenital pulmonary valve regurgitation
Q22.3	Other congenital malformations of pulmonary valve
	Congenital malformation of pulmonary valve NOS
Q22.4	Congenital tricuspid stenosis
	Tricuspid atresia
Q22.5	Ebstein's anomaly
Q22.6	Hypoplastic right heart syndrome
Q22.8	Other congenital malformations of tricuspid valve
Q22.9	Congenital malformation of tricuspid valve, unspecified
Q23	Congenital malformations of aortic and mitral valves
Q23.0	Congenital stenosis of aortic valve
	Congenital aortic: . atresia
	. stenosis
	<i>Excludes:</i> congenital subaortic stenosis (Q24.4)
	that in hypoplastic left heart syndrome (Q23.4)
Q23.1	Congenital insufficiency of aortic valve
	Congenital aortic insufficiency
Q23.10	Bicuspid aortic valve
Q23.2	Congenital mitral stenosis
	Congenital mitral atresia
Q23.3	Congenital mitral insufficiency
Q23.4	Hypoplastic left heart syndrome
	Atresia, or marked hypoplasia of aortic orifice or valve, with hypoplasia of ascending aorta and defective development of left ventricle (with mitral valve stenosis or atresia)
Q23.8	Other congenital malformations of aortic and mitral valves
Q23.9	Congenital malformation of aortic and mitral valves, unspecified

Q24 Other congenital malformations of heart*Excludes:*

Q24.0

Dextrocardia

Excludes:

dextrocardia with situs inversus (Q89.3)
 isomerism of atrial appendages (with
 asplenia or polysplenia) (Q20.6)
 mirror image atrial arrangement with situs
 inversus (Q89.3)

Q24.1

Laevocardia

Q24.2

Cor triatriatum

Q24.3

Pulmonary infundibular stenosis

Q24.4

Congenital subaortic stenosis

Q24.5

Malformation of coronary vessels
 Congenital coronary (artery) aneurysm

Q24.6

Congenital heart block

Q24.8

Other specified congenital malformations of the heart

Congenital malformation of: . myocardium
 . pericardium

Malposition of heart

Uhl's disease

Congenital cardiomegaly

Fallot's trilogy

Ectopia cordis

Q24.80

Congenital diverticulum of left ventricle

Q24.9

Congenital malformations of the heart, unspecified

Congenital: . anomaly
 . disease NOS of heart

Q25 Congenital malformations of great arteries

Q25.0

Patent ductus arteriosus

PDA

Patent ductus Botallo

Persistent ductus arteriosus

Q25.1

Coarctation of aorta

Q25.10

Preductal coarctation of aorta

Q25.11

Postductal coarctation of aorta

Q25.19

Coarctation of aorta unspecified

Q25.2

Atresia of aorta

Interrupted aortic arch

Q25.3

Stenosis of aorta

Supravalvular aortic stenosis

Excludes: congenital aortic stenosis (valvular) (Q23.0)

Q25.4

Other congenital malformations of aorta

Absence }

Aplasia } of aorta

Persistent convolutions of aortic arch

Excludes: hypoplasia of aorta in hypoplastic left heart
 syndrome (Q23.4)

Q25.40

Hypoplasia of aorta

Tubular hypoplasia of aorta

Q25.41

Persistent right aortic arch

Q25.42

Overriding aorta

Q25.43

Aneurysm of sinus of Valsalva (ruptured)

Q25.44

Double aortic arch

Vascular ring due to double aortic arch

Q25.45

Congenital aneurysm of aorta

Congenital dilatation of aorta

Q25.5

Atresia of pulmonary artery

Q25.6

Stenosis of pulmonary artery

Q25.7

Other congenital malformations of pulmonary artery

Agenesis }

Anomaly } of pulmonary artery

Hypoplasia }

Q25.70

Pulmonary arteriovenous aneurysm

Q25.71

Aberrant pulmonary artery

Q25.72

Congenital aneurysm of pulmonary artery

Congenital dilatation of pulmonary artery

Q25.8	Other congenital malformations of great arteries
Q25.80	Vascular ring due to anomalous right subclavian artery
Q25.81	Vascular ring, other and unspecified
	<i>Excludes:</i> vascular ring due to double aortic arch (Q25.44)
Q25.9	Congenital malformations of great arteries, unspecified
Q26	Congenital malformations of great veins
Q26.0	Congenital stenosis of vena cava
Q26.00	Congenital stenosis of inferior vena cava
Q26.01	Congenital stenosis of superior vena cava
Q26.1	Persistent left superior vena cava
Q26.2	Total anomalous pulmonary venous connection
	Total anomalous pulmonary venous drainage, TAPVD
Q26.20	Total anomalous pulmonary venous connection-subdiaphragmatic
Q26.21	Total anomalous pulmonary venous connection-supradiaphragmatic
Q26.3	Partial anomalous pulmonary venous connection
Q26.4	Anomalous pulmonary venous connection, unspecified
Q26.5	Anomalous portal venous connection
Q26.6	Portal vein-hepatic artery fistula
Q26.8	Other congenital malformations of great veins
	Absence of vena cava (inferior) (superior)
	Azygos continuation of inferior vena cava
	Persistent left posterior cardinal vein
	Scimitar syndrome
Q26.9	Congenital malformation of great vein, unspecified
	Anomaly of vena cava (inferior) (superior) NOS

Q27	Other congenital malformations of peripheral vascular system
<i>Excludes:</i>	anomalies of: . cerebral and precerebral vessels (Q28.0-Q28.3)
	. coronary vessels (Q24.5)
	. pulmonary artery (Q25.5-Q25.7)
	congenital retinal aneurysm (Q14.1)
	haemangioma and lymphangioma (D18.-)
	congenital naevi (Q82.5-)
Q27.0	Congenital absence and hypoplasia of umbilical artery
	Single umbilical artery
Q27.1	Congenital renal artery stenosis
Q27.2	Other congenital malformations of renal artery
	Congenital malformation of renal artery NOS
	Multiple renal arteries
Q27.3	Peripheral arteriovenous malformation
	Arteriovenous aneurysm
	<i>Excludes:</i> acquired arteriovenous aneurysm (I77.0)
Q27.4	Congenital phlebectasia
Q27.8	Other specified congenital malformations of peripheral vascular system
	Absence, atresia of artery or vein NEC Congenital:
	. aneurysm (peripheral)
	. stricture, artery
	. varix
Q27.80	Aberrant subclavian artery
	Anomalous right subclavian artery
	<i>Excludes:</i> vascular ring due to anomalous right subclavian artery (Q25.80)
Q27.9	Congenital malformation of peripheral vascular system, unspecified
	Anomaly of artery or vein NOS

Q28 Other congenital malformations of circulatory system*Excludes:* congenital aneurysm:

- . NOS (Q27.8)
- . coronary (Q24.5)
- . peripheral (Q27.8)
- . pulmonary (Q25.7)
- . retinal (Q14.1)
- . aneurysm of sinus of Valsalva (ruptured) (Q25.43)
- ruptured:

- . cerebral arteriovenous malformation (I60.8)
- . malformation of precerebral vessels (I72.-)
- Von Hippel-Lindau syndrome (Q85.82)

- Q28.0 Arteriovenous malformation of precerebral vessels
Congenital arteriovenous precerebral aneurysm (nonruptured)
- Q28.1 Other malformations of precerebral vessels
Congenital: . malformation of precerebral vessels NOS
. precerebral aneurysm (nonruptured)
- Q28.2 Arteriovenous malformation of cerebral vessels
Arteriovenous malformation of brain NOS
Congenital arteriovenous cerebral aneurysm (nonruptured)
See also Sturge-Weber(-Dimitri) syndrome (Q85.81)
- Q28.3 Other malformations of cerebral vessels
Congenital: . cerebral aneurysm (nonruptured)
. malformation of cerebral vessels NOS
- Q28.8 Other specified congenital malformations of circulatory system
Congenital aneurysm, specified site NEC
Congenital lymphatic abnormalities
- Q28.9 Congenital malformation of circulatory system, unspecified

Q30-Q34 Congenital malformations of the respiratory system**Q30 Congenital malformations of nose***Excludes:* congenital deviation of nasal septum (Q67.4)

- Q30.0 Choanal atresia
Atresia }
Congenital stenosis } of nares (anterior)(posterior)
CHARGE association
- Q30.1 Agenesis and underdevelopment of nose
Congenital absence of nose
- Q30.2 Fissured, notched and cleft nose
- Q30.3 Congenital perforated nasal septum
- Q30.8 Other congenital malformations of nose
Accessory nose
Congenital anomaly of nasal sinus wall
- Q30.9 Congenital malformation of nose, unspecified
- Q31 Congenital malformations of larynx**
- Q31.0 Web of larynx
Web of larynx: . NOS
. glottic
. subglottic
- Q31.1 Congenital subglottic stenosis
- Q31.2 Laryngeal hypoplasia
- Q31.3 Laryngocele
- Q31.4 Congenital laryngeal stridor
Congenital stridor (larynx) NOS
- Q31.40 Congenital laryngomalacia
- Q31.48 Other congenital laryngeal stridor
- Q31.8 Other congenital malformations of larynx
Absence }
Agenesis } of cricoid cartilage, epiglottis, glottis,
Atresia } larynx or thyroid cartilage
Cleft thyroid cartilage
Congenital stenosis of larynx NEC
Fissure of epiglottis
Posterior cleft of cricoid cartilage
- Q31.80 Congenital laryngeal cleft
- Q31.9 Congenital malformation of larynx, unspecified

Q32 Congenital malformations of trachea and bronchus*Excludes:* congenital bronchiectasis (Q33.4)

