

第十七章 先天性畸形、變形與染色體異常 (Q00-Q99)

Chapter XVII Congenital malformations, deformations and chromosomal abnormalities (Q00-Q99)

排除：先天性(新陳)代謝(性)缺陷(障礙、異常、偏差) (E70-E90)

Excludes: inborn errors of metabolism (E70-E90)

本章包含下列各節(群組)：

This chapter contains the following blocks:

Q00-Q07 神經系統先天性畸形

Q00-Q07 Congenital malformations of the nervous system

Q10-Q18 眼、耳、顏面和頸的先天性畸形

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

Q20-Q28 循環系統先天性畸形

Q20-Q28 Congenital malformations of the circulatory system

Q30-Q34 呼吸系統先天性畸形

Q30-Q34 Congenital malformations of the respiratory system

Q35-Q37 唇裂和腭裂

Q35-Q37 Cleft lip and cleft palate

Q38-Q45 消化系統其他先天性畸形

Q38-Q45 Other congenital malformations of the digestive system

Q50-Q56 生殖器官先天性畸形

Q50-Q56 Congenital malformations of genital organs

Q60-Q64 泌尿系統先天性畸形

Q60-Q64 Congenital malformations of the urinary system

Q65-Q79 肌肉骨骼系統先天性畸形和變形

Q65-Q79 Congenital malformations and deformations of the musculoskeletal system

Q80-Q89 其他先天性畸形

Q80-Q89 Other congenital malformations

Q90-Q99 染色體異常，他處未歸類者

Q90-Q99 Chromosomal abnormalities, not elsewhere classified

神經系統先天性畸形 (Q00-Q07)

Congenital malformations of the nervous system (Q00-Q07)

Q00 無腦症及類似畸形

Q00 Anencephaly and similar malformations

Q00.0 無腦(症、畸形)

Q00.0 Anencephaly

無頭(腦)(症、畸形)

Acephaly

無顱(蓋)(症、畸形)

Acrania

無腦脊髓(症、畸形)

Amyelencephaly

半側無頭(腦)(症、畸形)

半頭(腦)(症、畸形)

Q00.1 顱脊柱裂(症、畸形)

Q00.2 枕骨裂露腦(症、畸形)

Q01 腦膨出(症、畸形)

包含：腦脊髓膨出(症、畸形)

積水性(水囊)腦膨出(症、畸形)

顱積水性(水囊)腦膜膨出(症、畸形)

腦膜膨出(症、畸形)，(大)腦的

腦膜腦膨出(症、畸形)

排除：Meckel-Gruber (二氏)症候群
(Q61.9)

Q01.0 額部腦膨出(症、畸形)

Q01.1 鼻額部腦膨出(症、畸形)

Q01.2 枕部腦膨出(症、畸形)

Q01.8 其他部位腦膨出(症、畸形)

Q01.9 腦膨出(症、畸形)，未特定者

Q02 小頭(症、畸形)

積水性小頭(症、畸形)

腦小症(畸形)

排除：Meckel-Gruber (二氏)症候群
(Q61.9)

Q03 先天性水腦(症、畸形)

包含：新生兒水腦(症、畸形)

排除：Arnold-Chiari (二氏)症候群
(Q07.0)

水腦(症、畸形)：

- 後天性 (G91.-)
- 先天性弓蟲(病)症所致者
(P37.1)
- 伴有(合併或併發)脊椎(柱)
裂 (Q05.0-Q05.4)

Q03.0 Sylvius (氏)腦導水管(症、畸形)

(下列)腦導水管：

- 異常(畸形)
- 阻塞，先天性

Hemianencephaly

Hemicephaly

Q00.1 Craniorachischisis

Q00.2 Iniencephaly

Q01 Encephalocele

Includes: encephalomyelocele

hydroencephalocele

hydromeningocele, cranial

meningocele, cerebral

meningoencephalocele

Excludes: Meckel-Gruber syndrome(Q61.9)

Q01.0 Frontal encephalocele

Q01.1 Nasofrontal encephalocele

Q01.2 Occipital encephalocele

Q01.8 Encephalocele of other sites

Q01.9 Encephalocele, unspecified

Q02 Microcephaly

Hydromicrocephaly

Micrencephalon

Excludes: Meckel-Gruber syndrome (Q61.9)

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

• 狹窄

**Q03.1 Magendie (氏)孔及 Luschka (氏)孔
閉鎖(第 IV 腦室內側孔及外側孔閉
鎖)**

Dandy-Walk (二氏)症候群

Q03.8 其他先天性水腦(症、畸形)

Q03.9 先天性水腦(症、畸形)，未特定者

Q04 腦其他先天性畸形

排除：獨眼(症、畸形) (Q87.0)

巨頭(症、畸形) (Q75.3)

Q04.0 胼胝體先天性畸形

胼胝體生成(發育)不良(症、畸形)

Q04.1 無嗅腦(症、畸形)

Q04.2 全前腦(症、畸形)

Q04.3 腦其他短缺畸形

部份腦 { 缺損
生成或發育不良
發育不良(萎縮)
發育不全

無腦回(症、畸形)

積水性無腦(症、畸形)

平腦(症、畸形)(腦回發育不全)

小腦(症、畸形)

腦回肥厚(症、畸形)

排除：胼胝體先天性畸形 (Q04.0)

**Q04.4 中隔 - 眼的發育不良(症、畸形)(腦
中隔)**

Q04.5 巨腦(症、畸形)

Q04.6 先天性大腦囊腫(症、畸形)

孔洞腦(症、畸形)

裂腦(症、畸形)

排除：後天性孔洞腦畸形囊腫(症、畸形)
(G93.0)

Q04.8 腦其他特定的先天性畸形

巨腦回(症、畸形)

• stenosis

**Q03.1 Atresia of foramina of Magendie and
Luschka**

Dandy-Walker syndrome

Q03.8 Other congenital hydrocephalus

Q03.9 Congenital hydrocephalus, unspecified

**Q04 Other congenital malformations of
brain**

Excludes: cyclopia (Q87.0)

macrocephaly (Q75.3)

**Q04.0 Congenital malformations of corpus
callosum**

Agenesis of corpus callosum

Q04.1 Arrhinencephaly

Q04.2 Holoprosencephaly

Q04.3 Other reduction deformities of brain

Absence
Agenesis
Aplasia
Hypoplasia } of part of brain

Agyria

Hydranencephaly

Lissencephaly

Microgyria

Pachygyria

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

Q04.4 Septo-optic dysplasia

Q04.5 Megalencephaly

Q04.6 Congenital cerebral cysts

Porencephaly

Schizencephaly

Excludes: acquired porencephalic cyst (G93.0)

**Q04.8 Other specified congenital malformations
of brain**

Macrogyria

Q04.9 腦先天性畸形，未特定者

先天性：

腦其他未特定的

- 異常(畸形)
- 變形
- 疾病或病灶
- 多重異常(畸形)

Q04.9 Congenital malformation of brain, unspecified

Congenital:

• anomaly
• deformity
• disease or lesion
• multiple anomalies

NOS of brain

Q05 脊椎(柱)裂

包 含：(脊椎的)積水性腦脊膜膨出(症、畸形)

(脊椎的)腦脊膜膨出(症、畸形)

脊膜髓膨出(症、畸形)

脊髓膨出(症、畸形)

脊髓膜膨出(症、畸形)

脊柱裂

(開口性)(囊腫性)脊椎(柱)裂

脊髓中央管膨出(症、畸形)

排 除：Arnold-Chiari (二氏)症候群 (Q07.0)

隱性脊椎(柱)裂(症、畸形)

(Q76.0)

Q05 Spina bifida

Includes: hydromeningocele(spinal)

meningocele (spinal)

meningomyelocele

myelocele

myelomeningocele

rachischisis

spina bifida (aperta)(cystica)

syringomyelocele

Excludes: Arnold-Chiari syndrome (Q07.0)

spina bifida occulta (Q76.0)

Q05.0 頸椎裂伴有(合併或併發)水腦(症、畸形)

Q05.0 Cervical spina bifida with hydrocephalus

Q05.1 胸椎裂伴有(合併或併發)水腦(症、畸形)

Q05.1 Thoracic spina bifida with hydrocephalus

下列脊椎(柱)裂：

• 背(部) } 伴有(合併或併發)
• 胸腰(部) } 水腦(症、畸形)

Spina bifida:

• dorsal } with hydrocephalus
• thoracolumbar }

Q05.2 腰椎裂伴有(合併或併發)水腦(症、畸形)

Q05.2 Lumbar spina bifida with hydrocephalus

腰薦椎裂伴有(合併或併發)水腦(症、畸形)

Lumbosacral spina bifida with hydrocephalus

Q05.3 薦椎裂伴有(合併或併發)水腦(症、畸形)

Q05.3 Sacral spina bifida with hydrocephalus

Q05.4 未特定的脊椎裂伴有(合併或併發)水腦(症、畸形)

Q05.4 Unspecified spina bifida with hydrocephalus

Q05.5 頸椎裂未伴有(未合併或未併發)水腦(症、畸形)

Q05.5 Cervical spina bifida without hydrocephalus

Q05.6 胸椎裂未伴有(未合併或未併發)水腦(症、畸形)

Q05.6 Thoracic spina bifida without hydrocephalus

(下列)脊椎(柱)裂：

Spina bifida:

- 背(部)，其他未特定者
- 胸腰(部)，其他未特定者

Q05.7 腰椎裂未伴有(未合併或未併發)水腦(症、畸形)

腰薦椎裂，其他未特定者

Q05.8 薦椎裂未伴有(未合併或未併發)水腦(症、畸形)

Q05.9 脊椎(柱)裂，未特定者

- dorsal NOS
- thoracolumbar 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 脊髓其他先天性畸形

Q06.0 無脊髓(症、畸形)

Q06.1 脊髓發育不全及發育不良

脊髓發育不全

脊髓發育不全

脊髓發育不良

Q06.2 脊髓裂

Q06.3 其他先天性脊髓尾畸形

Q06.4 脊髓積水

椎管積水

Q06.8 脊髓其他特定的先天性畸形

Q06.9 脊髓先天性畸形，未特定者

(下列)先天性：

脊髓或腦(脊髓)膜
其他未特定的

- 異常(畸形)
- 變形
- 疾病或病灶

Q06 Other congenital malformations of spinal cord

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 malformation of spinal cord, unspecified

Congenital:

- anomaly
- deformity
- disease or lesion

NOS of spinal
cord or meninges

Q07 神經系統其他先天性畸形

排除：家族性自主神經機(功)能不良(障礙、失調、異常) [Riley-Day
(二氏)] (G90.1)
(非惡性)神經纖維瘤症 (Q85.0)

Q07.0 Arnold-Chiari (二氏)症候群

Q07.8 神經系統其他特定的先天性畸形

神經生成(發育)不良

臂神經叢移位

頷(顎)動性眨眼症候群

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

Agensis of nerve

Displacement of brachial plexus

Jaw-winking syndrome

Q07.9 神經系統先天性畸形，未特定者

先天性：

神經系統其 他未特定的	{	<ul style="list-style-type: none"> • 異常(畸形) • 變形 • 疾病或病灶
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Q07.8 Congenital malformation of nervous system, unspecified

Congenital:

<ul style="list-style-type: none"> • anomaly • deformity • disease or lesion 	}	NOS of nervous system
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眼、耳、顏面和頸的先天性畸形 (Q10-Q18)

排除：唇裂及腭裂 (Q35-Q37)

下列先天性畸形：

- 頸椎 (Q05.0, Q05.5, Q67.5, Q76.0-Q76.4)
- 喉 (Q31.-)
- 唇，他處未歸類者 (Q31.-)
- 鼻 (Q30.-)
- 副甲狀腺 (Q89.2)
- 甲狀腺 (Q89.2)

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

Excludes : cleft lip and cleft palate (Q35-Q37)

congenital malformation 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)

Q10 眼瞼、淚器及眼窩先天性畸形

排除：(下列)隱眼畸形：

- 其他未特定者 (Q11.2)
- 症候群 (Q87.0)

Q10 Congenital malformations of eyelid, lacrimal apparatus and orbit

Excludes: cryptophthalmos:

- NOS (Q11.2)
- syndrome (Q87.0)

Q10.0 先天性眼瞼下垂

Q10.1 先天性眼瞼外翻

Q10.2 先天性眼瞼內翻

Q10.3 眼瞼其他先天性畸形

眼瞼缺損[無眼瞼](症)

下列缺損或生成(或發育)不良：

- 睫毛
- 眼瞼

副(的)：

- 眼瞼
- 眼肌

眼裂狹小[眼瞼閉鎖](症)，先天性

眼瞼缺損[無眼瞼](症)

眼瞼先天性畸形，其他未特定者

Q10.0 Congenital ptosis

Q10.1 Congenital ectropion

Q10.2 Congenital entropion

Q10.3 Other congenital malformations of eyelid

Ablepharon

Absence of agenesis of:

- cilia
- eyelid

Accessory:

- eyelid
- eye muscle

Blepharophimosis, congenital

Coloboma of eyelid

Congenital malformation of eyelid NOS

Q10.4 淚器缺損及生成(或發育)不良

淚點缺損(症)

Q10.5 淚管先天性狹窄及縮窄(症、畸形)

Q10.6 淚器先天性畸形

淚器先天性畸形，其他未特定者

Q10.7 眼窩先天性畸形

Q11 無眼、小眼及巨眼(症、畸形)

Q11.0 囊腫性眼球(症、畸形)

Q11.1 其他無眼(症、畸形)

眼的 { 生成(或發育)不良
發育不良(萎縮)

Q11.2 小眼(症、畸形)，其他未特定者

隱眼(症、畸形)

眼發育不良

眼發育不全

未成熟之眼

排除：隱眼(症、畸形)症候群 (Q87.0)

Q11.3 巨眼畸形

排除：先天性青光眼(所致)的巨眼(症、畸形) (Q15.0)

Q12 先天性水晶體畸形

Q12.0 先天性白內障

Q12.1 先天性水晶體移位(症、畸形)

Q12.2 水晶體缺損(症、畸形)

Q12.3 先天性無水晶體(症、畸形)