Q32.0 Congenital tracheomalacia

Q32.1 Other congenital malformations of trachea

Anomaly of tracheal cartilage

Atresia of trachea

Congenital: . dilatation }
 . malformation } of trachea
 . tracheocele

Q32.10 Congenital tracheal stenosis

Complete (cartilaginous) tracheal ring [stovepipe trachea]

Q32.11 Congenital tracheo-oesophageal cleft

Excludes: congenital tracheo-oesophageal fistula (Q39.1, Q39.2)

Q32.2 Congenital bronchomalacia

Q32.20 Primary congenital bronchomalacia

Q32.21 Secondary congenital bronchomalacia

Congenital bronchomalacia associated with vascular ring

Q32.3 Congenital stenosis of bronchus

Q32.4 Other congenital malformations of bronchus

Congenital malformation of bronchus NOS

Q32.40 Tracheal bronchus

Q32.41 Bronchus picus

Q32.42 Congenital diverticulum of bronchus

Q32.43 Absence of bronchus

Agenesis }

Atresia } of bronchus

Q33 Congenital malformations of lung

Q33.0 Congenital cystic lung

Congenital: . cystic lung disease

. bronchogenic cyst

Excludes: cystic lung disease, acquired or unspecified (J98.4)

Q33.00 Congenital single lung cyst

Q33.01 Congenital polycystic lung
 Congenital multiple lung cysts

Q33.02 Congenital honeycomb lung

Q33.1 Accessory lobe of lung

Q33.10 Azygos lobe of lung

Q33.2 Sequestration of lung

Q33.3 Agenesis of lung

Absence of lung (lobe)

Q33.4 Congenital bronchiectasis

Q33.5 Ectopic tissue in lung

Q33.6 Hypoplasia and dysplasia of lung

Excludes: pulmonary hypoplasia associated with:

. short gestation (P28.0)

. prolonged rupture of membranes (P01.1)

Q33.8 Other congenital malformations of lung

Q33.80 Congenital (cystic) adenomatoid malformation of the lung

Q33.81 Broncho-pulmonary isomerism

Q33.9 Congenital malformation of lung, unspecified

Q34 Other congenital malformations of respiratory system

Q34.0 Anomaly of pleura

Q34.1 Congenital cyst of mediastinum

Q34.8 Other specified congenital malformations of respiratory system

Atresia of nasopharynx

Q34.80 Congenital pulmonary lymphangiectasis

Q34.9 Congenital malformation of respiratory system, unspecified

Congenital: . absence }

. anomaly NOS } of respiratory organ

Added December 2007:

For facial clefts, codes Q35-37 the coding committee has decided to recommend use of WHO updates instead of the BPA extensions (BPA extensions are crossed through below with WHO update replacements shown). Only for Q369 the coding committee recommend to use the BPA version instead of the WHO update.

Q35-Q37 Cleft lip and cleft palate

Excludes: Robin's syndrome (Q87.08)

Q35 Cleft palate

Includes: fissure of palate
palatoschisis

Excludes: cleft palate with cleft lip (Q37.-)

Q35.0	Cleft hard palate, bilateral
Q35.1	Cleft hard palate
Q35.3	Cleft soft palate
Q35.5	Cleft hard palate with cleft soft palate
Q35.10	Cleft hard palate, unilateral
Q35.19	Cleft hard palate, unspecified
Q35.2	Cleft soft palate, bilateral
Q35.30	Cleft soft palate, unilateral
Q35.39	Cleft soft palate, unspecified
Q35.4	Cleft hard palate with cleft soft palate, bilateral
	Bilateral complete cleft palate
Q35.50	Cleft hard palate with cleft soft palate, unilateral
	Unilateral complete cleft palate
Q35.59	Cleft hard palate with cleft soft palate, unspecified
	Complete cleft palate, unspecified
Q35.6	Cleft palate, medial
	Median cleft of soft and/or hard palate
Q35.60	Central complete cleft palate
Q35.61	Central incomplete cleft palate

Q35.7	Cleft uvula
Q35.8	Cleft palate, unspecified, bilateral
Q35.90	Cleft palate, unspecified, unilateral
Q35.99	Cleft palate, unspecified
Q35.9	Cleft palate, unspecified

Q36 Cleft lip

Includes: cheiloschisis
congenital fissure of lip
harelip
labium leporinum

Excludes: cleft lip with cleft palate (Q37.-)

Q36.0	Cleft lip, bilateral
Q36.1	Cleft lip, median
Q36.90	Cleft lip, specified as unilateral
Q36.99	Cleft lip NOS

Q37 Cleft palate with cleft lip

Q37.0	Cleft hard palate with bilateral cleft lip
Q37.1	Cleft hard palate with unilateral cleft lip
Q37.10	Cleft hard palate with cleft lip, specified as unilateral
Q37.19	Cleft hard palate with cleft lip, NOS
Q37.2	Cleft soft palate with bilateral cleft lip
Q37.3	Cleft soft palate with unilateral cleft lip
Q37.4	Cleft hard and soft palate with bilateral cleft lip
Q37.5	Cleft hard and soft palate with unilateral cleft lip
Q37.50	Cleft hard and soft palate with cleft lip, specified as unilateral
Q37.59	Cleft hard and soft palate with cleft lip NOS
Q37.8	Unspecified cleft palate with bilateral cleft lip
Q37.9	Unspecified cleft palate with unilateral cleft lip
Q37.90	Unspecified, cleft palate with cleft lip, specified as unilateral
Q37.99	Cleft palate with cleft lip NOS

Q38-Q45 Other congenital malformations of the digestive system

Excludes: hernia: . inguinal (K40)
 . femoral (K41)
 . umbilical (K42)
 . ventral (K43)

Q38 Other congenital malformations of tongue, mouth and pharynx

Excludes: macrostomia (Q18.4)
 microstomia (Q18.5)

Q38.0 Congenital malformations of lips, not elsewhere classified
 Congenital malformation of lip NOS
 Labial pit
 Van der Woude's syndrome

Excludes: cleft lip (Q36.-)
 . with cleft palate (Q37.-)
 macrocheilia (Q18.6)
 microcheilia (Q18.7)

Q38.00 Congenital fistula of lip

Q38.08 Other congenital malformations of lips, not elsewhere classified

Q38.1 Ankyloglossia
 Tongue tie

Q38.2 Macroglossia

Q38.3 Other congenital malformations of tongue

Bifid tongue
 Congenital: . adhesion of tongue
 . fissure of tongue
 . dislocation or displacement of tongue

Hypoglossia

Hypoplasia of tongue

Microglossia

Lobulated tongue

Hamartomata of tongue

Q38.30 Aglossia

Q38.39 Congenital malformation of tongue NOS

Q38.4 Congenital malformations of salivary glands and ducts
 Absence }
 Accessory } (of) salivary gland or duct
 Atresia }

Congenital fistula of salivary gland

Q38.5 Congenital malformations of palate, not elsewhere classified

Absence of uvula

Congenital malformation of palate NOS

Excludes: cleft palate (Q35.-)
 . with cleft lip (Q37.-)

Q38.50 High arched palate

Q38.58 Other congenital malformations of palate, not elsewhere classified

Q38.6 Other congenital malformations of mouth

Congenital malformation of mouth NOS

Q38.7 Pharyngeal pouch

Diverticulum of pharynx

Excludes: pharyngeal pouch syndrome (D82.1)

Q38.8 Other congenital malformations of pharynx

Congenital malformation of pharynx NOS

Q38.80 Congenital palato-oesophageal incoordination

Naso-pharyngeal dysmotility

Q39 Congenital malformations of oesophagus

Excludes: congenital tracheo-oesophageal cleft (Q32.11)

Q39.0 Atresia of oesophagus without fistula

Atresia of oesophagus NOS

Q39.1 Atresia of oesophagus with tracheo-oesophageal fistula

Atresia of oesophagus with broncho-oesophageal fistula

Q39.10 Atresia of oesophagus with fistula between trachea and upper oesophageal pouch

Q39.11 Atresia of oesophagus with fistula between trachea and lower oesophageal pouch

Q39.2 Congenital tracheo-oesophageal fistula without atresia

Congenital tracheo-oesophageal fistula NOS, TOF

Q39.20	Congenital broncho-oesophageal fistula without atresia	Q40.2	Other specified congenital malformations of stomach
Q39.3	Congenital stenosis and stricture of oesophagus		Megalogastria
Q39.4	Oesophageal web		Microgastria
			Congenital: . displacement of stomach
Q39.5	Congenital dilatation of oesophagus		. diverticulum of stomach
Q39.50	Congenital cardiospasm		. hourglass stomach
	Achalasia of cardia, congenital		
			Prepyloric diaphragm
Q39.6	Diverticulum of oesophagus	Q40.21	Dysmotility of stomach
	Oesophageal pouch		Pseudo-obstruction of stomach
		Q40.22	Duplication of stomach
Q39.8	Other congenital malformations of oesophagus	Q40.3	Congenital malformation of stomach, unspecified
	Absent oesophagus	Q40.8	Other specified congenital malformations of upper alimentary tract
	Congenital displacement of oesophagus		Pyloric atresia
Q39.80	Congenital duplication of oesophagus	Q40.9	Congenital malformation of upper alimentary tract,unspecified
Q39.81	Oesophageal dysmotility		Congenital: . anomaly }
	Pseudo-obstruction of oesophagus		. deformity } NOS of upper alimentary tract
Q39.9	Congenital malformation of oesophagus, unspecified		
Q40	Other congenital malformations of upper alimentary tract	Q41	Congenital absence, atresia and stenosis of small intestine
Q40.0	Congenital hypertrophic pyloric stenosis		<i>Includes:</i> congenital obstruction, occlusion and
	Congenital or infantile: . constriction }		stricture of small intestine or intestine NOS
	. hypertrophy }	Q41.0	<i>Excludes:</i> meconium ileus (E84.1)
	. spasm } of pylorus	Q41.1	Congenital absence, atresia and stenosis of duodenum
	. stenosis }		Congenital absence, atresia and stenosis of jejunum
	. stricture }		Apple peel syndrome
	Pyloric stenosis, NOS, in infant less than three months old	Q41.2	Imperforate jejunum
	Infantile hypertrophic pyloric stenosis		Congenital absence, atresia and stenosis of ileum
Q40.1	Congenital hiatus hernia	Q41.8	Congenital absence, atresia and stenosis of other specified
	Displacement of cardia through oesophageal hiatus		parts of small intestine
	Partial thoracic stomach		Congenital absence, atresia and stenosis of multiple
	<i>Excludes:</i> congenital diaphragmatic hernia (Q79.0)		regions of small intestine
		Q41.9	Congenital absence, atresia and stenosis of small intestine,
			part unspecified
			Congenital absence, atresia and stenosis of intestine NOS

Q42 Congenital absence, atresia and stenosis of large intestine*Includes:* congenital obstruction, occlusion and stricture of large intestine

Q42.0 Congenital absence, atresia and stenosis of rectum with fistula
 For Q42.0 the following *optional* fifth character codes may be used if desired to indicate the type of fistula:

- 0 rectourethral
- 1 rectovesical
- 2 rectovulval
- 3 rectocutaneous
- 4 rectocloacal
- 8 other specified (see below)

N.B. For Congenital absence, atresia and stenosis of rectum with rectovaginal fistula, use Q42.0 and Q52.2

For Congenital gastrointestinal-urinary tract fistula without rectal absence, atresia or stenosis, use Q64.74

Q42.1 Congenital absence, atresia and stenosis of rectum without fistula
 Imperforate rectum

Q42.2 Congenital absence, atresia and stenosis of anus with fistula
 For Q42.2 the following *optional* fifth character codes may be used if desired to indicate the type of fistula:

- 0 anocutaneous
- 1 anovestibular
- 8 other

Q42.3 Congenital absence, atresia, stenosis of anus without fistula
 Imperforate anus
 Congenital anal stenosis

Q42.8 Congenital absence, atresia and stenosis of other parts of large intestine

Congenital absence, atresia and stenosis of appendix

Q42.9 Congenital absence, atresia and stenosis of large intestine, part unspecified

Q42.90 Colonic atresia

Q43 Other congenital malformations of intestine

Q43.0 Meckel's diverticulum

Q43.00 Persistent omphalomesenteric duct

Persistent vitelline duct

Q43.01 Omphalomesenteric band

Q43.02 Omphalomesenteric cyst

Q43.1 Hirschsprung's disease

Aganglioneosis

Congenital (aganglionic) megacolon

Hirschsprung's disease NOS

Q43.10 Short segment Hirschsprung's disease

Q43.11 Long segment Hirschsprung's disease

Q43.12 Total colonic aganglioneosis

Q43.13 Total intestinal aganglioneosis

Q43.2 Other congenital functional disorders of colon

Congenital dilatation of colon

Congenital macrocolon, not aganglionic

Small left colon syndrome

Megacystis, microcolon, hypoperistalsis syndrome

Neuronal intestinal dysplasia

Hyperganglioneosis

Q43.20 Large intestinal dysmotility

Pseudo-obstruction of large intestine

Q43.3 Congenital malformations of intestinal fixation

Jackson's membrane

Universal mesentery

Other anomalies of mesentery

Q43.30 Malrotation of colon

Rotation: . failure of }

. incomplete } of caecum and colon

. insufficient }

Q43.31 Congenital intraabdominal adhesions [bands]

Congenital adhesions [bands]: . omental, anomalous
 . peritoneal

Ladd's bands

Q43.38 Other congenital malformations of intestinal fixation

Q43.4	Duplication of intestine Duplication of anus, appendix, caecum and intestine Enterogenous cyst	Q44.1	Other congenital malformations of gallbladder Congenital malformation of gallbladder NOS Intrahepatic gallbladder
Q43.5	Ectopic anus Misplaced anus	Q44.2	Duplication of gallbladder Atresia of bile ducts
Q43.6	Congenital fistula of rectum and anus <i>Excludes:</i> congenital fistula: . rectovaginal (Q52.2) . urethrorectal (Q64.7)	Q44.20	Biliary atresia NOS Intrahepatic biliary atresia
	pilonidal fistula or sinus (L05.-) congenital fistula of rectum and anus with absence, atresia and stenosis (Q42.0, Q42.2)	Q44.21	Extrahepatic biliary atresia
Q43.7	Persistent cloaca Cloaca NOS	Q44.3	Congenital stenosis and stricture of bile ducts
Q43.8	Other specified congenital malformations of intestine Congenital: . blind loop syndrome . diverticulitis, colon . diverticulum, intestine	Q44.4	Choledochal cyst
	Dolichocolon Megaloappendix Megaloduodenum Transposition of: . appendix . colon . intestine	Q44.5	Other congenital malformations of bile ducts Accessory hepatic duct Congenital malformation of bile duct NOS Duplication: . biliary duct . cystic duct
	Persistent inversion of appendix	Q44.6	Cystic disease of liver Fibrocystic disease of liver
Q43.80	Microcolon	Q44.7	Other congenital malformations of liver Accessory liver
Q43.81	Small intestinal dysmotility Pseudo-obstruction of small intestine	Q44.70	Congenital: . hepatomegaly . malformation of liver NOS
Q43.82	Generalised intestinal dysmotility	Q44.71	Absence or agenesis of liver, total or lobe
Q43.83	Congenital intestinal blind loop	Q44.72	Alagille's syndrome
		Q44.73	Congenital atrophy of left lobe of liver
		Q44.74	Riedel's lobe of liver
		Q44.75	Ectopic liver Focal nodular hypoplasia of liver
Q43.9	Congenital malformation of intestine, unspecified	Q45	Other congenital malformations of digestive system
Q44	Congenital malformations of gallbladder, bile ducts and liver	<i>Excludes:</i>	congenital: . diaphragmatic hernia (Q79.0) . hiatus hernia (Q40.1)
Q44.0	Agenesis, aplasia and hypoplasia of gallbladder Congenital absence of gallbladder	Q45.0	Agenesis, aplasia and hypoplasia of pancreas Congenital absence of pancreas
		Q45.1	Annular pancreas
		Q45.2	Congenital pancreatic cyst

	Q45.3	Other congenital malformations of pancreas and pancreatic duct Accessory pancreas Congenital malformation of pancreas or pancreatic duct NOS <i>Excludes:</i> diabetes mellitus: .congenital (E10.-) .neonatal (P70.2) fibrocystic disease of pancreas (E84.-)		Q50.3	Other congenital malformations of ovary Accessory ovary Dysplastic ovary Congenital malformation of ovary NOS
				Q50.30	Ovarian streak
	Q45.30	Ectopic pancreas		Q50.4	Embryonic cyst of fallopian tube Fimbrial cyst
	Q45.8	Other specified congenital malformations of digestive system		Q50.5	Embryonic cyst of broad ligament
	Q45.80	Absence (complete)(partial) of alimentary tract NOS			Cyst: .epooeophoron
	Q45.81	Duplication of digestive organs NOS			.Gartner's duct
	Q45.82	Malposition, congenital of digestive organs NOS			.parovarian
	Q45.83	Congenital mesenteric cyst			.of mesenteric remnant
				Q50.6	Other congenital malformations of fallopian tube and broad ligament
	Q45.9	Congenital malformation of digestive system, unspecified			Accessory } (of) fallopian tube or broad ligament
		Congenital: .anomaly }			Atresia }
		.deformity NOS } of digestive system			Congenital malformation of fallopian tube or broad ligament NOS
				Q50.60	Absence of fallopian tube or broad ligament
Q50-Q56		Congenital malformations of genital organs		Q51	Congenital malformations of uterus and cervix
<i>Excludes:</i>		androgen resistance syndrome [testicular feminisation syndrome] (E34.5)		Q51.0	Agensis and aplasia of uterus Congenital absence of uterus
		syndromes associated with anomalies in the number and form of chromosomes (Q90-Q99)		Q51.1	Doubling of uterus with doubling of cervix and vagina
				Q51.2	Other doubling of uterus Doubling of uterus NOS
Q50		Congenital malformations of ovaries, fallopian tubes and broad ligaments		Q51.3	Bicornate uterus Bicornuate uterus
Q50.0		Congenital absence of ovary		Q51.4	Unicornate uterus Unicornuate uterus
		<i>Excludes:</i> Turner's syndrome (Q96.-)		Q51.5	Agensis and aplasia of cervix Congenital absence of cervix
Q50.00		Congenital absence of ovary, unilateral		Q51.6	Embryonic cyst of cervix
Q50.01		Congenital absence of ovary, bilateral		Q51.7	Congenital fistula between uterus and digestive and urinary tracts
					Uterointestinal fistula
Q50.1		Developmental ovarian cyst			Uterovesical fistula
Q50.10		Developmental ovarian cyst, single			
Q50.11		Developmental ovarian cyst, multiple			
Q50.2		Congenital torsion of ovary			

Q55 Other congenital malformations of male genital organs*Excludes:* congenital hydrocele (P83.5)

hypospadias (Q54.-)