Q12.4 球形水晶體(症、畸形)

Q12.8 其他先天性水晶體畸形

Q12.9 先天性水晶體畸形，未特定者

Q13 前段眼先天性畸形

Q13.0 虹膜缺損

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 malformation of lacrimal apparatus
NOS

Q10.7 Congenital malformation of orbit

Q11 Anophthalmos, microphthalmos and macropthalmos

Q11.0 Cystic eyeball

Q11.1 Other anophthalmos

Agenesis }
Aplasia } of eye

Q11.2 Microphthalmos

Cryptophthalmos NOS

Dysplasia of eye

Hypoplasia of eye

Rudimentary eye

Excludes: cryptophthalmos syndrome (Q87.0)

Q11.3 Macropthalmos

Excludes: macropthalmos in congenital glaucoma (Q15.0)

Q12 Congenital lens malformations

Q12.0 Congenital cataract

Q12.1 Congenital displaced lens

Q12.2 Coloboma of lens

Q12.3 Congenital aphakia

Q12.4 Spherophakia

Q12.8 Other congenital lens malformations

Q12.9 Congenital lens malformation, unspecified

Q13 Congenital malformations of anterior segment of eye

Q13.0 Coloboma of iris

缺損，其他未特定者

Q13.1 無虹膜[虹膜缺損]

無虹膜[虹膜缺損]

Q13.2 虹膜其他先天性畸形

瞳孔(大小)不一，先天性

瞳孔閉鎖(症、畸形)

先天性虹膜畸形，其他未特定者

瞳孔異位(症、畸形)

Q13.3 先天性角膜混濁(症)

Q13.4 其他先天性角膜畸形

角膜先天性畸形，其他未特定者

小角膜(症、畸形)

Peter (氏)異常(畸形)

Q13.5 藍鞏膜(症)

Q13.8 前段眼其他先天性畸形

Rieger (氏)異常(畸形)

Q13.9 前段眼先天性畸形，未特定者

Q14 後段眼先天性畸形

Q14.0 玻璃體先天性畸形

先天性玻璃體混濁(症)

Q14.1 視網膜先天性畸形

先天性視網膜動脈瘤

Q14.2 視盤先天性畸形

視盤缺損

Q14.3 脈絡膜先天性畸形

Q14.8 後段眼其他先天性畸形

眼底缺損(症、畸形)

Q14.9 後段眼先天性畸形，未特定者

Q15 眼其他先天性畸形

排除：先天性眼球震顫 (H55)

眼白化症 (E70.3)

色素性視網膜炎 (H35.5)

Coloboma NOS

Q13.1 Absence of iris

Aniridia

Q13.2 Other congenital malformations of iris

Anisocoria, congenital

Atresia of pupil

Congenital malformation of iris NOS

Corectopia

Q13.3 Congenital corneal opacity

Q13.4 Other congenital corneal malformations

Congenital malformation of cornea NOS

Microcornea

Peter's anomaly

Q13.5 Blue sclera

Q13.8 Other congenital malformations of anterior segment of eye

Rieger's anomaly

Q13.9 Congenital malformation of anterior segment of eye, unspecified

Q14 Congenital malformations of posterior segment of eye

Q14.0 Congenital malformation of vitreous humour

Congenital vitreous opacity

Q14.1 Congenital malformation of retina

Congenital retinal aneurysm

Q14.2 Congenital malformation of optic disc

Coloboma of optic disc

Q14.3 Congenital malformation of choroid

Q14.8 Other congenital malformations of posterior segment of eye

Coloboma of fundus

Q14.9 Congenital malformation of posterior segment of eye, unspecified

Q15 Other congenital malformations of eye

Excludes: congenital nystagmus (H55)

ocular albinism (E70.3)

retinitis pigmentosa (H35.5)

Q15.0 先天性青光眼

牛眼症[先天性青光眼]
 新生兒青光眼
 眼水腫[眼積水]
 角膜球(狀)突，先天性
 先天性青光眼(所致)的巨眼(症、畸形)
 巨(球形)角膜(症、畸形)

Q15.8 眼其他特定的先天性畸形**Q15.9 眼先天性畸形，未特定者**

(下列)先天性：

眼其他未特定的 { • 異常(畸形)
 • 變形

Q16 導致聽覺(力)損傷的耳先天性畸形

~~排除~~：先天性耳聾 (H90.-)

Q16.0 先天性耳廓缺損[無耳廓]**Q16.1 (外)耳道先天性缺損[無(外)耳道]、閉鎖及縮窄**

骨性耳道閉鎖或縮窄

Q16.2 耳咽(鼓)管缺損[無耳咽(鼓)管]**Q16.3 聽小骨先天性畸形**

聽小骨融合

Q16.4 中耳其他先天性畸形

中耳先天性畸形，其他未特定者

Q16.5 內耳先天性畸形

(下列)異常(畸形)：

- 膜性迷路
- Corti (氏)器

Q16.9 導致聽覺(力)損傷的耳先天性畸形，未特定者

先天性耳缺損(無耳症)，其他未特定者

Q17 其他耳先天性畸形

~~排除~~：耳前竇 (Q18.1)

Q17.0 副耳(廓)

副耳屏(珠)

Q15.0 Congenital glaucoma

Buphthalmos
 Glaucoma of newborn
 Hydrophthalmos
 Keratoglobus, congenital
 Macrophthalmos in congenital glaucoma
 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 impairment of hearing

Excludes: congenital deafness (H90.-)

Q16.0 Congenital absence of (ear) auricle**Q16.1 Congenital absence, atresia and stricture of auditory canal (external)**

Atresia 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 malformation of middle ear NOS

Q16.5 Congenital malformation of inner ear

Anomaly:

- 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

多耳(症、畸形)
耳前附件或耳贅物
贅餘(的):
• 耳
• 耳垂

Polyotia
Preauricular appendage or tag
Supernumerary:
• ear
• lobule

Q17.1 巨耳(症、畸形)

Q17.2 小耳(症、畸形)

Q17.3 其他造形異常(畸形)耳

尖耳(症、畸形)

Q17.4 錯置(移位)耳

低位耳

排除: 頸部耳(狀)廓 (Q18.2)

Q17.5 招風耳(凸耳)

蝙蝠耳(招風耳)

Q17.8 其他特定的耳先天性畸形

耳垂先天性缺損[無耳垂]

Q17.9 耳先天性畸形，未特定者

耳先天性異常(畸形)，其他未特定者

Q17.1 Macrotia

Q17.2 Microtia

Q17.3 Other misshapen ear

Pointed 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

Congenital absence of lobe of ear

Q17.9 Congenital malformation of ear, unspecified

Congenital anomaly of ear NOS

Q18 顏面及頸其他先天性畸形

排除: 唇裂及腭裂 (Q35-Q37)

可歸類於 **Q67.0 - Q67.4** 的病況

顱骨及顏面骨先天性畸形 (Q75.-)

獨眼(症、畸形) (Q87.0)

齒(顏)面異常(包含咬合不正)
(K07.-)

影響顏面外觀的畸形症候群
(Q87.0)

甲(狀)舌管持續性殘留
(Q89.2)

Q18.0 鰓裂竇、瘻管及囊腫

鰓遺跡

Q18.1 耳前竇及囊腫

(下列)瘻管:

- 耳廓(瘻)，先天性
- 頸耳(瘻)

Q18 Other congenital malformations of face and neck

Excludes: cleft lip and cleft palate (Q35-Q37)

conditions classified to Q67.0-Q67.4

congenital malformations of skull
and face bones (Q75.-)

cyclopia (Q87.0)

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 其他鰓裂畸形

鰓裂畸形，其他未特定者

頸部耳(狀)廓

頭耳(併下頷(顎))畸形

Q18.3 頸蹼(畸形)

翼狀頸(皮)

Q18.4 巨口(症、畸形)

Q18.5 小口(症、畸形)

Q18.6 巨唇(症、畸形)

唇肥厚(肥大、增厚)，先天性

Q18.7 小唇(症、畸形)

Q18.8 顏面及頸其他特定的先天性畸形

內側的：

顏面及頸

- 囊腫
- 瘻管
- 竇

Q18.9 顏面及頸先天性畸形，未特定者

顏面及頸其他未特定的先天性異常(畸形)

Q18.2 Other branchial cleft malformations

Branchial cleft malformation 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
• sinus

} of face and neck

Q18.9 Congenital malformation of face and neck, unspecified

Congenital anomaly NOS of face and neck

循環系統先天性畸形 (Q20-Q28)

Q20 心臟腔室[房室]及連接處的先天性畸形

排除：右位心伴有(或合併)內臟反位
(Q89.3)

心房鏡像排列伴有(或合併)內臟
反位 (Q89.3)

Q20.0 共同動脈幹

持續性動脈幹殘留

Q20.1 右心室雙出口

Taussig-Bing (二氏)症候群

Q20.2 左心室雙出口

Q20.3 心室動脈連結不協調

主動脈右轉位

(完全的)大血管錯(轉)位

Q20.4 心室雙入口

共同心室

Congenital malformations of the circulatory system (Q20-Q28)

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

兩房一室的三腔室心臟
單一心室

Q20.5 不一致的房室連結

修正的錯(轉)位
左轉位
心室反向(內翻)

Q20.6 心房附件異構

伴有(合併或併發)無脾或多脾的心房附件
異構

Q20.8 心臟腔室[房室]及連接處的其他先天性畸形

Q20.9 心臟腔室[房室]及連接處的先天性畸形，未特定者

Cor triloculare biatriatum
Single ventricle

Q20.5 Discordant atrioventricular connection

Corrected transposition
Laevotransposition
Ventricular inversion

Q20.6 Isomerism of atrial appendages

Isomerism of atrial appendages with asplenia or polysplenia

Q20.8 Other congenital malformations of cardiac chambers and connections

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

Q21 心臟中隔先天性畸形

排除：後天性心臟中隔缺陷(損) (I51.0)

Q21.0 心室中隔缺陷(損)

Q21.1 心房中隔缺陷(損)

冠狀竇缺陷(損)
未關閉或持續性殘留的：
• 卵圓孔
• 第二孔心房中隔缺陷(損) (II 型)
靜脈竇缺陷(損)

Q21.2 房室中隔缺陷(損)

共同房室通道
心內膜墊缺陷(損)
第一孔心房中隔缺陷(損) (I 型)

Q21.3 法洛(Fallot)(氏)四重症

心室中隔缺陷(損)伴有(合併或併發)肺動脈狹窄或閉鎖、主動脈右跨位及右心室肥大(增大、肥厚)

Q21.4 主動脈及肺動脈中隔缺陷(損)

主動脈中隔缺陷(損)
主動脈肺動脈窗

Q21.8 心臟中隔其他先天性畸形

Eisenmenger (氏)症候群
法洛(Fallot)(氏)五重症

Q21 Congenital malformations of cardiac septa

Excludes: acquired cardiac septal defect (I51.0)

Q21.0 Ventricular septal defect

Q21.1 Atrial septal defect

Coronary sinus defect
Patent or persistent:
• foramen ovale
• ostium secundum defect (type II)
Sinus venosus defect

Q21.2 Atrioventricular septal defect

Common atrioventricular canal
Endocardial cushion defect
Ostium primum atrial septal defect (type I)

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

Eisenmenger's syndrome
Pentalogy of Fallot

Q21.9 心臟中隔先天性畸形，未特定者

(心臟)中隔缺陷(損)，其他未特定者

Q22 肺動脈瓣及三尖瓣先天性畸形

Q22.0 肺動脈瓣閉鎖

Q22.1 先天性肺動脈瓣狹窄

Q22.2 先天性肺動脈瓣機(功)能不全

先天性肺動脈瓣閉鎖不全(回流)

Q22.3 肺動脈瓣其他先天性畸形

肺動脈瓣先天性畸形，其他未特定者

Q22.4 先天性三尖瓣狹窄

三尖瓣閉鎖

Q22.5 Ebstein (氏)異常(畸形)

Q22.6 右心發育不全症候群

Q22.8 三尖瓣其他先天性畸形

Q22.9 三尖瓣先天性畸形，未特定者

Q23 主動脈瓣及二尖瓣先天性畸形

Q23.0 先天性主動脈瓣狹窄

先天性主動脈的：

- 閉鎖
- 狹窄

排除：先天性主動脈瓣下狹窄 (Q24.4)

於左心發育不全症候群者

(Q23.4)

Q23.1 先天性主動脈瓣機(功)能不全

二尖(瓣)性主動脈瓣

先天性主動脈機(功)能不全

Q23.2 先天性二尖瓣狹窄

先天性二尖瓣閉鎖

Q23.3 先天性二尖瓣機(功)能不全

Q23.4 左心發育不全症候群

主動脈開口或瓣膜閉鎖或明顯發育不全，且伴有(合併或併發)升主動脈發育不全及左心室發育不全(伴有或合併或併發二尖瓣狹窄或閉鎖)。

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

Excludes: congenital subaortic stenosis (Q24.4) that in hypoplastic left heart syndrome (Q23.4)

Q23.1 Congenital insufficiency of aortic valve

Bicuspid aortic valve

Congenital aortic insufficiency

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 主動脈及二尖瓣其他先天性畸形

Q23.9 主動脈及二尖瓣先天性畸形，未特定者

Q24 心臟其他先天性畸形

排除：心內膜纖維彈性增生(纖維變性) (I42.4)

Q24.0 右位心

排除：右位心伴有(合併或併發)內臟反位 (Q89.3)

心房附件異構(伴有或合併或併發無脾或多脾) (Q20.6)

鏡像心房排列伴有(合併或併發)內臟反位 (Q89.3)

Q24.1 左位心

Q24.2 三腔室心臟

Q24.3 肺動脈漏斗狀狹窄

Q24.4 先天性主動脈瓣下狹窄

Q24.5 冠狀血管畸形

先天性冠狀(動脈)動脈瘤

Q24.6 先天性心臟傳導阻斷

Q24.8 心臟其他特定的先天性畸形

(下列)先天性：

• 左心室憩室

• 下列畸形：

• 心肌

• 心包膜

心臟錯位

Uhl (氏)病

Q24.9 心臟先天性畸形，未特定者

先天性：

心臟其他未特定的 { • 異常(畸形)
• 疾病

Q25 大動脈先天性畸形

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: endocardial fibroelastosis (I42.4)