Q55.0 Absence and aplasia of testis

Q55.00 Absence and aplasia of testis, unilateral

Monorchism

Q55.01 Absence and aplasia of testis, bilateral

Anorchism

Q55.1 Hypoplasia of testis and scrotum

Fusion of testes

Q55.2 Other congenital malformations of testis and scrotum

Congenital malformation of testis or scrotum NOS

Polyorchism

Q55.20 Retractable testis

Q55.21 Bifid scrotum

Q55.3 Atresia of vas deferens

Q55.4 Other congenital malformations of vas deferens, epididymis, seminal vesicles and prostate

Absence or aplasia of: . prostate

. spermatic cord

Congenital malformation of vas deferens, epididymis, seminal vesicles or prostate NOS

Cysts of embryonal remnants [persistent Wolffian duct]

Q55.40 Congenital cyst of hydatid of Morgagni in male

Q55.5 Congenital absence and aplasia of penis

Q55.6 Other congenital malformations of penis

Congenital malformation of penis NOS

Curvature of penis lateral

Hypoplasia of penis

Micropenis

Penile duplication

Penoscrotal transposition

Q55.8

Other specified congenital malformations of male genital organs

Q55.9

Congenital malformation of male genital organ, unspecified

Congenital: . anomaly }

. deformity } NOS of male genital organ

Q56**Indeterminate sex and pseudohermaphroditism***Excludes:*

pseudohermaphroditism:

. female, with adrenocortical disorder (E25.-)

. male, with androgen resistance (E34.5)

. with specified chromosomal anomaly (Q96-Q99)

Q56.0

Hermaphroditism, not elsewhere classified

Ovotestis

Excludes: Chimera 46,XX/46,XY true hermaphrodite (Q99.0)

Q56.1

Male pseudohermaphroditism, not elsewhere classified

Male pseudohermaphroditism NOS

Q56.2

Female pseudohermaphroditism, not elsewhere classified

Female pseudohermaphroditism NOS

Q56.3

Pseudohermaphroditism, unspecified

Q56.4

Indeterminate sex, unspecified

Ambiguous genitalia

Q60-Q64**Congenital malformations of the urinary system****Q60****Renal agenesis and other reduction defects of kidney***Includes:*

atrophy of kidney: . congenital

. infantile

congenital absence of kidney

Q60.0

Renal agenesis, unilateral

Q60.1

Renal agenesis, bilateral

Q60.2

Renal agenesis, unspecified

Q60.3

Renal hypoplasia, unilateral

Q60.4

Renal hypoplasia, bilateral

Q60.5

Renal hypoplasia, unspecified

Q60.6

Potter's syndrome

Potter's sequence

Oligohydramnios sequence

Q61	Cystic kidney disease	Q62.1	Atresia and stenosis of ureter
<i>Excludes:</i>	acquired cyst of kidney (N28.1)		Congenital occlusion of ureter
Q61.0	Congenital single renal cyst		Impervious ureter
	Cyst of kidney (congenital)(single)	Q62.10	Congenital pelviureteric junction obstruction, unilateral
Q61.1	Polycystic kidney, infantile type	Q62.11	Congenital pelviureteric junction obstruction, bilateral
Q61.2	Polycystic kidney, adult type	Q62.12	Congenital vesicoureteric junction obstruction, unilateral
Q61.3	Polycystic kidney, unspecified	Q62.13	Congenital vesicoureteric junction obstruction, bilateral
Q61.4	Renal dysplasia	Q62.18	Other specified atresia and stenosis of ureter
Q61.40	Multicystic dysplastic kidney, unilateral		
	Cystic renal dysplasia, unilateral	Q62.2	Congenital megaloureter
Q61.41	Multicystic dysplastic kidney, bilateral		Congenital dilatation of ureter
	Cystic renal dysplasia, bilateral	Q62.3	Other obstructive defects of renal pelvis and ureter
Q61.48	Other specified renal dysplasia		Congenital ureterocele
		Q62.30	Ectopic ureterocele
Q61.5	Medullary cystic kidney	Q62.31	Orthotopic ureterocele
	Sponge kidney NOS	Q62.32	Congenital polyp of ureter
Q61.50	Juvenile medullary cystic kidney	Q62.33	Congenital hydroureter
	Nephronophthisis		
Q61.51	Adult type medullary cystic kidney		
Q61.52	Medullary sponge kidney	Q62.4	Agenesis of ureter
			Absent ureter
Q61.8	Other cystic kidney disease	Q62.5	Duplication of ureter
	Fibrocystic renal degeneration or disease		Accessory ureter
	Cystic kidney disease associated with:	Q62.50	Double ureter
	. tuberous sclerosis (Q85.1)		Duplex ureter
	. Zellweger's syndrome (Q87.83)		Complete duplication of ureter
	Glomerular cystic disease	Q62.51	Triple ureter
Q61.9	Cystic kidney disease, unspecified		
Q61.90	Meckel-Gruber syndrome		
	Microcephalus with cystic kidney disease		
Q62	Congenital obstructive defects of renal pelvis and congenital malformations of ureter		
Q62.0	Congenital hydronephrosis		
	Ante-natally diagnosed hydronephrosis		

Q62.6	Malposition of ureter	Q63.1	Lobulated, fused and horseshoe kidney
	Deviation }		Renal fusion anomalies without ectopia
	Displacement } (of) ureter or		<i>Excludes:</i> crossed ectopia of kidney with fusion anomaly (Q63.22)
	Ectopic } ureteric orifice	Q63.10	Horseshoe kidney
	Implantation, anomalous }	Q63.18	Other specified renal fusion anomaly
	For Q62.6 the following <i>optional</i> fifth character	Q63.19	Renal fusion anomaly, unspecified
	subdivision can be used if desired, to indicate		
	the site of ureteric drainage:	Q63.2	Ectopic kidney
	0 bladder neck		Renal ectopia
	1 urethra		Congenital displaced kidney
	2 vagina		Malrotation of kidney
	3 vulva	Q63.20	Pelvic kidney
	4 vas deferens	Q63.21	Crossed ectopia of kidney (without fusion)
	5 seminal vesicles	Q63.22	Crossed ectopia of kidney with fusion anomaly
	8 other	Q63.28	Other specified renal ectopia
		Q63.29	Renal ectopia, unspecified
Q62.7	Congenital vesico-uretero-renal reflux		
	Congenital vesicoureteric reflux	Q63.3	Hyperplastic and giant kidney
	VUR		
	<i>Excludes:</i> vesicoureteral-reflux-associated nephropathy (N13.7)	Q63.8	Other specified congenital malformations of kidney
Q62.70	Congenital vesico-uretero-renal reflux, unilateral	Q63.81	Congenital calyceal diverticulum
Q62.71	Congenital vesico-uretero-renal reflux, bilateral		
Q62.8	Other congenital malformations of ureter	Q63.9	Congenital malformation of kidney, unspecified
	Anomaly of ureter NOS		
Q63	Other congenital malformations of kidney	Q64	Other congenital malformations of urinary system
<i>Excludes:</i>	congenital nephrotic syndrome (N04.-)	Q64.0	Epispadias
Q63.0	Accessory kidney		<i>Excludes:</i> hypospadias (Q54.-)
Q63.00	Double or triple kidney	Q64.1	Exstrophy of urinary bladder
	Duplex or triplex kidney		Ectopia vesicae
			Extroversion of bladder
		Q64.10	Cloacal exstrophy
			Ectopia cloacae
		Q64.20	Congenital posterior urethral valves
		Q64.21	Congenital anterior urethral valves

Q64.3	Other atresia and stenosis of urethra and bladder neck
	Impervious urethra
Q64.30	Congenital bladder neck obstruction
Q64.31	Congenital stricture of urethra
	Congenital stricture of anterior urethra
Q64.32	Congenital stricture of urethral meatus
Q64.33	Hypoplasia of urethra
	Atresia of urethra
Q64.4	Malformation of urachus
Q64.40	Cyst of urachus
Q64.41	Patent urachus
Q64.42	Urachal diverticulum
Q64.48	Other specified malformation of urachus
	Prolapse of urachus
Q64.5	Congenital absence of bladder and urethra
Q64.6	Congenital diverticulum of bladder
	Congenital paraureteric diverticulum
Q64.7	Other congenital malformations of bladder and urethra
	Accessory: . bladder
	. urethra
	Congenital: . hernia of bladder
	. malformation of bladder or urethra NOS
	. prolapse of: . urethra
	. urinary meatus
Q64.70	Anterior urethral diverticulum
Q64.71	Congenital prolapse of bladder (mucosa)
Q64.72	Double urethra
	Double urinary meatus
Q64.73	Ectopic urethra or urethral orifice
Q64.74	Congenital gastrointestinal-urinary tract fistula
	Congenital: . urethrorectal fistula
	. rectovesical fistula
Q64.75	Congenital megalourethra
Q64.76	Megacystis-megaureter syndrome
Q64.78	Congenital urethral syngocele

Q64.8	Other specified congenital malformations of urinary system
Q64.9	Congenital malformation of urinary system, unspecified
	Congenital: . anomaly }
	. deformity } NOS of urinary system

Q65-Q79 Congenital malformations and deformations of musculoskeletal system

Q65	Congenital deformities of hip
	CDH
<i>Excludes:</i>	clicking hip (R29.4)
Q65.0	Congenital dislocation of hip, unilateral
Q65.1	Congenital dislocation of hip, bilateral
Q65.2	Congenital dislocation of hip, unspecified
Q65.3	Congenital subluxation of hip, unilateral
Q65.4	Congenital subluxation of hip, bilateral
Q65.5	Congenital subluxation of hip, unspecified
Q65.6	Unstable hip
	Dislocatable hip
	Subluxatable hip
Q65.60	Unstable hip, unilateral
Q65.61	Unstable hip, bilateral
Q65.8	Other congenital deformities of hip
Q65.80	Dysplastic hip, unilateral
	Congenital acetabular dysplasia, unilateral
Q65.81	Dysplastic hip, bilateral
	Congenital acetabular dysplasia, bilateral
Q65.82	Anteversion of femoral neck
	Anteversion of femur
Q65.83	Congenital coxa valga
Q65.84	Congenital coxa vara
Q65.9	Congenital deformity of hip, unspecified