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 heart

Congenital:

• diverticulum of left ventricle

• malformation of:

• myocardium

• pericardium

Malposition of heart

Uhl's disease

Q24.9 Congenital malformation of heart, unspecified

Congenital:

• anomaly

• disease

} NOS of heart

Q25 Congenital malformations of great arteries

Q25.0 開放性動脈導管

開放性 Botallo (氏)導管

持續性動脈導管殘留

Q25.1 主動脈縮窄

(導管前)(導管後)主動脈窄縮

Q25.2 主動脈閉鎖**Q25.3 主動脈狹窄**

瓣膜上主動脈狹窄

排除：先天性主動脈狹窄 (Q23.0)

Q25.4 主動脈其他先天性畸形

主動脈的 { 缺損
發育不良(萎縮)
先天性：
• 動脈瘤
• 擴張

主動脈竇[Valsalva 竇]動脈瘤(破裂)

雙主動脈弓[主動脈血管環]

主動脈發育不全

持續性：

• 主動脈弓捲曲

• 右主動脈弓

排除：左心發育不全症候群(所致)的
主動脈發育不全 (Q23.4)

Q25.5 肺動脈閉鎖**Q25.6 肺動脈狹窄****Q25.7 肺動脈其他先天性畸形**

迷走(畸形)肺動脈

{ 生成或發育不良
動脈瘤
異常(畸形)
發育不全

肺動靜脈血管瘤

Q25.8 大動脈其他先天性畸形**Q25.9 大動脈先天性畸形，未特定者****Q25.0 Patent ductus arteriosus**

Patent ductus Botallo

Persistent ductus arteriosus

Q25.1 Coarctation of aorta

Coarctation of aorta (preductal)(postductal)

Q25.2 Atresia of aorta**Q25.3 Stenosis of aorta**

Supravalvular aortic stenosis

Excludes: congenital aortic stenosis (Q23.0)

Q25.4 Other congenital malformations of aorta

Absence
Aplasia
Congenital:
• aneurysm
• dilatation } of aorta

Aneurysm of sinus of Valsalva (ruptured)

Double aortic arch [vascular ring of aorta]

Hypoplasia of aorta

Persistent:

• convolutions of aortic arch

• right aortic arch

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

Q25.5 Atresia of pulmonary artery**Q25.6 Stenosis of pulmonary artery****Q25.7 Other congenital malformations of pulmonary artery**

Aberrant pulmonary artery

Agenesis
Aneurysm
Anomaly
Hypoplasia } of pulmonary artery

Pulmonary arteriovenous aneurysm

Q25.8 Other congenital malformations of great arteries**Q25.9 Congenital malformation of great arteries, unspecified**

Q26 大靜脈先天性畸形

Q26.0 腔靜脈先天性狹窄

(上)(下)腔靜脈先天性狹窄

Q26.1 持續性左上腔靜脈殘留

Q26.2 肺靜脈連接處全部異常

Q26.3 肺靜脈連接處部份異常

Q26.4 肺靜脈連接處異常，未特定者

Q26.5 門靜脈連接處異常

Q26.6 門靜脈－肝動脈瘻管

Q26.8 大靜脈其他先天性畸形

(下)(上)腔靜脈缺損[無(下)(上)腔靜脈]

下腔靜脈的奇靜脈延伸

左後主靜脈持續性殘留

彎刀症候群

Q26.9 大靜脈先天性畸形，未特定者

(下)(上)腔靜脈異常(畸形)，其他未特定者

Q27 周邊血管系統其他先天性畸形

排除：下列異常(畸形)：

- (大)腦及腦前血管
(Q28.0-Q28.3)
- 冠狀血管 (Q24.5)
- 肺動脈 (Q25.5-Q25.7)
- 先天性視網膜動脈瘤 (Q14.1)
- 血管瘤及淋巴管瘤 (D18.-)

Q27.0 先天性臍動脈缺損及發育不全

單一臍動脈

Q27.1 先天性腎動脈狹窄

Q27.2 腎動脈其他先天性畸形

腎動脈先天性畸形，其他未特定者

多腎動脈

Q27.3 周邊動靜脈畸形

Q26 Congenital malformations of great veins

Q26.0 Congenital stenosis of vena cava

Congenital stenosis of vena cava (inferior)
(superior)

Q26.1 Persistent left superior vena cava

Q26.2 Total anomalous pulmonary venous connection

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

Excludes: 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.-)

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

動靜脈瘤

排除：後天性動靜脈瘤 (I77.0)

Q27.4 先天性靜脈擴張

Q27.8 周邊血管系統其他特定的先天性畸形

迷走(畸形)鎖骨下動脈

他處未歸類的動脈或靜脈

缺損
閉鎖

先天性：

- (周邊的)動脈瘤
- 縮窄，動脈
- 血管曲張

Q27.9 周邊血管系統先天性畸形，未特定者

動脈或靜脈異常(畸形)，其他未特定者

Q28 循環系統其他先天性畸形

排除：下列先天性動脈瘤：

- 其他未特定者 (Q27.8)
- 冠狀動脈的 (Q24.5)
- 周邊動脈的 (Q27.8)
- 肺動脈的 (Q25.7)
- 視網膜的 (Q14.1)

破裂的：

- 腦動靜脈畸形 (I60.8)
- 腦前血管畸形 (I72.-)

Q28.0 腦前血管動靜脈畸形

先天性腦前(未破裂的)動靜脈瘤

Q28.1 腦前血管其他畸形

先天性：

- 腦前血管畸形，其他未特定者
- 腦前動脈瘤(未破裂者)

Q28.2 腦血管動靜脈畸形

腦動靜脈畸形，其他未特定者

Arteriovenous aneurysm

Excludes: acquired arteriovenous aneurysm (I77.0)

Q27.4 Congenital phlebectasia

Q27.8 Other specified congenital malformations of peripheral vascular system

Aberrant subclavian artery

Absence
Atresia

} of artery or vein NEC

Congenital:

- aneurysm (peripheral)
- stricture, artery
- varix

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)

ruptured:

- cerebral arteriovenous malformation (I60.8)
- malformation of precerebral vessels (I72.-)

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

先天性腦動靜脈瘤(未破裂者)

Q28.3 大腦血管其他畸形

先天性：

- 腦動脈瘤(未破裂者)
- 腦血管畸形，其他未特定者

Q28.8 循環系統其他特定的先天性畸形

先天性動脈瘤，特定部位且他處未歸類者

Q28.9 循環系統先天性畸形，未特定者

Congenital arteriovenous cerebral aneurysm
(nonruptured)

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

Q28.9 Congenital malformation of circulatory system, unspecified

呼吸系統先天性畸形 (Q30-Q34)

Q30 鼻先天性畸形

~~排除~~：先天性鼻中隔偏曲 (Q67.4)

Q30.0 後鼻孔閉鎖(症)

(前)(後)鼻孔的 $\left\{ \begin{array}{l} \text{閉鎖} \\ \text{先天性狹窄} \end{array} \right.$

Q30.1 鼻生成不良及發育不全

先天性鼻缺損[無鼻]

Q30.2 鼻裂隙、切迹及裂開

Q30.3 先天性鼻中隔穿孔

Q30.8 其他先天性鼻畸形

副鼻

鼻竇壁先天性異常(畸形)

Q30.9 鼻先天性畸形，未特定者

Congenital malformations of the respiratory system(Q30-Q34)

Q30 Congenital malformations of nose

Excludes: congenital deviation of nasal septum
(Q67.4)

Q30.0 Choanal atresia

Atresia $\left\{ \begin{array}{l} \text{of nares (anterior)} \\ \text{Congenital stenosis (posterior)} \end{array} \right.$

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 喉先天性畸形

Q31.0 喉蹼

下列喉蹼：

- 其他未特定者
- 聲門(的)
- 聲門下(的)

Q31.1 先天性聲門下狹窄

Q31.2 喉發育不全

Q31.3 喉膨出

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 先天性喉喘鳴

(喉)先天性喘鳴，其他未特定者

Q31.8 喉其他先天性畸形

環狀軟骨、會厭、
聲門、喉或甲狀
軟骨的

缺損
生成(發育)不良
閉鎖

甲狀軟骨裂

喉先天性狹窄，他處未歸類者

會厭裂隙

環狀軟骨後裂

Q31.9 喉先天性畸形，未特定者

Q32 氣管及支氣管先天性畸形

~~排除~~ 先天性支氣管擴張症 (Q33.4)

Q32.0 先天性氣管軟化

Q32.1 氣管其他先天性畸形

氣管軟骨異常(畸形)

氣管閉鎖

(下列)先天性：

氣管的

• 擴張
• 畸形
• 狹窄

• 氣管膨出

Q32.2 先天性支氣管軟化

Q32.3 支氣管先天性狹窄

Q32.4 支氣管其他先天性畸形

支氣管的

缺損
生成(發育)不良
閉鎖
先天性畸形，其他未特定者
憩室

Q33 肺先天性畸形

Q33.0 先天性肺囊腫

(下列)先天性：

• 蜂巢肺

Q31.4 Congenital laryngeal stridor

Congenital stridor (larynx) NOS

Q31.8 Other congenital malformations of larynx

Absence
Agenesis
Atresia

of cricoid cartilage,
epiglottis, glottis, larynx
or thyroid cartilage

Cleft thyroid cartilage

Congenital stenosis of larynx NEC

Fissure of epiglottis

Posterior cleft of cricoid cartilage

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
• stenosis

of trachea

• tracheocele

Q32.2 Congenital bronchomalacia

Q32.3 Congenital stenosis of bronchus

Q32.4 Other congenital malformations of bronchus

Absence
Agenesis
Atresia
Congenital malformation NOS
Diverticulum

of bronchus

Q33 Congenital malformations of lung

Q33.0 Congenital cystic lung

Congenital:

• honeycomb lung

- 肺疾病
- 囊腫性
- 多囊腫性

排除：囊腫性肺疾病，後天或未特定者 (J98.4)

- lung disease
- cystic
- polycystic

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

Q33.1 肺副葉

Q33.2 肺分離

Q33.3 肺生成(發育)不良

肺(葉)缺損[無肺(葉)]

Q33.4 先天性支氣管擴張症

Q33.5 肺中異位組織

Q33.6 肺發育不全及發育不良

排除：妊娠(胎齡)過短相關的肺發育不全 (P28.0)

Q33.8 肺其他先天性畸形

Q33.9 肺先天性畸形，未特定者

Q33.1 Accessory lobe of lung

Q33.2 Sequestration of lung

Q33.3 Agenesis of lung

Abseence 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)

Q33.8 Other congenital malformations of lung

Q33.9 Congenital malformation of lung, unspecified

Q34 呼吸系統其他先天性畸形

Q34.0 肋膜畸形

Q34.1 縱隔先天性囊腫

Q34.8 呼吸系統其他特定的先天性畸形

鼻咽閉鎖

Q34.9 呼吸系統先天性畸形，未特定者

呼吸器官的 { 先天性：
• 缺損
• 異常(畸形)，其他未特定者

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

Q34.9 Congenital malformation of respiratory system, unspecified

Congenital: {
• absence
• anomaly NOS } of respiratory organ

唇裂和腭裂 (Q35-Q37)

排除：Robin (氏)症候群 (Q87.0)

Q35 腭裂

包含：腭裂隙

腭裂畸形

排除：腭裂伴有(合併或併發)唇裂 (Q37.-)

Cleft and cleft palate (Q35-Q37)

Excludes: Robin's syndrome (Q87.0)

Q35 Cleft palate

Includes: fissure of palate

Palatoschisis

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

- Q35.0 硬腭裂，雙側**
Q35.1 硬腭裂，單側
 硬腭裂，其他未特定者
Q35.2 軟腭裂，雙側
Q35.3 軟腭裂，單側
 軟腭裂，其他未特定者
Q35.4 硬腭裂伴有(合併或併發)軟腭裂，雙側
Q35.5 硬腭裂伴有(合併或併發)軟腭裂，單側
 硬腭裂伴有(合併或併發)軟腭裂，其他未特定者
Q35.6 腭裂，內側
Q35.7 懸壅垂裂
Q35.8 腭裂，未特定者，雙側
Q35.9 腭裂，未特定者，單側
 腭裂，其他未特定者

Q36 唇裂

包含：裂唇畸形
 先天性唇裂隙
 兔唇
 兔狀唇

排除：唇裂伴有(合併或併發)腭裂
 (Q37.-)

- Q36.0 唇裂，雙側**
Q36.1 唇裂，中線
Q36.9 唇裂，單側
 唇裂，其他未特定者

Q37 腭裂伴有(合併或併發)唇裂

- Q37.0 硬腭裂伴有(合併或併發)唇裂，雙側**
Q37.1 硬腭裂伴有(合併或併發)唇裂，單側
 硬腭裂伴有(合併或併發)唇裂，其他未特定者
Q37.2 軟腭裂伴有(合併或併發)唇裂，雙側
Q37.3 軟腭裂伴有(合併或併發)唇裂，單側
 軟腭裂伴有(合併或併發)唇裂，其他未特定者
Q37.4 硬腭裂及軟腭裂伴有(合併或併發)唇裂，雙側

- Q35.0 Cleft hard palate, bilateral**
Q35.1 Cleft hard palate, unilateral
 Cleft hard palate NOS
Q35.2 Cleft soft palate, bilateral
Q35.3 Cleft soft palate, unilateral
 Cleft soft palate NOS
Q35.4 Cleft hard palate with cleft soft palate, bilateral
Q35.5 Cleft hard palate with cleft soft palate, unilateral
 Cleft hard palate with cleft soft palate NOS
Q35.6 Cleft palate, medial
Q35.7 Cleft uvula
Q35.8 Cleft palate, unspecified, bilateral
Q35.9 Cleft palate, unspecified, unilateral
 Cleft palate NOS

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, medial
Q36.9 Cleft lip, unilateral
 Cleft lip NOS

Q37 Cleft palate with cleft lip

- Q37.0 Cleft hard palate with cleft lip, bilateral**
Q37.1 Cleft hard palate with cleft lip, unilateral
 Cleft hard palate with cleft lip NOS
Q37.2 Cleft soft palate with cleft lip, bilateral
Q37.3 Cleft soft palate with cleft lip, unilateral
 Cleft soft palate with cleft lip NOS
Q37.4 Cleft hard and soft palate with cleft lip, bilateral

- Q37.5 硬腭裂及軟腭裂伴有(合併或併發)唇裂，單側**
硬腭裂及軟顎裂伴有(合併或併發)唇裂，其他未特定者
- Q37.8 未特定腭裂伴有(合併或併發)唇裂，雙側**
- Q37.9 未特定腭裂伴有(合併或併發)唇裂，單側**
腭裂伴有(合併或併發)唇裂，其他未特定者

- Q37.5 Cleft hard and soft palate with cleft lip, unilateral**
Cleft hard and soft palate with cleft lip NOS
- Q37.8 Unspecified cleft palate with cleft lip, bilateral**
- Q37.9 Unspecified cleft palate with cleft lip, unilateral**
Cleft palate with cleft lip NOS

消化系統其他先天性畸形 (Q38-Q45)

Q38 舌、口腔及咽其他先天性畸形

排除：巨口(症、畸形) (Q18.4)
小口(症、畸形) (Q18.5)

Q38.0 唇先天性畸形，他處未歸類者

(下列)先天性：

- 唇瘻管
- 唇畸形，其他未特定者

Van der Woude (氏)症候群

排除：唇裂 (Q36.)