Q66 Congenital deformities of feet*Excludes:* reduction defects of feet (Q72.-)

valgus deformities (acquired) (M21.0)

varus deformities (acquired) (M21.1)

Q66.0 Talipes equinovarus

Q66.1 Talipes calcaneovarus

Q66.2 Metatarsus varus

Metatarsus adductus

Q66.3 Other congenital varus deformities of feet

Hallux varus, congenital

Q66.4 Talipes calcaneovalgus

Q66.5 Congenital pes planus

Flat foot: . congenital

. rigid

. spastic (everted)

Excludes: pes planus acquired (M21.4)

Q66.6 Other congenital valgus deformities of feet

Metatarsus valgus

Q66.7 Pes cavus

Q66.8 Other congenital deformities of feet

Clubfoot NOS

Hammer toe, congenital

Talipes: . NOS

. asymmetric

Tarsal coalition

Vertical talus

Q66.80 Rocker bottom foot

Q66.81 Congenital short Achilles tendon

Q66.9 Congenital deformity of feet, unspecified

Q67 Congenital musculoskeletal deformities of head, face, spine and chest*Excludes:* congenital malformation syndromes classified to Q87.-

Potter's sequence [syndrome] (Q60.6)

Q67.0 Facial asymmetry

Q67.1 Compression facies

Excludes: Potter's facies (Q60.6)

Q67.2 Dolichocephaly

Q67.3 Plagiocephaly

Asymmetric head

Q67.4 Other congenital deformities of skull, face and jaw

Hemifacial atrophy or hypertrophy

Squashed or bent nose, congenital

Excludes: dentofacial anomalies [including malocclusion] (K07.-)

syphilitic saddle nose (A50.5)

Goldenhar syndrome [oculo-auriculo-vertebral syndrome] (Q87.04)

Q67.40 Depressions in skull

Q67.41 Deviation of nasal septum, congenital

Q67.5 Congenital deformity of spine

Excludes: infantile idiopathic scoliosis (M41.0)

scoliosis due to congenital bony

malformation (Q76.3)

Q67.50 Congenital scoliosis, postural

Q67.52 Congenital postural curvature of spine, NOS

Q67.58 Other specified congenital deformity of spine

Q67.6 Pectus excavatum

Congenital funnel chest

Q67.7 Pectus carinatum

Congenital pigeon chest

Q67.8 Other congenital deformities of chest

Congenital deformity of chest wall NOS

Q68 Other congenital musculoskeletal deformities*Excludes:* reduction defects of limb(s) (Q71-Q73)

- Q68.0 Congenital deformity of sternocleidomastoid muscle
 Congenital (sternomastoid) torticollis
 Contracture of sternocleidomastoid (muscle)
 Sternomastoid tumour (congenital)
Excludes: sternomastoid swelling due to birth trauma (P15.2)
- Q68.1 Congenital deformity of hand
 Congenital clubfinger
 Camptodactyly
- Q68.10 Clinodactyly
- Q68.2 Congenital deformity of knee
 Q68.20 Congenital dislocation of knee
 Q68.21 Congenital genu recurvatum
 Q68.28 Other specified congenital deformity of knee
- Q68.3 Congenital bowing of femur
Excludes: anteversion of femur (neck) (Q65.8)
- Q68.4 Congenital bowing of tibia and fibula
 Q68.5 Congenital bowing of long bones of leg, unspecified
 Q68.8 Other specified congenital musculoskeletal deformities
 Congenital deformity of:
 . clavicle
 . elbow
 . forearm
 . scapula
 Congenital dislocation of shoulder
 Arthrogryposis NOS
Excludes: arthrogryposis multiplex congenita (Q74.3)
- Q68.80 Congenital dislocation of radial head

Q69 Polydactyly*Excludes:* acrocephalopolysyndactyly (Q87.01)

For Q69.0-Q69.2 the following RCPCH fifth-character extensions can be used if desired:

- 0 Preaxial
 1 Mesoaxial
 2 Postaxial
 9 unspecified
- Q69.0 Accessory finger(s)
 Supernumerary finger(s)
- Q69.1 Accessory thumb(s)
 Supernumerary thumb(s)
- Q69.2 Accessory toe(s)
 Supernumerary toe(s)
 Accessory [supernumerary] hallux
- Q69.9 Polydactyly, unspecified
 Supernumerary digit(s) NOS
- Q70 Syndactyly**
- Excludes:* acrocephalopolysyndactyly (Q87.00)
 acrocephalosyndactyly (Q87.01)
- Q70.0 Fused fingers
 Complex syndactyly of fingers with synostosis
- Q70.1 Webbed fingers
 Simple syndactyly of fingers without synostosis
- Q70.2 Fused toes
 Complex syndactyly of toes with synostosis
- Q70.3 Webbed toes
 Simple syndactyly of toes without synostosis
- Q70.4 Polysyndactyly
- Q70.9 Syndactyly, unspecified
- Q70.90 Symphalangism
 Symphalangy NOS

Q71 Reduction defects of upper limb

- Q71.0 Congenital complete absence of upper limb(s)
Amelia of upper limb
- Q71.1 Congenital absence of upper arm and forearm with hand present
Phocomelia of upper limb
- Q71.2 Congenital absence of both forearm and hand
- Q71.3 Congenital absence of hand and finger(s)
- Q71.30 Congenital absence of finger(s)
[Remainder of hand intact]
- Q71.31 Absence or hypoplasia of thumb
[Other digits intact]
- Q71.4 Longitudinal reduction defect of radius
Clubhand (congenital)
Radial clubhand
Absence of radius
Excludes: thrombocytopenia with absent radius syndrome (Q87.25)
Fanconi's anaemia with absent radius (D61.0)
- Q71.5 Longitudinal reduction defect of ulna
- Q71.6 Lobster-claw hand
Congenital cleft hand
- Q71.8 Other reduction defects of upper limb(s)
Congenital shortening of upper limb(s)
- Q71.9 Reduction defect of upper limb, unspecified
Congenital amputation of upper limb NOS
Constriction ring syndrome of upper limb NOS

Q72 Reduction defects of lower limb

- Q72.0 Congenital complete absence of lower limb(s)
Amelia of lower limb
- Q72.1 Congenital absence of thigh and lower leg with foot present
Phocomelia of lower limb
- Q72.2 Congenital absence of both lower leg and foot
- Q72.3 Congenital absence of foot and toe(s)

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

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

- Q72.4 Longitudinal reduction defect of femur
Proximal femoral focal deficiency

- Q72.5 Longitudinal reduction defect of tibia
Absence of tibia

- Q72.6 Longitudinal reduction defect of fibula
Absence of fibula

- Q72.7 Split foot

- Q72.8 Other reduction defects of lower limb(s)
Congenital shortening of lower limb(s)

- Q72.9 Reduction defect of lower limb, unspecified
Congenital amputation of lower limb NOS
Constriction ring syndrome of lower limb NOS

Q73 Reduction defects of unspecified limb

- Q73.0 Congenital absence of unspecified limb(s)
Amelia NOS
- Q73.1 Phocomelia, unspecified limb(s)
Phocomelia NOS
- Q73.8 Other reduction defects of unspecified limb(s)
Longitudinal reduction deformity of unspecified limb(s)
Ectromelia NOS }
Hemimelia NOS } of limb(s) NOS
Reduction defect }
Amputation of unspecified limb(s)
Constriction ring syndrome of unspecified limb(s)
- Q73.80 Absent digits NOS
Excludes: congenital absence of all fingers (Q71.80)
congenital absence of all toes (Q72.80)

Q74 Other congenital malformations of limb(s)*Excludes:* polydactyly (Q69.-)

reduction defect of limb (Q71-Q73)

syndactyly (Q70.-)

Q74.0 Other congenital malformations of upper limb(s), including shoulder girdle

Congenital pseudoarthrosis of clavicle

Congenital cubitus valgus or varus

Q74.00 Accessory carpal bones

Q74.01 Madelung's deformity

Q74.02 Cleidocranial dysostosis

Q74.03 Sprengel's deformity

Congenital elevation of the scapula

Q74.04 Macrodactylia (fingers)

Q74.05 Triphalangeal thumb

Q74.06 Radioulnar synostosis

Radioulnar dysostosis

Q74.07 Humero-ulnar synostosis

Q74.08 Humero-radial synostosis

Q74.09 Bifid digit(s) of upper limb

Q74.1 Congenital malformation of knee

Congenital: . absence of patella

. dislocation of patella

. genu: . valgum

. varum

Rudimentary patella

Excludes: congenital: . dislocation of knee(Q68.2)

. genu recurvatum(Q68.2)

nail patella syndrome(Q87.2)

Q74.2 Other congenital malformations of lower limb(s), including pelvic girdle

Congenital malformation (of): . ankle (joint)

. sacroiliac (joint)

Excludes: anteversion of femur (neck) (Q65.8)

Q74.20 Congenital fusion of sacroiliac joint

Q74.21 Astragaloscapoid synostosis

Q74.22 Congenital angulation of tibia

Q74.23 Bifid digit(s) of lower limb

Q74.3 Arthrogryposis multiplex congenita

Excludes: primary disorders of muscles (G71.-)

congenital viral myositis (P35.8)

infantile spinal muscular atrophy (G12.0)