- 伴有(合併或併發)腭裂 (Q37.-)
- 巨唇(症、畸形) (Q18.6)
- 小唇(症、畸形) (Q18.7)

Q38.1 舌黏連[舌繫帶短縮]

舌繫帶短縮

Q38.2 巨舌(症、畸形)

Q38.3 舌其他先天性畸形

無舌

分叉舌

(下列)先天性：

舌的 {

- 沾黏(黏連)
- 裂溝(溝裂)
- 畸形，其他未特定者

小舌(症、畸形)

舌發育不全(症、畸形)

小舌(症、畸形)

Other congenital malformations of the digestive system (Q38-Q45)

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:

- fistula of lip
- malformation of lip NOS

Van der Woude's syndrome

Excludes: cleft lip (Q36.-)

- with cleft palate (Q37.-)
- macrocheilia (Q18.6)
- microcheilia (Q18.7)

Q38.1 Ankyloglossia

Tongue tie

Q38.2 Macroglossia

Q38.3 Other congenital malformations of tongue

Aglossia

Bifid tongue

Congenital:

{

- adhesion
- fissure
- malformation NOS

} of tongue

Hypoglossia

Hypoplasia of tongue

Microglossia

Q38.4 唾液腺及唾管先天性畸形

唾液腺及唾管的 { 缺損
副腺
閉鎖
唾液腺先天性瘻管

Q38.5 腭先天性畸形，他處未歸類者

懸壅垂缺損[無懸壅垂]

腭先天性畸形，其他未特定者

高弓腭

排除：腭裂 (Q35.-)

• 伴有(合併或併發)唇裂
(Q37.-)

Q38.6 口腔其他先天性畸形

口腔先天性畸形，其他未特定者

Q38.7 咽(小)囊

咽憩室

排除：咽(小)囊症候群 (D82.1)

Q38.8 咽其他先天性畸形

咽先天性畸形，其他未特定者

Q39 食道先天性畸形

Q39.0 食道閉鎖未伴有(未合併或未併發)瘻管

食道閉鎖，其他未特定者

Q39.1 食道閉鎖伴有(合併或併發)氣管—食道瘻管

食道閉鎖伴有(合併或併發)支氣管—食道瘻管

Q39.2 先天性氣管—食道瘻管未伴有(未合併或未併發)閉鎖

先天性氣管—食道瘻管，其他未特定者

Q39.3 食道先天性狹窄及縮窄

Q39.4 食道蹼

Q39.5 食道先天性擴張

Q39.6 食道憩室

食道(盲)囊

Q38.4 Congenital malformations of salivary glands and ducts

Absence } (of) salivary gland or duct
Accessory }
Atresia }
Congenital fistula of salivary gland

Q38.5 Congenital malformations of palate, not elsewhere classified

Absence of uvula

Congenital malformation of palate NOS

High arched palate

Excludes: cleft palate (Q35.-)

• with cleft lip (Q37.-)

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

Q39 Congenital malformations of oesophagus

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

Q39.2 Congenital tracheo-oesophageal fistula without atresia

Congenital tracheo-oesophageal fistula NOS

Q39.3 Congenital stenosis and stricture of oesophagus

Q39.4 Oesophageal web

Q39.5 Congenital dilatation of oesophagus

Q39.6 Diverticulum of oesophagus

Oesophageal pouch

Q39.8 食道其他先天性畸形

食道(的) { 缺損
先天性移位
雙套

Q39.9 食道先天性畸形，未特定者

Q40 上消化道其他先天性畸形

Q40.0 先天性肥厚(增厚、肥大)性幽門狹窄

先天性或嬰兒性：

幽門的 { • 窄縮
• 肥厚(增厚、肥大)
• 痙攣
• 狹窄
• 縮窄

Q40.1 先天性裂孔疝氣(疝脫)

經由食道裂孔的賁門移位

排除：先天性橫膈疝氣(疝脫) (Q79.0)

Q40.2 胃其他特定的先天性畸形

(下列)先天性：

- 賁門痙攣
- 胃移位
- 胃憩室
- 葫蘆(沙漏)型胃

雙(套)胃(畸形)

巨胃(症、畸形)

小胃(症、畸形)

Q40.3 胃先天性畸形，未特定者

Q40.8 上消化道其他特定的先天性畸形

Q40.9 上消化道先天性畸形，未特定者

先天性：

Q39.8 Other congenital malformations of oesophagus

Absent
Congenital displacement
Duplication } (of) oesophagus

Q39.9 Congenital malformation of oesophagus, unspecified

Q40 Other congenital malformations of upper alimentary tract

Q40.0 Congenital hypertrophic pyloric stenosis

Congenital or infantile:

{ • constriction
• hypertrophy
• spasm
• stenosis
• stricture } of pylorus

Q40.1 Congenital hiatus hernia

Displacement of cardia through oesophageal hiatus

Excludes: congenital diaphragmatic hernia (Q79.0)

Q40.2 Other specified congenital malformations of stomach

Congenital:

- cardiospasm
- displacement of stomach
- diverticulum of stomach
- hourglass stomach

Duplication of stomach

Megalogastria

Microgastria

Q40.3 Congenital malformation of stomach, unspecified

Q40.8 Other specified congenital malformations of upper alimentary tract

Q40.9 Congenital malformation of upper alimentary tract, unspecified

Congenital:

} NOS of upper alimentary tracts

上消化道其他未特定的 {

- 異常(畸形)
- 變形

- anomaly
- deformity

Q41 小腸先天性缺損、閉鎖及狹窄

包含：小腸或腸先天性阻塞、閉塞及狹窄，其他未特定者

排除：胎便性腸阻塞 (E84.1)

Q41.0 十二指腸先天性缺損、閉鎖及狹窄

Q41.1 空腸先天性缺損、閉鎖及狹窄

蘋果皮症候群

無孔空腸[空腸閉鎖不通]

Q41.2 迴腸先天性缺損[無迴腸]、閉鎖及狹窄

Q41.8 小腸其他特定部位的先天性缺損、閉鎖及狹窄

Q41.9 小腸先天性缺損[無小腸]、閉鎖及狹窄，部位未特定者

腸先天性缺損[無腸]、閉鎖及狹窄，其他未特定者

Q42 大腸先天性缺損、閉鎖及狹窄

包含：大腸先天性阻塞、閉鎖及狹窄

Q42.0 直腸先天性缺損[無直腸]、閉鎖及狹窄，伴有(合併或併發)瘻管

Q42.1 直腸先天性缺損[無直腸]、閉鎖及狹窄，未伴有(未合併或未併發)瘻管

無孔直腸[直腸閉鎖不通]

Q42.2 肛門缺損[無肛門]、閉鎖及狹窄，伴有(合併或併發)瘻管

Q42.3 肛門缺損[無肛門]、閉鎖及狹窄，未伴有(未合併或未併發)瘻管

無孔肛門[肛門閉鎖不通]

Q42.8 大腸其他部位先天性缺損[無部份大腸]、閉鎖及狹窄

Q42.9 大腸先天性缺損[無大腸]、閉鎖及狹窄，部位未特定者

Q41 Congenital absence, atresia and stenosis of small intestine

Includes: congenital obstruction, occlusion and stricture of small intestine or intestine NOS

Excludes: meconium ileus (E84.1)

Q41.0 Congenital absence, atresia and stenosis of duodenum

Q41.1 Congenital absence, atresia and stenosis of jejunum

Apple peel syndrome

Imperforate jejunum

Q41.2 Congenital absence, atresia and stenosis of ileum

Q41.8 Congenital absence, atresia and stenosis of other specified parts 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

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

Imperforate rectum

Q42.2 Congenital absence, atresia and stenosis of anus with fistula

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

Imperforate anus

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

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

Q43 腸道其他先天性畸形

Q43.0 Meckel (氏)憩室

持續(殘留)的：

- 臍腸系膜導管
- 卵黃管

Q43.1 Hirschsprung (氏)病 [先天性巨大結腸症]

無神經節細胞症

先天性(無神經節性)巨大結腸症

Q43.2 結腸其他先天性功(機)能疾患

結腸先天性擴張(症)

Q43.3 先天性腸固定畸形

先天性沾黏(黏連)[帶]：

- 網膜的，異常的
- 腹膜的

Jackson (氏)膜

結腸旋轉不良

(下列)旋轉：

盲腸及結腸的 $\left\{ \begin{array}{l} \cdot \text{衰竭} \\ \cdot \text{不完全} \\ \cdot \text{不足} \end{array} \right.$

普遍存在的腸系膜[腸系膜過多]

Q43.4 雙套腸

Q43.5 異位肛門

Q43.6 直腸及肛門先天性瘻管

排除：(下列)先天性瘻管：

- 直腸陰道的 (Q52.2)
- 尿道直腸的 (Q64.7)
- 潛毛性瘻管或竇 (L05.-)
- 伴有(合併或併發)缺損、閉鎖及狹窄 (Q42.0-Q42.2)

Q43.7 持續(殘留)的泄殖腔

泄殖腔，其他未特定者

Q43.8 腸其他特定的先天性畸形

(下列)先天性：

- (腸)盲環症候群

Q43 Other congenital malformations of intestine

Q43.0 Meckel's diverticulum

Persistent:

- omphalomesenteric duct
- vitelline duct

Q43.1 Hirschsprung's disease

Aganglionosis

Congenital (aganglionic) megacolon

Q43.2 Other congenital functional disorders of colon

Congenital dilatation of colon

Q43.3 Congenital malformations of intestinal fixation

Congenital adhesions [bands]:

- omental, anomalous
- peritoneal

Jackson's membrane

Malrotation of colon

Rotation :

$\left\{ \begin{array}{l} \cdot \text{failure of} \\ \cdot \text{incomplete} \\ \cdot \text{insufficient} \end{array} \right\}$ of caecum and colon

Universal mesentery

Q43.4 Duplication of intestine

Q43.5 Ectopic anus

Q43.6 Congenital fistula of rectum and anus

Excludes: congenital fistula:

- rectovaginal (Q52.2)
- urethrorectal (Q64.7)
- pilonidal fistula or sinus (L05.-)
- with absence, atresia and stenosis (Q42.0, Q42.2)

Q43.7 Persistent cloaca

Cloaca NOS

Q43.8 Other specified congenital malformations of intestine

Congenital:

- blind loop syndrome

- 憩室炎，結腸
- 憩室，腸
- 長結腸(症、畸形)
- 巨大闌尾(症、畸形)
- 巨大十二指腸(症、畸形)
- 小結腸(症、畸形)
- (下列)轉位(錯位)：
 - 闌尾
 - 結腸
 - 腸

- diverticulitis, colon
- diverticulum, intestine
- Dolichocolon
- Megaloappendix
- Megaloduodenum
- Microcolon
- Transposition of:
 - appendix
 - colon
 - intestine

Q43.9 腸先天性畸形，未特定者

Q43.9 Congenital malformation of intestine, unspecified

Q44 膽囊、膽管及肝的先天性畸形

Q44 Congenital malformations of gallbladder, bile ducts and liver

Q44.0 膽囊生成不良、發育不良及發育不全

膽囊先天性缺損[無膽囊]

Q44.0 Agenesis, aplasia and hypoplasia of gallbladder

Congenital absence of gallbladder

Q44.1 膽囊其他先天性畸形

膽囊先天性畸形，其他未特定者

肝內膽囊

Q44.1 Other congenital malformations of gallbladder

Congenital malformation of gallbladder NOS

Intrahepatic gallbladder

Q44.2 膽管閉鎖

Q44.2 Atresia of bile ducts

Q44.3 膽管先天性狹窄及縮窄

Q44.3 Congenital stenosis and stricture of bile ducts

Q44.4 總膽管囊腫

Q44.4 Choledochal cyst

Q44.5 膽管其他先天性畸形

Q44.5 Other congenital malformations of bile ducts

副肝管

膽管先天性畸形，其他未特定者

雙套：

- 膽管
- 膽囊管

Accessory hepatic duct

Congenital malformation of bile duct NOS

Duplication :

- biliary duct
- cystic duct

Q44.6 肝囊腫性疾病

肝纖維囊腫性疾病

Q44.6 Cystic disease of liver

Fibrocystic disease of liver

Q44.7 肝其他先天性畸形

副肝

Alagille (氏)症候群

先天性：

- 肝缺損[無肝]
- 肝腫大

Q44.7 Other congenital malformations of liver

Accessory liver

Alagille's syndrome

Congenital:

- absence of liver
- hepatomegaly

• 肝畸形，其他未特定者

• malformation of liver NOS

Q45 消化系統其他先天性畸形

排除：先天性：

- 橫膈疝氣(疝脫) (Q79.0)
- 裂孔疝氣(疝脫) (Q40.1)

Q45.0 胰生成不良、發育不良及發育不全

胰先天性缺損[無胰(臟)]

Q45.1 環狀胰

Q45.2 先天性胰囊腫

Q45.3 胰及胰管其他先天性畸形

副胰

胰或胰管先天性畸形，其他未特定者

排除：(下列)糖尿病：

- 先天性 (E10.-)
- 新生兒 (P70.2)
- 胰纖維囊腫性疾病 (E84.-)

Q45.8 消化系統其他特定的先天性畸形

消化道(完全)(部份)缺損，其他未特定者

其他未特定的消化道 { 雙套
轉(錯)位，
先天性

Q45.9 消化系統先天性畸形，未特定者

先天性：

消化系統其他 { 異常(畸形)
未特定的 { 變形

生殖器官先天性畸形 (Q50-Q56)

排除：雄性(激)素阻抗症候群 (E34.5)

Q45 Other congenital malformations of digestive system

Excludes: congenital:

- diaphragmatic hernia(Q79.0)
- hiatus hernia (Q40.1)

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

Excludes: diabetes mellitus:

- congenital (E10.-)
- neonatal (P70.2)
- fibrocystic disease of pancreas (E84.-)

Q45.8 Other specified congenital malformations of digestive system

Absence (complete) (partial) of alimentary tract NOS

Duplication } of digestive
Malposition, congenital } organs NOS

Q45.9 Congenital malformation of digestive system, unspecified

Congenital:

• anomaly } NOS of digestive system
• deformity }

Congenital malformations of genital organs (Q50-Q56)

Excludes: androgen resistance syndrome (E34.5)

(與)染色體數目及形狀異常(畸形)相關的症候群 (Q90-Q99)

睪丸雌化(女性化)症候群 (E34.5)

syndromes associated with anomalies in the number and form of chromosomes (Q90-Q99)

testicular feminization syndrome (E34.5)

Q50 卵巢、輸卵管及子宮闊韌帶的先天性畸形

Q50.0 先天性卵巢缺損

排除：Turner (氏)症候群 (Q96.-)

Q50.1 發育性卵巢囊腫

Q50.2 卵巢先天性扭轉

Q50.3 卵巢其他先天性畸形

副卵巢

卵巢先天性畸形，其他未特定者

卵巢痕

Q50.4 輸卵管胚囊

(卵巢)繖囊

Q50.5 子宮闊韌帶胚囊

(下列)囊腫：

- 卵巢冠
- Gartner (氏)管[卵巢冠縱管]
- 卵巢旁

Q50.6 輸卵管及子宮闊韌帶其他先天性畸形

輸卵管或子宮闊韌帶(之) $\left\{ \begin{array}{l} \text{缺損} \\ \text{副(的)} \\ \text{閉鎖} \end{array} \right.$

輸卵管或子宮闊韌帶先天性畸形，其他未特定者

Q50 Congenital malformations of ovaries, fallopian tubes and broad ligaments

Q50.0 Congenital absence of ovary

Excludes: Turner's syndrome (Q96.-)

Q50.1 Developmental ovarian cyst

Q50.2 Congenital torsion of ovary

Q50.3 Other congenital malformations of ovary

Accessory ovary

Congenital malformation of ovary NOS

Ovarian streak

Q50.4 Embryonic cyst of fallopian tube

Fimbrial cyst

Q50.5 Embryonic cyst of broad ligament

Cyst:

- epoophoron
- Gartner's duct
- parovarian

Q50.6 Other congenital malformations of fallopian tube and broad ligament

Absence

Accessory

Atresia

$\left. \begin{array}{l} \text{Absence} \\ \text{Accessory} \\ \text{Atresia} \end{array} \right\} \text{ (of) fallopian tube or broad ligament}$

Congenital malformation of fallopian tube or broad ligament NOS

Q51 子宮及子宮頸先天性畸形

Q51.0 子宮生成不良及發育不良

先天性子宮缺損[無子宮]

Q51.1 雙(套)子宮伴有(合併或併發)雙(套)子宮頸及雙(套)陰道

Q51.2 其他雙(套)子宮

雙(套)子宮，其他未特定者

Q51.3 雙角子宮

Q51.4 單角子宮

Q51.5 子宮頸生成不良及發育不良

先天性子宮頸缺損[無子宮頸]

Q51 Congenital malformations of uterus and cervix

Q51.0 Agenesia and aplasia of uterus

Congenital absence of uterus

Q51.1 Doubling of uterus with doubling of cervix and vagina

Q51.2 Other doubling of uterus

Doubling of uterus NOS

Q51.3 Bicornate uterus

Q51.4 Unicornate uterus

Q51.5 Agenesia and aplasia of cervix

Congenital absence of cervix

- Q51.6 子宮頸胚囊
- Q51.7 子宮、消化道及泌尿道[尿路]間的先天性瘻管
- Q51.8 子宮及子宮頸其他先天性畸形
- 子宮及子宮頸發育不全
- Q51.9 子宮及子宮頸先天性畸形，未特定者

Q52 女性生殖器其他先天性畸形

- Q52.0 先天性陰道缺損[無陰道]
- Q52.1 雙(套)陰道
- 陰道(分)隔
- 排除：**雙(套)陰道伴有(合併或併發)雙(套)子宮及雙(套)子宮頸 (Q51.1)
- Q52.2 先天性直腸陰道瘻管
- 排除：**泄殖腔 (Q43.7)
- Q52.3 處女膜未穿孔[處女膜閉鎖]
- Q52.4 陰道其他先天性畸形
- 陰道先天性畸形，其他未特定者
(下列)囊腫：
- Nuck (氏)管，先天性
 - 胚胎性陰道的
- Q52.5 陰唇融合
- Q52.6 陰蒂先天性畸形
- Q52.7 外陰其他先天性畸形
- (下列)先天性：
- | | |
|----|--|
| 外陰 | $\left\{ \begin{array}{l} \text{缺損} \\ \text{囊腫} \\ \text{先天性畸形，其他未特定者} \end{array} \right.$ |
| | |
| | |
- Q52.8 女性生殖器其他特定的先天性畸形
- Q52.9 女性生殖器先天性畸形，未特定者

Q53 隱睪(症)[睪丸未降]

- Q53.0 睪丸異位[異位睪丸]
- 單側或雙側睪丸異位

- Q51.6 Embryonic cyst of cervix
- Q51.7 Congenital fistulae between uterus and digestive and urinary tracts
- Q51.8 Other congenital malformations of uterus and cervix
- Hypoplasia of uterus and cervix
- Q51.9 Congenital malformation of uterus and cervix, unspecified

Q52 Other congenital malformations of female genitalia

- Q52.0 Congenital absence of vagina
- Q52.1 Doubling of vagina
- Septate vagina
- Excludes:** doubling of vagina with doubling of uterus and cervix (Q51.1)
- Q52.2 Congenital rectovaginal fistula
- Excludes:** cloaca (Q43.7)
- Q52.3 Imperforate hymen
- Q52.4 Other congenital malformations of vagina
- Congenital malformation of vagina NOS
- Cyst:
- canal of Nuck, congenital
 - embryonic vaginal
- Q52.5 Fusion of labia
- Q52.6 Congenital malformation of clitoris
- Q52.7 Other congenital malformations of vulva
- Congenital:
- | | |
|---|--|
| <ul style="list-style-type: none"> • absence • cyst • malformation NOS | $\left. \vphantom{\begin{array}{l} \bullet \\ \bullet \\ \bullet \end{array}} \right\} \text{ of vulva}$ |
| | |
| | |
- Q52.8 Other specified congenital malformations of female genitalia
- Q52.9 Congenital malformation of female genitalia, unspecified

Q53 Undescended testicle

- Q53.0 Ectopic testis
- Unilateral or bilateral ectopic testes

Q53.1 隱睪(症)，單側

Q53.2 隱睪(症)，雙側

Q53.9 隱睪(症)，未特定者

隱睪(症)，其他未特定者

Q54 尿道下裂

排除：尿道上裂 (Q64.0)

Q54.0 尿道下裂，龜頭的

(下列)尿道下裂：

- 冠狀部
- 龜頭部

Q54.1 尿道下裂，陰莖的

Q54.2 尿道下裂，陰莖陰囊的

Q54.3 尿道下裂，會陰的

Q54.4 先天性弓彎形陰莖(可能會有痛性勃起)

Q54.8 其他尿道下裂

Q54.9 尿道下裂，未特定者

Q55 男性生殖器其他先天性畸形

排除：先天性陰囊水腫 (P83.5)

尿道下裂 (Q54.-)

Q55.0 睪丸缺損[無睪丸]及發育不良

單睪丸

Q55.1 睪丸及陰囊發育不全

睪丸融合

Q55.2 睪丸及陰囊其他先天性畸形

睪丸或陰囊先天性畸形，其他未特定者

多睪丸

可縮性睪丸

移行性睪丸

Q55.3 輸精管閉鎖

Q55.4 輸精管、副睪、精囊及前列腺[攝護腺]其他先天性畸形

缺損或發育不良：

- 前列腺[攝護腺]
- 精索

Q53.1 Undescended testicle, unilateral

Q53.2 Undescended testicle, bilateral

Q53.9 Undescended testicle, unspecified

Cryptorchism NOS

Q54 Hypospadias

Excludes: epispadias (Q64.0)

Q54.0 Hypospadias, balanic

Hypospadias:

- coronal
- glandular

Q54.1 Hypospadias, penile

Q54.2 Hypospadias, penoscrotal

Q54.3 Hypospadias, perineal

Q54.4 Congenital chordee

Q54.8 Other hypospadias

Q54.9 Hypospadias, unspecified

Q55 Other congenital malformations of male genital organs

Excludes: congenital hydrocele (P83.5)

hypospadias (Q54.-)

Q55.0 Absence and aplasia of testis

Monorchism

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

Retractile testis

Testis migrans

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

輸精管、副睪、精囊或前列腺[攝護腺]的
先天性畸形，其他未特定者

Q55.5 陰莖先天性缺損[無陰莖]及發育不良

Q55.6 陰莖其他先天性畸形

陰莖先天性畸形，其他未特定者

陰莖(側向)彎曲

陰莖發育不全

Q55.8 男性生殖器其他特定的先天性畸形

Q55.9 男性生殖器先天性畸形，未特定者

先天性：

男性生殖器其他
未特定的

{

- 異常(畸形)
- 變形

Q56 不確定性別[性別不明]及假性陰陽人[兩性同體]

排除：假性陰陽人[兩性同體]

- 女性，伴有(合併或併發)腎上腺皮質疾患 (E25.-)
- 男性，伴有(合併或併發)雄性(激)素抵抗 (E34.5)
- 伴有(合併或併發)特定的染色體異常(畸形) (Q96-Q99)

Q56.0 假性陰陽人[兩性同體]，他處未歸類者

卵睪[兩性生殖腺]

Q56.1 男性假性陰陽人，他處未歸類者

男性假性陰陽人，其他未特定者

Q56.2 女性假性陰陽人，他處未歸類者

女性假性陰陽人，其他未特定者

Q56.3 假性陰陽人[兩性同體]，未特定者

Q56.4 不確定性別[性別不明]，未特定者

性別不明確(隱晦不明)的生殖器

泌尿系統先天性畸形 (Q60-Q64)

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

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

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

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

Congenital malformations of the urinary system (Q60-Q64)

Q60 腎生成不良及腎其他短少(縮減)缺陷

包含:(下列)腎萎縮:

- 先天性
- 嬰兒性

腎先天性缺損[無腎(臟)]

- Q60.0 腎生成不良, 單側
- Q60.1 腎生成不良, 雙側
- Q60.2 腎生成不良, 未特定者
- Q60.3 腎發育不全, 單側
- Q60.4 腎發育不全, 雙側
- Q60.5 腎發育不全, 未特定者
- Q60.6 Potter (氏)症候群

Q61 囊腫性腎疾病

排除:後天性腎囊腫 (N28.1)

Potter (氏)症候群 (Q60.6)

- Q61.0 先天性單一腎囊腫
(先天性)(單一)腎囊腫
- Q61.1 多囊(腫)性腎, 嬰兒型
- Q61.2 多囊(腫)性腎, 成人型
- Q61.3 多囊(腫)性腎, 未特定者
- Q61.4 腎發育不良
- Q61.5 髓囊腫性腎
海綿狀腎, 其他未特定者
- Q61.8 其他囊腫性腎疾病
(下列)纖維囊腫性:
 - 腎
 - 腎退化變性或腎疾病
- Q61.9 囊腫性腎疾病, 未特定者
Meckel-Gruber (二氏)症候群

Q62 腎盂先天性阻塞性缺陷及輸尿管先天性畸形

- Q62.0 先天性水腎[腎積水]
- Q62.1 輸尿管閉鎖及狹窄
下列先天性閉塞:
 - 輸尿管

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

Q61 Cystic kidney disease

Excludes: acquired cyst of kidney (N28.1)

Potter's syndrome (Q60.6)

- Q61.0 Congenital single renal cyst
Cyst of kidney (congenital) (single)
- Q61.1 Polycystic kidney, infantile type
- Q61.2 Polycystic kidney, adult type
- Q61.3 Polycystic kidney, unspecified
- Q61.4 Renal dysplasia
- Q61.5 Medullary cystic kidney
Sponge kidney NOS
- Q61.8 Other cystic kidney diseases
Fibrocystic:
 - kidney
 - renal degeneration or disease
- Q61.9 Cystic kidney disease, unspecified
Meckel-Gruber syndrome

Q62 Congenital obstructive defects of renal pelvis and congenital malformations of ureter

- Q62.0 Congenital hydronephrosis
- Q62.1 Atresia and stenosis of ureter
Congenital occlusion of:
 - ureter

- 輸尿管腎盂結合處
 - 輸尿管膀胱開口處
- 輸尿管不通(透)

Q62.2 先天性巨大輸尿管

先天性輸尿管擴張

Q62.3 腎盂及輸尿管其他阻塞性缺陷

先天性輸尿管膨出

Q62.4 輸尿管生成不良

輸尿管缺損[無輸尿管]