Q74.8 Other specified congenital malformations of limb(s)

Q74.80 Brachydactyly

Q74.81 Congenital overgrowth of limb(s)

Congenital hemihypertrophy

Q74.82 Congenital undergrowth of limb(s)

Excludes: hemiatrophy NOS (R68.82)

Q74.83 Congenital limb asymmetry, unspecified

Q74.84 Larsen's syndrome

Q74.9 Unspecified congenital malformation of limb(s)

Congenital anomaly of limb(s) NOS

Q75 Other congenital malformations of skull and face bones*Excludes:* congenital malformation of face NOS (Q18.-)

congenital malformation syndromes classified to Q87.-

dentofacial anomalies [including malocclusion] (K07.-)

musculoskeletal deformities of head and face (Q67.0-Q67.4)

skull defects associated with congenital anomalies of

brain such as: . anencephaly (Q00.0)

. encephalocele (Q01.-)

. hydrocephalus (Q03.-)

. microcephaly (Q02)

Q75.0	Craniosynostosis Imperfect fusion of skull Pfeiffer syndrome <i>Excludes:</i> thanatophoric dwarfism/trigonocephaly association (Q77.1) acrocephalo(poly)syndactyly (Q87.0-) clover leaf skull (Kleeblattsch„del deformity syndrome) (Q03.80)	Q75.8	Other specified congenital malformations of skull and face bones Absence of skull bone, congenital Congenital deformity of forehead Platybasia
Q75.00	Coronal craniosynostosis	Q75.80	Localised skull defects
Q75.01	Brachycephaly Sagittal craniosynostosis Scaphocephaly	Q75.81	Frontonasal dysplasia Median cleft facial syndrome
Q75.02	Trigonocephaly <i>Excludes:</i> thanatophoric dwarfism (Q77.1)	Q75.9	Congenital malformation of skull and face bones, unspecified Congenital anomaly of: . face bones NOS . skull NOS
Q75.03	Craniosynostosis of other multiple sutures Acrocephaly Oxycephaly Turricephaly	Q76 <i>Excludes:</i>	Congenital malformations of spine and bony thorax congenital musculoskeletal deformities of spine and chest (Q67.5-Q67.8)
Q75.1	Craniofacial dysostosis Crouzon's disease	Q76.0	Spina bifida occulta <i>Excludes:</i> meningocele (spinal) (Q05.-) spina bifida (aperta)(cystica) (Q05.-)
Q75.2	Hypertelorism	Q76.1	Klippel-Feil syndrome Cervical fusion syndrome
Q75.3	Macrocephaly	Q76.2	Congenital spondylolisthesis Congenital spondylolysis <i>Excludes:</i> spondylolisthesis (acquired) (M43.1) spondylolysis (acquired) (M43.0)
Q75.30	Familial (benign) macrocephaly	Q76.3	Congenital scoliosis due to congenital bony malformation Kyphoscoliosis due to congenital bony malformation Fusion or failure of segmentation with scoliosis
Q75.4	Mandibulofacial dysostosis <i>Note:</i> Code Q75.4 is to be used for the isolated anomaly of skull and face bones. When this condition occurs as part of Treacher Collins [-Franceschetti] [-Klein] syndrome use (Q87.0A).	Q76.30	Single hemivertebra with congenital scoliosis
Q75.5	Oculomandibular dysostosis <i>Note:</i> Code Q75.5 is to be used for the isolated anomaly of skull and face bones. When this condition occurs as part of Hallerman-Streiff syndrome use (Q87.05).	Q76.38	Congenital scoliosis due to other specified congenital bony malformation

Q76.4	Other congenital malformations of spine, not associated with scoliosis
	Congenital:
	. fusion of spine }
	. gibbus }
	. kyphosis }
	. lordosis }
	. malformation of lumbosacral (joint) (region) }
	Malformation of spine }
	Platyspondylisis }
	Supernumerary vertebra }
Q76.40	Congenital absence of vertebra(e)
Q76.41	Congenital anomalies of sacral vertebrae
	Sacral agenesis
Q76.42	Congenital anomalies of other vertebrae
Q76.43	Congenital lordosis, postural
Q76.5	Cervical rib
	Supernumerary rib in cervical region
Q76.6	Other congenital malformations of ribs
	Congenital malformation of ribs NOS
	<i>Excludes:</i> short rib syndrome (Q77.2)
Q76.60	Congenital absence of rib
Q76.61	Congenital fusion of ribs
Q76.62	Accessory rib
	<i>Excludes:</i> cervical rib (Q76.5)
Q76.7	Congenital malformation of sternum
	Misshapen sternum
	<i>Excludes:</i> pectus excavatum (Q67.6)
	pectus carinatum (Q67.7)
Q76.70	Congenital absence of sternum
Q76.71	Sternum bifidum
Q76.78	Other specified congenital malformation of sternum

Q76.8	Other congenital malformations of bony thorax
Q76.9	Congenital malformation of bony thorax, unspecified
Q77	Osteochondrodysplasia with defects of growth of tubular bones and spine
<i>Excludes:</i>	mucopolysaccharidosis (E76.0-E76.3)
Q77.0	Achondrogenesis
Q77.00	Achondrogenesis, type I
Q77.01	Achondrogenesis, type II
Q77.02	Hypochondrogenesis
Q77.1	Thanatophoric short stature
	Thanatophoric dwarfism/trigonocephaly association
	Thanatophoric dysplasia (with clover leaf skull)
Q77.2	Short rib syndrome
	Asphyxiating thoracic dysplasia [Jeune]
	Jeune's syndrome
Q77.3	Chondrodysplasia punctata
	Chondrodystrophia calcificans congenita
	Conradi (-Hunerman) syndrome
	Congenital multiple epiphyseal dysplasia
	Rhizomelic syndrome
	<i>Excludes:</i> warfarin embryopathy (Q86.2)
Q77.4	Achondroplasia
	Achondroplastic dwarfism
	Hypochondroplasia
Q77.5	Diastrophic dysplasia
	Diastrophic dwarfism
Q77.6	Chondroectodermal dysplasia
	Ellis-van Creveld syndrome
Q77.7	Spondyloepiphyseal dysplasia

Q77.8	Other osteochondrodysplasia with defects of growth of tubular bones and spine Acrodysostosis Kniest dysplasia
Q77.80	Metatropic dwarfism
	Metatropic dysplasia
Q77.81	Metaphyseal chondrodysplasia Metaphyseal dysostosis
Q77.9	Osteochondrodysplasia with defects of growth of tubular bones and spine, unspecified

Q78 Other osteochondrodysplasias

Q78.0	Osteogenesis imperfecta Fragilitas ossium Osteopsathyrosis
Q78.00	Osteogenesis imperfecta congenita
Q78.08	Other osteogenesis imperfecta Osteogenesis imperfecta tarda
Q78.1	Polyostotic fibrous dysplasia McCune-Albright(-Sternberg) syndrome
Q78.2	Osteopetrosis Albers-Schönberg syndrome Marble bone disease
Q78.3	Progressive diaphyseal dysplasia Camurati-Engelmann syndrome
Q78.4	Enchondromatosis
Q78.40	Enchondromatosis with haemangiomata Maffucci's syndrome [Kast's syndrome]
Q78.48	Other specified enchondromatosis Dyschondroplasia Ollier's disease Osteochondromatosis syndrome <i>Excludes:</i> osteochondromatosis, NOS (D48.0)

Q78.5	Metaphyseal dysplasia Pyle's syndrome
Q78.6	Multiple congenital exostoses Diaphyseal aclasis
Q78.8	Other specified osteochondrodysplasias <i>Excludes:</i> chondrodystrophic myotonia [Schwartz-Jampel] (G71.16)
Q78.80	Osteopoikilosis
Q78.9	Osteochondrodysplasia, unspecified Osteodystrophy NOS

Q79 Congenital malformations of the musculoskeletal system, not elsewhere classified

<i>Excludes:</i>	congenital (sternomastoid) torticollis (Q68.0)
Q79.0	Congenital diaphragmatic hernia <i>Excludes:</i> congenital hiatus hernia (Q40.1)
Q79.00	Congenital anterior (foramen of Morgagni) hernia
Q79.01	Congenital posterolateral (foramen of Bochdalek) hernia
Q79.1	Other congenital malformations of diaphragm Congenital malformation of diaphragm NOS
Q79.10	Congenital eventration of diaphragm
Q79.11	Congenital absent hemidiaphragm, (unilateral)
Q79.12	Congenital absent diaphragm Congenital absent hemidiaphragm, bilateral
Q79.2	Exomphalos Omphalocele <i>Excludes:</i> umbilical hernia (K42.-)
Q79.3	Gastroschisis
Q79.4	Prune belly syndrome
Q79.5	Other congenital malformations of abdominal wall <i>Excludes:</i> umbilical hernia (K42.-)
Q79.6	Ehlers-Danlos syndrome

Q79.8	Other congenital malformations of the musculoskeletal system Accessory muscle Popliteal web syndrome Congenital shortening of tendon <i>Excludes:</i> achilles tendon (Q66.81)
Q79.80	Congenital constriction bands
Q79.81	Absence of muscle and/or tendon
Q79.82	Poland's anomaly [syndrome]
Q79.9	Congenital malformation of musculoskeletal system, unspecified Congenital: . anomaly NOS } . deformity NOS } of musculoskeletal system NOS Unspecified anomalies of muscle, tendon, bones, cartilage or connective tissue