Q62.5 雙(套)輸尿管

副
雙(套) } 輸尿管

Q62.6 輸尿管轉(錯)位

輸尿管或輸尿管開口的

{	偏移
	移位
	異位
	異常植入

Q62.7 先天性膀胱—輸尿管—腎逆(回)流

Q62.8 輸尿管其他先天性畸形

輸尿管異常(畸形)，其他未特定者

Q63 腎其他先天性畸形

~~排除~~：先天性腎病症候群 (NO4.-)

Q63.0 副腎

Q63.1 分葉、融合及馬蹄形腎

Q63.2 異位腎

先天性腎移位

腎旋轉異常

Q63.3 增生(殖)性腎及巨(大)腎

Q63.8 腎其他特定的先天性畸形

先天性腎結石

Q63.9 腎先天性畸形，未特定者

Q64 泌尿系統其他先天性畸形

- ureteropelvic junction
 - ureterovesical orifice
- Impervious ureter

Q62.2 Congenital megaloureter

Congenital dilatation of ureter

Q62.3 Other obstructive defects of renal pelvis and ureter

Congenital ureterocele

Q62.4 Agenesis of ureter

Absent ureter

Q62.5 Duplication of ureter

Accessory }
Double } ureter

Q62.6 Malposition of ureter

Deviation
Displacement
Ectopic
Implantation, anomalous

{	(of)ureter or ureteric orifice
---	--------------------------------------

Q62.7 Congenital vesico-uretero-renal reflux

Q62.8 Other congenital malformations of ureter

Anomaly of ureter NOS

Q63 Other congenital malformations of kidney

Excludes: congenital nephrotic syndrome (NO4.-)

Q63.0 Accessory kidney

Q63.1 Lobulated, fused and horseshoe kidney

Q63.2 Ectopic kidney

Congenital displaced kidney

Malrotation of kidney

Q63.3 Hyperplastic and giant kidney

Q63.8 Other specified congenital malformations of kidney

Congenital renal calculi

Q63.9 Congenital malformation of kidney, unspecified

Q64 Other congenital malformations of urinary system

Q64.0 尿道上裂

~~排除~~：尿道下裂 (Q54.-)

Q64.1 膀胱外翻

膀胱異位

膀胱外翻

Q64.2 先天性後尿道瓣膜

Q64.3 尿道及膀胱頸其他閉鎖及狹窄

(下列)先天性：

- 膀胱頸阻塞

- 下列縮窄：

- 尿道

- 尿道口

- 膀胱尿道口

尿道不通(透)

Q64.4 臍尿管畸形

臍尿管囊腫

開放性臍尿管

臍尿管脫垂

Q64.5 膀胱及尿道先天性缺損[無膀胱且無尿道]

Q64.6 膀胱先天性憩室

Q64.7 膀胱及尿道其他先天性畸形

副(的)：

- 膀胱

- 尿道

先天性：

- 膀胱疝氣(疝脫)

- 膀胱或尿道畸形，其他未特定者

- 下列脫垂：

- 膀胱(黏膜)

- 尿道

- 尿道口

- 尿道直腸瘻管

雙(套)：

- 尿道

- 泌尿道口

Q64.0 Epispadias

Excludes: hypospadias (Q54.-)

Q64.1 Exstrophy of urinary bladder

Ectopia vesicae

Extroversion of bladder

Q64.2 Congenital posterior urethral valves

Q64.3 Other atresia and stenosis of urethra and bladder neck

Congenital:

- bladder neck obstruction

- stricture of:

- urethra

- urinary meatus

- vesicourethral orifice

Impervious urethra

Q64.4 Malformation of urachus

Cyst of urachus

Patent urachus

Prolapse of urachus

Q64.5 Congenital absence of bladder and urethra

Q64.6 Congenital diverticulum of bladder

Q64.7 Other congenital malformations of bladder and urethra

Accessory:

- bladder

- urethra

Congenital:

- hernia of bladder

- malformation of bladder or urethra NOS

- prolapse of :

- bladder (mucosa)

- urethra

- urinary meatus

- urethrorectal fistula

Double:

- urethra

- urinary meatus

Q64.8 泌尿系統其他特定的先天性畸形

Q64.9 泌尿系統先天性畸形，未特定者

先天性：

泌尿系統其他未特定的

{	• 異常(畸形)
	• 變形

Q64.8 Other specified congenital malformations of urinary system

Q64.9 Congenital malformation of urinary system, unspecified

Congenital:

{	• anomaly
	• deformity

NOS of urinary system

肌肉骨骼系統先天性畸形和變形 (Q65-Q79)

Congenital malformations and deformations of the musculoskeletal system (Q65-Q79)

Q65 髖(部)先天性變形

排除：彈響髖(部) (R29.4)

Q65.0 先天性髖(關節)脫位(白)，單側

Q65.1 先天性髖(關節)脫位(白)，雙側

Q65.2 先天性髖(關節)脫位(白)，未特定者

Q65.3 先天性髖(關節)不全脫位(白)，單側

Q65.4 先天性髖(關節)不全脫位(白)，雙側

Q65.5 先天性髖(關節)不全脫位(白)，未特定者

Q65.6 不穩定的髖(部)

易脫位(白)的髖(關節)

易不全脫位(白)的髖(關節)

Q65.8 髖(部)其他先天性變形

股骨頸前傾

先天性髖臼發育不良

先天性髖(部)：

• 外翻

• 內翻

Q65.9 髖(部)先天性變形，未特定者

Q66 足先天性變形

排除：足短縮缺陷 (Q72.-)

(後天性)外翻變形 (M21.0)

(後天性)內翻變形 (M21.1)

Q66.0 馬蹄內翻足

Q66.1 仰趾內翻足

Q66.2 內翻蹠

Q66.3 其他先天性足內翻變形

Q65 Congenital deformities of hip

Excludes: 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

Q65.8 Other congenital deformities of hip

Anteversion of femoral neck

Congenital acetabular dysplasia

Congenital coxa:

• valga

• 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

Q66.3 Other congenital varus deformities of feet

大趾內翻，先天性

Q66.4 仰趾外翻足

Q66.5 先天性扁平足

(下列)扁平足：

- 先天性
- 僵硬性
- 僵直性(外翻性)

Q66.6 其他先天性足外翻變形

蹠外翻

Q66.7 弓形足

Q66.8 足其他先天性變形

杵狀足，其他未特定者

槌狀趾，先天性

(下列)畸形足：

- 其他未特定者
- 不對稱

跗骨併合

垂直距骨

Q66.9 足先天性變形，未特定者

Q67 頭、顏面、脊椎及胸的先天性肌肉骨骼變形

排除：歸類於 **Q87.** - 分類項的先天性畸形症候群

Potter (氏)症候群 (Q60.6)

Q67.0 顏面不對稱(變形)

Q67.1 顏面受壓[扁臉](變形)

Q67.2 長頭(變形)

Q67.3 斜頭(變形)

Q67.4 顱骨、顏面及頷(顎)其他先天性變形

顱骨凹陷(變形)

鼻中隔偏曲，先天性

半側顏面萎縮或肥厚(增生、肥大)

扁鼻或塌(彎)鼻，先天性

排除：齒顏(面)骨異常(畸形或變形)[含咬合不正] (K07.-)

梅毒性鞍狀鼻 (A50.5)

Hallux varus, congenital

Q66.4 Talipes calcaneovalgus

Q66.5 Congenital pes planus

Flat foot:

- congenital
- rigid
- spastic (everted)

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

Q66.9 Congenital deformity of feet, unspecified

Q67 Congenital musculoskeletal deformities of head, face, spine and chest

Excludes: congenital malformation syndromes classified to Q87. -

Potter's syndrome (Q60.6)

Q67.0 Facial asymmetry

Q67.1 Compression facies

Q67.2 Dolichocephaly

Q67.3 Plagiocephaly

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

Depressions in skull

Deviation of nasal septum, congenital

Hemifacial atrophy or hypertrophy

Squashed or bent nose, congenital

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

syphilitic saddle nose (A50.5)

Q67.5 脊椎(柱)先天性變形

先天性脊椎側彎(症)：

- 其他未特定者
- 姿勢[姿位]性

排除：嬰兒不明原因(特發)性脊椎側彎(症) (M41.0)

先天性骨性畸形所致的脊椎側彎(症) (Q76.3)

Q67.6 漏斗胸

先天性漏斗胸

Q67.7 雞胸

先天性雞胸

Q67.8 胸部其他先天性變形

胸壁先天性變形，其他未特定者

Q68 肌肉骨骼其他先天性變形

排除：肢體短縮缺陷 (Q71-Q73)

Q68.0 胸鎖乳突肌先天性變形

先天性(胸骨乳突)斜頸(症)
胸鎖乳突(肌)攣縮(症)
(先天性)胸骨乳突腫瘤

Q68.1 手先天性變形

先天性杵狀指
(先天性)鏟狀手

Q68.2 膝先天性變形

先天性：
• 膝脫位(白)
• 膝反屈

Q68.3 股骨先天性弓(彎)曲

排除：股骨(頸)前傾 (Q65.8)

Q68.4 先天性脛骨及腓骨弓(彎)曲**Q68.5 先天性小腿長骨弓(彎)曲，未特定者****Q68.8 其他特定的先天性肌肉骨骼變形**

先天性：
• 下列變形：
• 鎖骨

Q67.5 Congenital deformity of spine

Congenital scoliosis:

- NOS
- postural

Excludes: infantile idiopathic scoliosis (M41.0)

scoliosis due to congenital bony malformation (Q76.3)

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)

Q68.1 Congenital deformity of hand

Congenital clubfinger
Spade-like hand (congenital)

Q68.2 Congenital deformity of knee

Congenital:
• dislocation of knee
• genu recurvatum

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

- 肘
- 前臂
- 肩胛骨

• 下列脫位(白)：

- 肘
- 肩

- elbow
- forearm
- scapula

• dislocation of:

- elbow
- shoulder

Q69 多指[趾](畸形或變形)

Q69.0 副指(畸形或變形)

Q69.1 副拇指(畸形或變形)

Q69.2 副趾(畸形或變形)

副拇趾(畸形或變形)

Q69.9 多指[趾](畸形或變形)，未特定者

贅餘指(畸形或變形)，未特定者

Q70 併指[趾](畸形或變形)

Q70.0 融合指(畸形或變形)

手指複雜性併指畸形伴有(合併或併發)骨性接合

Q70.1 指蹼(畸形或變形)

手指單純性併指畸形未伴有(未合併或未併發)骨性接合

Q70.2 融合趾(畸形或變形)

足趾複雜性併指畸形伴有(合併或併發)骨性接合

Q70.3 趾蹼(畸形或變形)

足趾單純性併趾畸形未伴有(未合併或未併發)骨性接合

Q70.4 多指併指[趾](畸形或變形)

Q70.9 併指[趾](畸形或變形)，未特定者

併指[趾](畸形或變形)[指(趾)關節黏連]，其他未特定者

Q71 上肢短縮缺陷(損)(畸形或變形)

Q71.0 上肢先天性完全缺損[無上肢](畸形或變形)

Q71.1 上肢及前臂先天性缺損[無上肢且無前臂]伴有(或合併)存在之手(畸形或變形)

Q69 Polydactyly

Q69.0 Accessory finger(s)

Q69.1 Accessory thumb(s)

Q69.2 Accessory toe(s)

Accessory hallux

Q69.9 Polydactyly, unspecified

Supernumerary digit(s) NOS

Q70 Syndactyly

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

Symphalangy NOS

Q71 Reduction defects of upper limb

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

Q71.1 Congenital absence of upper arm and forearm with hand present

Q71.2 前臂及手指先天性缺損[無前臂且無手指](畸形或變形)

Q71.3 手及手指先天性缺損[無手且無手指](畸形或變形)

Q71.4 橈骨縱行短縮缺陷(損)(畸形或變形)
(先天性)杵狀手(畸形或變形)
橈側杵狀手(畸形或變形)

Q71.5 尺骨縱行短縮缺陷(損)(畸形或變形)

Q71.6 龍蝦爪形手(畸形或變形)

Q71.8 其他上肢短縮缺陷(畸形或變形)
上肢先天性短縮(畸形或變形)

Q71.9 上肢短縮缺陷(損)(畸形或變形)，未特定者

Q72 下肢短縮缺陷(損)(畸形或變形)

Q72.0 先天性下肢完全缺損[無下肢](畸形或變形)

Q72.1 先天性腿(大腿及小腿)缺損[無腿]伴有(或合併)存在之足(畸形或變形)

Q72.2 小腿及足先天性缺損[無小腿且無足](畸形或變形)

Q72.3 足及趾先天性缺損[無足且無趾](畸形或變形)

Q72.4 股骨縱行短縮缺陷(損)(畸形或變形)
近端股骨局部缺乏(症)(畸形或變形)

Q72.5 脛骨縱行短縮缺陷(損)(畸形或變形)

Q72.6 腓骨縱行短縮缺陷(損)(畸形或變形)

Q72.7 裂足[足裂](畸形或變形)

Q72.8 下肢其他短縮缺陷(損)(畸形或變形)
下肢先天性短(縮)(畸形或變形)

Q72.9 下肢短縮缺陷(損)(畸形或變形)，未特定者

Q73 未特定的肢體短縮缺陷(損)(畸形或變形)

Q73.0 未特定的肢體先天性缺損(畸形或變形)
無肢(畸形或變形)，其他未特定者

Q 73.1 短肢[海豹肢](畸形或變形)，未特定肢體者
短肢[海豹肢](畸形或變形)，其他未特定者

Q71.2 Congenital absence of both forearm and hand

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

Q71.4 Longitudinal reduction defect of radius
Clubhand (congenital)
Radial clubhand

Q71.5 Longitudinal reduction defect of ulna

Q71.6 Lobster-claw hand

Q71.8 Other reduction defects of upper limb(s)
Congenital shortening of upper limb(s)