Q80-Q89 Other congenital malformations

Q80	Congenital ichthyosis
<i>Excludes:</i>	Refsum's disease (G60.1)
Q80.0	Ichthyosis vulgaris
Q80.1	X-linked ichthyosis
Q80.2	Lamellar ichthyosis (Non-bullous ichthyosiform erythroderma) Severe form known as - Collodion baby
Q80.3	Congenital bullous ichthyosiform erythroderma (Epidermolytic hyperkeratosis)
Q80.4	Harlequin fetus
Q80.8	Other congenital ichthyosis <i>Excludes:</i> Sjogren-Larsson syndrome (Q87.1A)
Q80.9	Congenital ichthyosis unspecified
Q81	Epidermolysis bullosa
Q81.0	Epidermolysis bullosa simplex <i>Excludes:</i> Cockayne's syndrome (Q87.1)
Q81.1	Epidermolysis bullosa letalis Herlitz' syndrome
Q81.2	Epidermolysis bullosa dystrophica

Q81.8	Other epidermolysis bullosa
Q81.9	Epidermolysis bullosa, unspecified
Q82	Other congenital malformations of skin
<i>Excludes:</i>	acrodermatitis enteropathica (E83.2) congenital erythropoietic porphyria (E80.0) pilonidal cyst or sinus (L05.-) Sturge-Weber(-Dimitri) syndrome (Q85.8)
Q82.0	Hereditary lymphoedema
Q82.1	Xeroderma pigmentosum
Q82.2	Mastocytosis Urticaria pigmentosa <i>Excludes:</i> malignant mastocytosis (C96.2)
Q82.3	Incontinentia pigmenti
Q82.4	Ectodermal dysplasia (anhidrotic) <i>Excludes:</i> Ellis-van Creveld syndrome (Q77.6) ectodermal dysplasia, hidrotic (Q82.82)
Q82.5	Congenital non-neoplastic naevus Birthmark NOS Naevus: . sanguineous . vascular NOS . verrucous <i>Excludes:</i> caf, au lait spots (L81.3) lentigo (L81.4) naevus: . NOS (D22.-) . araneus (I78.1) . melanocytic (D22.-) . pigmented (D22.-) . spider (I78.1) . stellar (I78.1) capillary haemangioma (D18.00) cavernous haemangioma (D18.01) mixed haemangioma (D18.02)

Q82.50	Naevus flammeus [Portwine stain]
Q82.51	Strawberry naevus
	<i>Note:</i> This term should be used for typical strawberry naevi. Massive, non-superficial or otherwise atypical lesions should be coded to D18.0-.
Q82.52	Mongolian blue spot
Q82.58	Other specified congenital non-neoplastic naevus
Q82.8	Other specified congenital malformations of skin
	Benign familiar pemphigus [Hailey-Hailey]
	Cutis laxa (hyperelastica)
	Dermatoglyphic anomalies [<i>Excludes:</i> abnormal palmar creases - Q82.80]
	Inherited keratosis palmaris et plantaris
	Keratosis follicularis [Darier-White]
	<i>Excludes:</i> Ehlers-Danlos syndrome (Q79.6)
Q82.80	Abnormal palmar creases
Q82.81	Accessory skin tags
Q82.82	Ectodermal dysplasia, hidrotic
	<i>Excludes:</i> ectodermal dysplasia, anhidrotic (Q82.4)
Q82.83	Hypomelanosis of Ito
Q82.9	Congenital malformation of skin, unspecified
Q83	Congenital malformations of breast
<i>Excludes:</i>	absence of pectoral muscle (Q79.81)
Q83.0	Congenital absence of breast with absent nipple
Q83.1	Accessory breast
	Supernumerary breast
Q83.2	Absent nipple
Q83.3	Accessory nipple
	Supernumerary nipple
Q83.8	Other congenital malformations of breast
	Hypoplasia of breast
Q83.9	Congenital malformation of breast, unspecified

Q84	Other congenital malformations of integument
Q84.0	Congenital alopecia
	Congenital atrichosis
Q84.1	Congenital morphological disturbances of hair, not elsewhere classified
	Beaded hair
	Monilethrix
	Pili annulati
	Pili torti
	<i>Excludes:</i> Menkes' kinky hair syndrome (E83.0)
Q84.2	Other congenital malformations of hair
	Congenital malformation of hair NOS
	Persistent lanugo
Q84.20	Congenital hypertrichosis
Q84.3	Anonychia
	Congenital absent nails
	<i>Excludes:</i> nail patella syndrome (Q87.2)
Q84.4	Congenital leukonychia
Q84.5	Enlarged and hypertrophic nails
	Congenital onychauxis
	Pachyonychia
Q84.6	Other congenital malformations of nails
	Congenital: . clubnail
	. koilonychia
	. malformation of nail NOS
Q84.8	Other specified congenital malformations of integument
Q84.80	Aplasia cutis congenita
Q84.9	Congenital malformation of integument, unspecified
	Congenital: . anomaly NOS }
	. deformity NOS } of integument NOS

Q85 Phakomatoses, not elsewhere classified

Excludes: ataxia-telangiectasia [Louis-Bar] (G11.30)
familial dysautonomia [Riley-Day] (G90.1)

Q85.0 Neurofibromatosis (nonmalignant)
Von Recklinghausen's disease

Q85.1 Tuberous sclerosis
Bourneville's disease
Epiloia

Q85.8 Other phakomatoses, not elsewhere classified
Excludes: Meckel-Gruber syndrome (Q61.9)

Q85.80 Peutz-Jeghers syndrome
Q85.81 Sturge-Weber(-Dimitri) syndrome
Q85.82 Von Hippel-Lindau syndrome
Q85.83 Gardner's syndrome
Osteomatosis-intestinal polyposis syndrome

Q85.9 Phakomatosis, unspecified
Hamartosis NOS

Q86 Congenital malformation syndromes due to known exogenous causes, not elsewhere classified

Excludes: iodine-deficiency-related hypothyroidism (E00-E02)
nonteratogenic effects of substances transmitted
via placenta or breast milk (P04.-)

Q86.0 Fetal alcohol syndrome (dysmorphic)
Q86.1 Fetal hydantoin syndrome
Q86.2 Dysmorphism due to warfarin
Q86.8 Other congenital malformation syndromes due to known
exogenous causes

Congenital malformations due to methylmercury
Q86.80 Congenital malformations due to valproate
Q86.81 Congenital malformations due to Vitamin A
Q86.82 Congenital malformations due to thalidomide
Q86.83 Congenital malformations due to cytotoxic agents
Q86.84 Congenital malformations due to other drugs
Q86.85 Congenital malformations due to ionising radiation

Q87 Other specified congenital malformation syndromes affecting multiple systems

Q87.0 Congenital malformation syndromes predominantly affecting
facial appearance

Excludes: cherubism (K10.80)
Waardenburg's syndrome (E70.30)

Q87.00 Acrocephalopolysyndactyly
Acrocephalopolysyndactyly type I, Noack syndrome
Acrocephalopolysyndactyly type II, Carpenter syndrome

Q87.01 Acrocephalosyndactyly
Apert's syndrome
Vogt cephalodactyly

Q87.02 Cryptophthalmos syndrome
Q87.03 Cyclopia [cyclops][cyclopism][synophthalmia]
Q87.04 Goldenhar syndrome

Oculo-auriculo-vertebral syndrome [Hemifacial microsomia
syndrome]

Q87.05 Hallerman-Streiff syndrome
Excludes: (isolated) oculomandibular dysostosis (Q75.5)

Q87.06 Moebius syndrome
Q87.07 Oro-facial-digital syndrome
Oro-facial-digital syndrome types I and II
Mohr syndrome

Q87.08 Pierre Robin sequence
Robin syndrome/sequence

Q87.09 Stickler syndrome
Hereditary progressive arthro-ophthalmopathy

Q87.0A Treacher Collins [-Franceschetti] [-Klein] syndrome
Excludes: (isolated) mandibulofacial dysostosis (Q75.4)

Q87.0B Trico-rhino-phalangeal syndrome
Type I

Type II [Langer-Giedion]

Q87.0C Whistling face syndrome
Q87.0D Ullrich-Feichtiger's syndrome
Dyscraniopygophalangism

Q87.0E	Pena-Shokeir syndrome	Q87.24	Sirenomelia syndrome
	Camptodactyly-ankyloses-facial anomalies-pulmonary hypoplasia syndrome	Q87.25	Thrombocytopenia with absent radius syndrome
Q87.0F	Other specified congenital malformation syndromes predominantly affecting facial appearance		TAR syndrome
		Q87.26	VATER association
			VACTERL association
Q87.1	Congenital malformation syndromes predominantly associated with short stature	Q87.28	Other specified congenital malformation syndromes predominantly involving limbs
	<i>Excludes:</i> Ellis-van Creveld syndrome (Q77.6)		
Q87.10	Aarskog syndrome	Q87.3	Congenital malformation syndromes involving early overgrowth
Q87.11	Cockayne syndrome	Q87.30	Beckwith-Wiedemann syndrome
Q87.12	Cornelia de Lange syndrome		Beckwith's syndrome
	Amsterdam dwarf [Brachmann-de Lange syndrome]	Q87.31	Sotos syndrome
Q87.13	Dubowitz syndrome		Cerebral gigantism
Q87.14	Noonan syndrome	Q87.32	Weaver syndrome
Q87.15	Prader-Willi syndrome	Q87.38	Other specified congenital malformation syndromes involving early overgrowth
Q87.16	Robinow-Silverman-Smith syndrome	Q87.4	Marfan's syndrome
Q87.17	Russell-Silver syndrome		Arachnodactyly NOS
Q87.18	Seckel syndrome	Q87.5	Other congenital malformation syndromes with other skeletal changes
	Bird-headed dwarfism		
	Microcephalic primordial dwarfism	Q87.8	Other specified congenital malformation syndromes, not elsewhere classified
Q87.19	Smith-Lemli-Opitz syndrome		
	7-dehydrocholesterol reductase deficiency	Q87.80	Alport's syndrome
Q87.1A	Sjogren-Larsson syndrome	Q87.81	Laurence-Moon-Biedl syndrome
	Fatty alcohol:nicotinamide adenine dinucleotide oxidoreductase deficiency		Laurence-Moon(-Bardet)-Biedl syndrome
Q87.1B	Other specified congenital malformation syndromes predominantly associated with short stature	Q87.83	Zellweger syndrome
			<i>Note:</i> this is a peroxisomal disorder
			<i>Excludes:</i> Zellweger-like syndrome (E88.8F)
Q87.2	Congenital malformation syndromes predominantly involving limbs		pseudo-Zellweger syndrome (E88.8J)
	<i>Excludes:</i> Fanconi's anaemia with absent radius (D61.0)	Q87.84	William's syndrome
Q87.20	Holt-Oram syndrome	Q87.85	Angelman's syndrome
Q87.21	Klippel-Tr,naunay-Weber syndrome		[Happy puppet syndrome]
Q87.22	Nail patella syndrome		
Q87.23	Rubinstein-Taybi syndrome		

Q89 Other congenital malformations, not elsewhere classified

Q89.0	Congenital malformations of spleen
	Congenital splenomegaly [hyperplasia of spleen]
	Hypoplasia of }
	Mis-shapen }
	Accessory } spleen
	Ectopic }
	<i>Excludes:</i> isomerism of atrial appendages (with asplenia or polysplenia) (Q20.6)
Q89.00	Congenital asplenia
	Congenital absence of spleen
Q89.08	Other specified congenital malformation of spleen
Q89.1	Congenital malformations of adrenal gland
	Accessory } adrenal gland
	Ectopic }
	<i>Excludes:</i> congenital adrenal hyperplasia (E25.0)
Q89.10	Congenital absence of adrenal gland
Q89.11	Congenital adrenal hypoplasia
Q89.18	Other specified congenital malformation of adrenal gland
Q89.2	Congenital malformations of other endocrine glands
Q89.20	Congenital malformations of pituitary gland
Q89.21	Congenital malformations of thyroid gland
Q89.22	Persistent thyroglossal duct
Q89.23	Thyroglossal cyst
Q89.24	Congenital malformations of parathyroid gland
Q89.25	Congenital malformations of thymus
Q89.3	Situs inversus
	<i>Excludes:</i> dextrocardia NOS (Q24.0)
Q89.30	Dextrocardia with situs inversus
Q89.31	Mirror-image atrial arrangement with situs inversus
Q89.32	Situs inversus abdominalis
	Situs transversus abdominalis

	Transposition of abdominal viscera
Q89.33	Situs inversus thoracis
	Situs transversus thoracis
	Transposition of thoracic viscera
Q89.34	Kartagener's syndrome
	Kartagener's triad
	<i>Excludes:</i> other immotile cilia syndromes (J98.80)
Q89.38	Other specified situs inversus
Q89.4	Conjoined twins
Q89.40	Dicephaly
	Two heads
Q89.41	Craniopagus
	Head-joined twins
Q89.42	Thoracopagus
	Thorax-joined twins
Q89.43	Xiphopagus
	Xiphoid and pelvis-joined twins
Q89.44	Pygopagus
	Buttock-joined twins
Q89.45	Double monster
Q89.48	Other specified conjoined twins
Q89.7	Multiple congenital malformations, not elsewhere classified
	Multiple congenital: . anomalies NOS
	. deformities NOS
	<i>Excludes:</i> congenital malformation syndromes affecting multiple systems (Q87.-)
Q89.8	Other specified congenital malformations
Q89.80	Caudal dysplasia sequence
Q89.9	Congenital malformation, unspecified
	Congenital: . anomaly NOS
	. deformity NOS

Q90-Q99	Chromosomal abnormalities, not elsewhere classified
Q90	Down's syndrome
Q90.0	Trisomy 21, meiotic nondisjunction
Q90.1	Trisomy 21, mosaicism (mitotic nondisjunction)
Q90.2	Trisomy 21, translocation
Q90.9	Down's syndrome, unspecified
	Trisomy 21 NOS
Q91	Edward's syndrome and Patau's syndrome
Q91.0	Trisomy 18, meiotic nondisjunction
Q91.1	Trisomy 18, mosaicism (mitotic nondisjunction)
Q91.2	Trisomy 18, translocation
Q91.3	Edward's syndrome, unspecified
Q91.4	Trisomy 13, meiotic nondisjunction
Q91.5	Trisomy 13, mosaicism (mitotic nondisjunction)
Q91.6	Trisomy 13, translocation
Q91.7	Patau's syndrome, unspecified
Q92	Other trisomies and partial trisomies of the autosomes, not elsewhere classified
<i>Includes:</i>	unbalanced translocations and insertions
<i>Excludes:</i>	trisomies of chromosomes 13, 18, 21 (Q90-Q91)
Q92.0	Whole chromosome trisomy, meiotic nondisjunction
Q92.1	Whole chromosome trisomy, mosaicism (mitotic nondisjunction)
Q92.2	Major partial trisomy
	Whole arm or more duplicated
Q92.3	Minor partial trisomy
	Less than whole arm duplicated
Q92.4	Duplications seen only at prometaphase
Q92.5	Duplications with other complex rearrangements
Q92.6	Extra marker chromosomes
Q92.7	Triploidy and polyploidy
Q92.8	Other specified trisomies and partial trisomies of autosomes
Q92.9	Trisomy and partial trisomy of autosomes, unspecified

Q93	Monosomies and deletions from the autosomes, not elsewhere classified
Q93.0	Whole chromosome monosomy, meiotic nondisjunction
Q93.1	Whole chromosome monosomy, mosaicism (mitotic nondisjunction)
Q93.2	Chromosome replaced with ring or dicentric
Q93.3	Deletion of short arm of chromosome 4
	Wolff-Hirschorn syndrome
Q93.4	Deletion of short arm of chromosome 5
	Cri du chat syndrome
Q93.5	Other deletions of part of a chromosome
	Deletion of long arm of chromosome 13
	Deletion of long or short arm of chromosome 18
	[18p- or 18q syndrome]
Q93.50	Deletion of long arm of chromosome 21
	Anti-mongolism syndrome
Q93.6	Deletions seen only at prometaphase
Q93.7	Deletions with other complex rearrangements
Q93.8	Other deletions from the autosomes
Q93.9	Deletion from autosomes, unspecified
Q95	Balanced rearrangements and structural markers, not elsewhere classified
<i>Includes:</i>	Robertsonian and balanced reciprocal translocations and insertions
Q95.0	Balanced translocation and insertion in normal individual
Q95.1	Chromosome inversion in normal individual
Q95.2	Balanced autosomal rearrangement in abnormal individual
Q95.3	Balanced sex/autosomal rearrangement in abnormal individual
Q95.4	Individuals with marker heterochromatin
Q95.5	Individuals with autosomal fragile site
Q95.8	Other balanced rearrangements and structural markers
Q95.9	Balanced rearrangement and structural marker, unspecified

Q96	Turner's syndrome		
<i>Excludes:</i>	Noonan's syndrome (Q87.14)		
Q96.0	Karyotype 45,X		
Q96.1	Karyotype 46,X iso (Xq)		
Q96.2	Karyotype 46,X with abnormal sex chromosome, except iso (Xq)		
Q96.3	Mosaicism, 45,X/46,XX or XY		
Q96.4	Mosaicism, 45,X/other cell line(s) with abnormal sex chromosome		
Q96.8	Other variants of Turner's syndrome		
Q96.9	Turner's syndrome, unspecified		
Q97	Other sex chromosome abnormalities, female phenotype, not elsewhere classified		
Q97.0	Karyotype 47,XXX		
Q97.1	Female with more than three X chromosomes		
Q97.2	Mosaicism, lines with various numbers of X chromosomes		
Q97.3	Female with 46,XY karyotype		
	<i>Excludes:</i> Drash syndrome (N07)		
Q97.8	Other specified sex chromosome abnormalities, female phenotype		
Q97.9	Sex chromosome abnormality, female phenotype, unspecified		
Q98	Other sex chromosome abnormalities, male phenotype, not elsewhere classified		
Q98.0	Klinefelter's syndrome karyotype 47,XXY		
Q98.1	Klinefelter's syndrome, male with more than two X chromosomes		
Q98.2	Klinefelter's syndrome, male with 46,XX karyotype		
Q98.3	Other male with 46,XX karyotype		
Q98.4	Klinefelter's syndrome, unspecified		
Q98.5	Karyotype 47,XYY		
Q98.6	Male with structurally abnormal sex chromosome		
Q98.7	Male with sex chromosome mosaicism		
Q98.8	Other specified sex chromosome abnormalities, male phenotype		
Q98.9	Sex chromosome abnormality, male phenotype, unspecified		
Q99	Other chromosome abnormalities, not elsewhere classified		
Q99.0	Chimera 46,XX/46,XY		
			Chimera 46,XX/46,XY true hermaphrodite
		Q99.1	46,XX true hermaphrodite
			46,XX with streak gonads
			46,XY with streak gonads
			Pure gonadal dysgenesis
		Q99.2	Fragile X chromosome
			Fragile X syndrome
		Q99.8	Other specified chromosome abnormalities
		Q99.9	Chromosomal abnormality, unspecified