Q71.9 Reduction defect of upper limb(s), unspecified

Q72 Reduction defects of lower limb

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

Q72.1 Congenital absence of thigh and lower leg with foot present

Q72.2 Congenital absence of both lower leg and foot

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

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

Q72.5 Longitudinal reduction defect of tibia

Q72.6 Longitudinal reduction defect 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

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 未特定的肢體其他短縮缺陷(損)(畸形或變形)

未特定的肢體縱行短縮變形(畸形)

其他未特定肢體的 { 肢骨不全[短肢]，其他未特定者
半肢，其他未特定者
短縮缺陷(損)

Q74 肢體其他先天性畸形

排除：多指[趾](畸形或變形) (Q69.-)

肢體短縮缺陷(損)(畸形或變形)
(Q71-Q73)

併指[趾](畸形或變形) (Q70.-)

Q74.0 上肢其他先天性畸形，含肩胛帶

副腕骨(畸形或變形)

鎖顳骨成骨不全(畸形或變形)

先天性鎖骨假性關節(畸形或變形)

巨指[趾](症)(畸形或變形)

Madelung (氏)變形(畸形)

橈尺骨骨性接合(畸形或變形)

Sprengel (氏)變形(畸形)

姆指三指節(畸形或變形)

Q74.1 膝先天性畸形

(下列)先天性：

• 髌骨缺損[無髌骨](畸形或變形)

• 髌骨脫位(白)(畸形或變形)

• 膝(畸形或變形)：

• 外翻

• 內翻

發育不完全[未成熟]的髌骨

排除：先天性(畸形或變形)：

• 膝脫位(白) (Q68.2)

• 膝反屈 (Q68.2)

指甲髌骨症候群 (Q87.2)

Q74.2 下肢其他先天性畸形，含骨盆帶

先天性(畸形或變形)：

• 骶髂關節融合

Q73.8 Other reduction defects of unspecified limb(s)

Longitudinal reduction deformity of unspecified limb(s)

Ectromelia NOS

Hemimelia NOS

Reduction defect

} of limb(s) NOS

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

Accessory carpal bones

Cleidocranial dysostosis

Congenital pseudarthrosis of clavicle

Macroactylia (fingers)

Madelung's deformity

Radioulnar synostosis

Sprengel's deformity

Triphalangeal thumb

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:

• fusion of sacroiliac joint

• (下列)畸形：

- 踝(關節)
- 骶髂(關節)

排除：股骨(頸)前傾 (Q65.8)

Q74.3 先天性多重(發)關節彎曲症(畸形或變形)

Q74.8 肢體其他特定的先天性畸形

Q74.9 肢體未特定的先天性畸形

先天性肢體異常(畸形)，其他未特定者

Q75 顱骨及顏面骨其他先天性畸形

排除：顏面先天性畸形，其他未特定者 (Q18.-)

歸類於 **Q87.-** 的先天性畸形症候群

齒(顏)面異常(畸形)[含咬合不正] (K07.-)

頭及顏面肌肉骨骼變形 (Q67.0-Q67.4)

(與)先天性(大)腦異常(畸形)相關的顱骨缺陷，如：

- 無腦(症、畸形) (Q00.0)
- 腦膨出(症、畸形) (Q01.-)
- 水腦(症、畸形) (Q03.-)
- 小頭(症、畸形) (Q02)

Q75.0 顱骨縫過早封閉(畸形或變形)

尖頭(畸形或變形)

顱骨融合不全(畸形或變形)

尖頭(畸形或變形)

三角頭(畸形或變形)

Q75.1 顱顏(面)骨成骨不全

Crouzon (氏)病

Q75.2 眼距過寬(畸形或變形)

Q75.3 巨頭(症、畸形或變形)

Q75.4 下頷顏面骨成骨不全(畸形或變形)

Q75.5 眼下頷成骨不全(畸形或變形)

• malformation (of):

- ankle (joint)
- sacroiliac (joint)

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

Q74.3 Arthrogryposis multiplex congenita

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

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

Acrocephaly

Imperfect fusion of skull

Oxycephaly

Trigonocephaly

Q75.1 Craniofacial dysostosis

Crouzon's disease

Q75.2 Hypertelorism

Q75.3 Macrocephaly

Q75.4 Mandibulofacial dysostosis

Q75.5 Oculomandibular dysostosis

Q75.8 顱骨及顏面骨其他特定的畸形

顱骨缺損[無顱骨]，先天性
前額先天性變形
扁顱底(後腦)(畸形或變形)

Q75.9 顱骨及顏面骨先天性畸形，未特定者

下列先天性異常(畸形)：
• 顏面骨，其他未特定者
• 顱骨，其他未特定者

Q76 脊椎及胸廓先天性畸形

排除：脊椎(柱)及胸(廓)先天性肌肉骨骼變形 (Q67.5-Q67.8)

Q76.0 隱性脊椎(柱)裂

排除：(脊椎的)腦脊髓膜膨出(症、畸形)(Q05.-)
(開口性)(囊腫性)脊椎(柱)裂(Q05.-)

Q76.1 Klippel-Feil (二氏)症候群

頸(椎)融合症候群

Q76.2 先天性脊椎滑脫症

排除：(後天性)脊椎滑脫症 (M43.1)
(後天性)脊椎崩解(脫離)症(M43.0)

Q76.3 先天性骨性畸形所致的先天性脊椎側彎(症、畸形或變形)

半脊椎融合或未分節伴有(合併或併發)脊椎側彎(症、畸形或變形)

Q76.4 脊椎(柱)其他先天性畸形，(與)脊椎側彎(症)無關者

先天性(畸形)：

未特定
或未伴
隨脊椎
側彎
(症)的

- 脊椎(體)缺損
- 脊椎(柱)融合
- 脊椎(柱)後彎
- 脊椎(柱)前彎
- 下列畸形
 - 腰骶[薦](關節)(區段、部位)
 - 半椎

Q75.8 Other specified congenital malformations of skull and face bones

Absence of skull bone, congenital
Congenital deformity of forehead
Platybasia

Q75.9 Congenital malformation of skull and face bones, unspecified

Congenital anomaly of:
• face bones NOS
• skull NOS

Q76 Congenital malformations of spine and bony thorax

Excludes: congenital musculoskeletal deformities of spine and chest (Q67.5-Q67.8)

Q76.0 Spina bifida occulta

Excludes: meningocele (spinal)(Q05.-)
spina bifida (aperta)(cystica)(Q05.-)

Q76.1 Klippel-Feil syndrome

Cervical fusion syndrome

Q76.2 Congenital spondylolisthesis

Excludes: spondylolisthesis (acquired) (M43.1)
spondylolysis (acquired) (M43.0)

Q76.3 Congenital scoliosis due to congenital bony malformation

Hemivertebra fusion or failure of segmentation with scoliosis

Q76.4 Other congenital malformations of spine, not associated with scoliosis

Congenital:

- absence of vertebra
- fusion of spine
- kyphosis
- lordosis
- malformation of lumbosacral (joint) (region)
- Hemivertebra

unspecified
or not
associated
with
scoliosis

脊椎(柱)
脊椎(體)扁平
贅餘脊椎(體)

Malformation of spine
Platyspondylism
Supernumerary vertebra

Q76.5 頸肋骨

頸部區段(部位)贅餘肋骨

Q76.6 肋骨其他先天性畸形

副肋骨

先天性：

- 肋骨缺損[無肋骨]
- 肋骨融合
- 肋骨畸形，其他未特定者

排除：短肋症候群 (Q77.2)

Q76.7 胸骨先天性畸形

胸骨先天性缺損[無胸骨]

胸骨裂

Q76.8 骨性胸廓其他先天性畸形

Q76.9 骨性胸廓先天性畸形，未特定者

Q77 骨軟骨發育不良伴有(合併或併發)管狀骨及脊椎的生長缺陷

排除：黏多糖症 (E76.0-E76.3)

Q77.0 軟骨生成(成骨)不良

軟骨生成(成骨)不全

Q77.1 致死性矮小

Q77.2 短肋症候群

窒息性胸發育不良[Jeune (氏)]

Q77.3 點狀軟骨發育不良

Q77.4 軟骨發育不良

軟骨發育不全

Q77.5 彎曲變型性發育不良[彎曲侏儒症]

Q77.6 軟骨外胚層發育不良

Ellis-van Creveld (二氏)症候群

Q77.7 脊椎骨骺發育不良

Q77.8 其他骨軟骨發育不良伴有(合併或併發)管狀骨及脊椎生長缺陷

Q76.5 Cervical rib

Supernumerary rib in cervical region

Q76.6 Other congenital malformations of ribs

Accessory rib

Congenital:

- absence of rib
- fusion of ribs
- malformation of ribs NOS

Excludes: short rib syndrome (Q77.2)

Q76.7 Congenital malformation of sternum

Congenital absence of sternum

Sternum bifidum

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

Excludes: mucopolysaccharidosis (E76.0-E76.3)

Q77.0 Achondrogenesis

Hypochondrogenesis

Q77.1 Thanatophoric short stature

Q77.2 Short rib syndrome

Asphyxiating thoracic dysplasia [Jeune]

Q77.3 Chondrodysplasia punctata

Q77.4 Achondroplasia

Hypochondroplasia

Q77.5 Diastrophic dysplasia

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

Q77.9 骨軟骨發育不良伴有(合併或併發)
管狀骨及脊椎生長缺陷，未特定者

Q78 其他骨軟骨發育不良

Q78.0 骨生成(成骨)不全

骨易碎
骨脆症

Q78.1 多骨性纖維性發育不良

Albright (-McCune)(-Sternberg)(氏)症候群

Q78.2 骨質石化症

Albers-Schönberg (二氏)症候群

Q78.3 漸進性骨幹發育不良

Camurati-Engelmann (二氏)症候群

Q78.4 內生軟骨瘤症

Maffucci (氏)症候群
Ollier (氏)病

Q78.5 骨幹骺端發育不良

Pyle (氏)症候群

Q78.6 先天性多發性外生骨贅

骨幹連接症

Q78.8 其他特定的骨軟骨發育不良

(全身)骨脆弱性硬化症

Q78.9 骨軟骨發育不良，未特定者

軟骨失養(症)，其他未特定者
骨失養(症)，其他未特定者

**Q79 肌肉骨骼系統先天性畸形，他處
未歸類者**

~~排除~~：先天性(胸骨乳突)斜頸(症)
(Q68.0)

Q79.0 先天性橫膈疝氣(疝脫)

~~排除~~：先天性裂孔疝氣(疝脫)(Q40.1)

Q79.1 橫膈膜其他先天性畸形

橫膈膜缺損[無橫膈膜]
橫膈膜先天性畸形，其他未特定者
橫膈膜膨出

Q79.2 (先天性)臍膨出[臍疝氣(疝脫)]

Q77.9 Osteochondrodysplasia with defects of
growth of tubular bones and spine,
unspecified

Q78 Other osteochondrodysplasias

Q78.0 Osteogenesis imperfecta

Fragilitas ossium
Osteopsathyrosis

Q78.1 Polyostotic fibrous dysplasia

Albright (-McCune)(-Sternberg) syndrome

Q78.2 Osteopetrosis

Albers-Schönberg syndrome

Q78.3 Progressive diaphyseal dysplasia

Camurati-Engelmann syndrome

Q78.4 Enchondromatosis

Maffucci's syndrome
Ollier's disease

Q78.5 Metaphyseal dysplasia

Pyle's syndrome

Q78.6 Multiple congenital exostoses

Diaphyseal aclasis

Q78.8 Other specified osteochondrodysplasias

Osteopoikilosis

Q78.9 Osteochondrodysplasia, unspecified

Chondrodystrophy NOS
Osteodystrophy NOS

**Q79 Congenital malformations of
musculoskeletal system, not elsewhere
classified**

Excludes: congenital (sternomastoid) torticollis
(Q68.0)

Q79.0 Congenital diaphragmatic hernia

Excludes: congenital hiatus hernia (Q40.1)

Q79.1 Other congenital malformations of
diaphragm

Absence of diaphragm
Congenital malformation of diaphragm NOS
Eventration of diaphragm

Q79.2 Exomphalos

臍突出[臍疝氣(疝脫)]

排除：臍疝氣(疝脫) (K42.-)

Q79.3 裂腹(症、畸形)

Q79.4 梅腹症候群

Q79.5 腹壁其他先天性畸形

排除：臍疝氣(疝脫) (K42.-)

Q79.6 Ehlers-Danlos (二氏)症候群

Q79.8 肌肉骨骼系統其他先天性畸形

下列缺損：

• 肌肉

• 肌腱

副肌

先天性肌萎縮

先天性：

• 束狀帶(窄縮帶)

• 肌腱短縮

Poland (氏)症候群

Q79.9 肌肉骨骼系統先天性畸形，未特定者

先天性：

其他未特定肌肉
骨骼系統的

{	• 異常(畸形)，其他未特定者
	• 變形，其他未特定者

Omphalocele

Excludes: umbilical hernia (K42.-)

Q79.3 Gastroschisis

Q79.4 Prune belly syndrome

Q79.5 Other congenital malformations of abdominal wall

Excludes : umbilical hernia (K42.-)

Q79.6 Ehlers-Danlos syndrome

Q79.8 Other congenital malformations of musculoskeletal system

Absence of:

• muscle

• tendon

Accessory muscle

Amyotrophia congenita

Congenital:

• constricting bands

• shortening of tendon

Poland's syndrome

Q79.9 Congenital malformation of musculoskeletal system, unspecified

Congenital :

• anomaly NOS

• deformity NOS

} of musculoskeletal system NOS

其他先天性畸形 (Q80-Q89)

Q80 先天性魚鱗癬(症)

排除：Refsum's (氏)病 (G60.1)

Q80.0 尋常魚鱗癬(症)

Q80.1 X-性聯遺傳魚鱗癬(症)

Q80.2 層狀魚鱗癬(症)

火棉膠嬰兒

Q80.3 先天性(大)水疱性魚鱗癬狀紅皮症

Q80.4 斑色胎

Q80.8 其他先天性魚鱗癬(症)

Q80.9 先天性魚鱗癬(症)，未特定者

Other congenital malformations (Q80-Q89)

Q80 Congenital ichthyosis

Excludes : Refsum's disease (G60.1)

Q80.0 Ichthyosis vulgaris

Q80.1 X-linked ichthyosis

Q80.2 Lamellar ichthyosis

Collodion baby

Q80.3 Congenital bullous ichthyosiform erythroderma

Q80.4 Harlequin fetus

Q80.8 Other congenital ichthyosis

Q80.9 Congenital ichthyosis, unspecified

Q81 (大)水疱性表皮鬆解(症)**Q81.0 單純(大)水疱性表皮鬆解(症)**

排除：Cockayne (氏)症候群 (Q87.1)

Q81.1 致死性(大)水疱性表皮鬆解(症)

Herlitz (氏)症候群

Q81.2 失養性(大)水疱性表皮鬆解(症)**Q81.8 其他(大)水疱性表皮鬆解(症)****Q81.9 (大)水疱性表皮鬆解(症)，未特定者****Q82 皮膚其他先天性畸形**

排除：腸病變性肢皮炎 (E83.2)

先天性紅血球生成性紫質沉著
(積)症 (E80.0)

潛毛性囊腫或竇 (L05.-)

Sturge-Weber(-Dimitri)(二氏)症
候群 (Q85.8)

Q82.0 遺傳性淋巴水腫**Q82.1 色素性乾皮症****Q82.2 肥大細胞增多症**

色素性蕁麻疹

排除：惡性肥大細胞增多症 (C96.2)

Q82.3 色素失調症**Q82.4 (無汗性)外胚層發育不良**

排除：Ellis-van Creveld (二氏)症候群
(Q77.6)

Q82.5 先天性非腫瘤性痣

胎痣，其他未特定者

(下列之)痣：

- (火)燄色的
- 葡萄酒色的
- 多血管的
- 草莓狀
- 血管性，其他未特定者
- 疣狀

排除：咖啡牛奶斑 (L81.3)

色斑[小痣] (L81.4)

(下列之)痣：

- 其他未特定者 (D22.-)

Q81 Epidermolysis bullosa**Q81.0 Epidermolysis bullosa simplex**

Excludes: 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**

Excludes: 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

Excludes: malignant mastocytosis (C96.2)

Q82.3 Incontinentia pigmenti**Q82.4 Ectodermal dysplasia (anhidrotic)**

Excludes: Ellis-van Creveld syndrome (Q77.6)

Q82.5 Congenital non-neoplastic naevus

Birthmark NOS

Naevus:

- flammeus
- portwine
- sanguineous
- strawberry
- vascular NOS
- verrucous

Excludes: café au lait spots (L81.3)

lentigo (L81.4)

naevus:

- NOS (D22.-)

- 蛛狀 (I78.1)
- 黑色素細胞性 (D22.-)
- 色素性 (D22.-)
- 蜘蛛 (I78.1)
- 星狀 (I78.1)

- araneus (I78.1)
- melanocytic (D22.-)
- pigmented (D22.-)
- spider (I78.1)
- stellar (I78.1)

Q82.8 皮膚其他特定的先天性畸形

不正常掌紋
皮膚贅片
良性家族性天疱瘡[Hailey-Hailey (二氏)]
(超彈性)皮膚鬆垂症
皮紋異常(畸形)[手足之指(趾)掌(蹠)紋]
遺傳性掌蹠角化症
毛囊角化症[Darier-White (二氏)]
排除：Ehlers-Danlos (二氏)症候群
(Q79.6)

Q82.9 皮膚先天性畸形，未特定者

Q83 乳房先天性畸形

排除：胸肌缺損[無胸肌] (Q79.8)

Q83.0 先天性乳房缺損[無乳房]伴有(合併或併發)乳頭缺損[無乳頭]

Q83.1 副乳(房)

贅餘乳房

Q83.2 無乳頭

Q83.3 副乳頭

贅餘乳頭

Q83.8 乳房其他先天性畸形

乳房發育不全

Q83.9 乳房先天性畸形，未特定者

Q84 體被其他先天性畸形

Q84.0 先天性禿

先天性無毛髮(症)

Q84.1 毛髮先天性形態(學)障礙，他處未歸類者

念珠狀毛髮(症)

念珠形毛髮(症)

環形毛髮(症)

Q82.8 Other specified congenital malformations of skin

Abnormal palmar creases
Accessory skin tags
Benign familial pemphigus [Hailey-Hailey]
Cutis laxa (hyperelastica)
Dermatoglyphic anomalies
Inherited keratosis palmaris et plantaris
Keratosis follicularis [Darier-White]
Excludes: Ehlers-Danlos syndrome (Q79.6)

Q82.9 Congenital malformation of skin, unspecified

Q83 Congenital malformations of breast

Excludes: absence of pectoral muscle (Q79.8)

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

排除：Menkes (氏)捲毛症候群 (E83.0)

Q84.2 毛髮其他先天性畸形

先天性：

- 多毛(髮)症
- 毛髮畸形，其他未特定者

持續性胎毛(殘留)

Q84.3 無[指、趾]甲症

排除：指[趾]甲膈骨症候群 (Q87.2)

Q84.4 先天性[指、趾]甲白斑症

Q84.5 腫大及肥厚(增生、肥大)的指[趾]甲

先天性指[趾]甲肥厚(症)

[指、趾]甲肥厚(症)

Q84.6 指[趾]甲其他先天性畸形

先天性(畸形)：

- 杵狀指[趾]甲
- 匙形指[趾]甲
- 指[趾]甲畸形，其他未特定者

Q84.8 體被[皮膚]其他特定的先天性畸形

先天性皮膚發育不良

Q84.9 皮膚先天性畸形，未特定者

先天性：

其他未特定體被(被
膜、皮膚)的

{	• 異常(畸形)，其他未特定者
	• 變形，其他未特定者

Q85 斑痣性瘤症，他處未歸類者

排除：毛細血管擴張性運動失調

[Louis-Bar (二氏)] (G11.3)

家族性自主神經機能不良(障

礙、失調、異常) [Riley-Day

(二氏)] (G90.1)

Q85.0 (非惡性)神經纖維瘤症

Von Recklinghausen (氏)病

Q85.1 結節性(腦)硬化症

Bourneville (氏)病

結節性(腦)硬化

Q85.8 其他斑痣性瘤症，他處未歸類者

(下列)症候群：

Excludes: Menkes' kinky hair syndrome (E83.0)

Q84.2 Other congenital malformations of hair

Congenital:

- hypertrichosis
- malformation of hair NOS

Persistent lanugo

Q84.3 Anonychia

Excludes: 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

Aplasia cutis congenita

Q84.9 Congenital malformation of integument, unspecified

Congenital:

- | | |
|-----------------|---------------------|
| • anomaly NOS | } of integument NOS |
| • deformity NOS | |

Q85 Phakomatoses, not elsewhere classified

Excludes: ataxia telangiectasia [Louis-Bar]

(G11.3)

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

Syndrome:

- Peutz-Jeghers (二氏)
- Sturge-Weber (-Dimitri) (二氏)
- von Hippel-Lindau (二氏)

排除：Meckel-Gruber (二氏)症候群
(Q61.9)

Q85.9 斑痣性瘤症，未特定者

缺陷(瘤)症，其他未特定者

Q86 外在原因所致的先天性畸形症候群，他處未歸類者

排除：缺碘性甲狀腺低能症 (E00-E02)

經胎盤或母乳傳遞物質的非畸
胎效應(作用、影響) (P04.-)

Q86.0 [異(變、畸)形性]胎兒酒精症候群

Q86.1 胎兒內蘊脈症候群

Meadow (氏)症候群

Q86.2 Warfarin 所致的異(變、畸)形

Q86.8 已知外在原因所致的其他先天性畸形症候群

Q87 影響多(重)系統其他特定的先天性畸形症候群

Q87.0 影響顏面為主的先天性畸形症候群

尖頭多指[趾]併指[趾](症、畸形)
尖頭併指[趾](症、畸形)[Apert (氏)]
隱眼症候群
獨眼(症、畸形)
(下列)症候群：
• Goldenhar (氏)
• Moebius (氏)
• 口—(顏)面—指[趾]
• Robin (氏)
• Treacher Collins (氏)
吹哨狀(顏)面

- Peutz-Jeghers
- Sturge-Weber (-Dimitri)
- von Hippel-Lindau

Excludes: Meckel-Gruber syndrome (Q61.9)

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

Meadow's syndrome

Q86.2 Dysmorphism due to warfarin

Q86.8 Other congenital malformation syndromes due to known exogenous causes

Q87 Other specified congenital malformation syndromes affecting multiple systems

Q87.0 Congenital malformation syndromes predominantly affecting facial appearance

Acrocephalopolysyndactyly
Acrocephalosyndactyly [Apert]
Cryptophthalmos syndrome
Cyclopia
Syndrome:
• Goldenhar
• Moebius
• oro-facial-digital
• Robin
• Treacher Collins
Whistling face

Q87.1 (與)身材(體型)短小相關為主的先天性畸形症候群

(下列)症候群：

- Aarskog (氏)
- Cockayne (氏)
- De Lange (氏)
- Dubowitz (氏)
- Noonan (氏)
- Prader-Willi (氏)
- Robinow-Silverman-Smith (三氏)
- Russell-Silver (二氏)
- Seckel (氏)
- Smith-Lemli-Opitz (三氏)

排除：Ellis-van Creveld (二氏)症候群 (Q77.6)

Q87.2 侵及(侵害、涉及)肢體為主的先天性畸形症候群

(下列)症候群：

- Holt-Oram (二氏)
- Klippel-Tr'enaunay-Weber (三氏)
- 指[趾]甲贅骨
- Rubinstein-Taybi (二氏)
- 併腿無足[人魚狀]畸形
- 血小板減少伴有(合併或併發)橈骨缺損(無橈骨)[TAR]
- VATER[脊椎缺損、肛門閉鎖、氣管食道瘻、橈骨及腎發育異常]

Q87.3 涉及早期過度生長的先天性畸形症候群

(下列)症候群：

- Beckwith-Wiedemann (二氏)
- Sotos (氏)
- Weaver (氏)

Q87.4 Marfan (氏)症候群

Q87.5 其他先天性畸形症候群伴有(合併或併發)其他骨骼變化

Q87.8 其他特定的先天性畸形症候群，他處未歸類者

(下列)症候群：

- Alport (氏)

Q87.1 Congenital malformation syndromes predominantly associated with short stature

Syndrome:

- Aarskog
- Cockayne
- De Lange
- Dubowitz
- Noonan
- Prader-Willi
- Robinow-Silverman-Smith
- Russell-Silver
- Seckel
- Smith-Lemli-Opitz

Excludes: Ellis-van Creveld syndrome (Q77.6)

Q87.2 Congenital malformation syndromes predominantly involving limbs

Syndrome:

- Holt-Oram
- Klippel-Tr'enaunay-Weber
- nail patella
- Rubinstein-Taybi
- sirenomelia
- thrombocytopenia with absent radius [TAR]
- VATER

Q87.3 Congenital malformation syndromes involving early overgrowth

Syndrome:

- Beckwith-Wiedemann
- Sotos
- Weaver

Q87.4 Marfan's syndrome

Q87.5 Other congenital malformation syndromes with other skeletal changes

Q87.8 Other specified congenital malformation syndromes, not elsewhere classified

Syndrome:

- Alport

- Laurence-Moon(-Bardet)-Biedl (三氏)
- Zellweger (氏)

- Laurence-Moon (-Bardet)-Biedl
- Zellweger

Q89 其他先天性畸形，他處未歸類者

Q89.0 脾先天性畸形

(先天性)無脾(症、畸形)

先天性脾腫大

排除：心房附件異構(伴有或合併有無脾或多脾)(Q20.6)

Q89.1 腎上腺先天性畸形

排除：先天性腎上腺增生(殖)(E25.0)

Q89.2 其他內分泌腺先天性畸形

副甲狀腺或甲狀腺先天性畸形

甲狀舌管持續性殘留

甲狀舌管囊腫

Q89.3 內臟反位

右位心伴有(合併或併發)內臟反位

鏡像心房排列伴有(合併或併發)內臟反位

內臟反位或逆(倒轉)位：

- 腹
- 胸

內臟轉(錯)位：

- 腹
- 胸

排除：右位心，其他未特定者 (Q24.0)

Q89.4 連體雙胞胎

頭連(雙胎)

雙頭畸(怪)胎

雙畸(怪)胎

臂部相連(雙胎)

胸部相連(雙胎)

Q89.7 多重(發)先天性畸形，他處未歸類者

畸(怪)胎，其他未特定者

先天性多重的：

Q89 Other congenital malformations, not elsewhere classified

Q89.0 Congenital malformations of spleen

Asplenia (congenital)

Congenital splenomegaly

Excludes: isomerism of atrial appendages (with asplenia or polysplenia)(Q20.6)

Q89.1 Congenital malformations of adrenal gland

Excludes: congenital adrenal hyperplasia (E25.0)

Q89.2 Congenital malformations of other endocrine glands

Congenital malformation of parathyroid or thyroid gland

Persistent thyroglossal duct

Thyroglossal cyst

Q89.3 Situs inversus

Dextrocardia with situs inversus

Mirror-image atrial arrangement with situs inversus

Situs inversus or transversus:

- abdominalis
- thoracis

Transposition of viscera:

- abdominal
- thoracic

Excludes: dextrocardia NOS(Q24.0)

Q89.4 Conjoined twins

Craniopagus

Dicephaly

Double monster

Pygopagus

Thoracopagus

Q89.7 Multiple congenital malformations, not elsewhere classified

Monster NOS

Multiple congenital:

• 異常(畸形)，其他未特定者

• 變形，其他未特定者

排除：影響多處系統的先天性畸形症候群 (Q87.-)

• anomalies NOS

• deformities NOS

Excludes: congenital malformation syndromes affecting multiple systems (Q87.-)

Q89.8 其他特定的先天性畸形

Q89.9 先天性畸形，未特定者

先天性：

• 異常(畸形)，其他未特定者

• 變形，其他未特定者

Q89.8 Other specified congenital malformations

Q89.9 Congenital malformation, unspecified

Congenital:

• anomaly NOS

• deformity NOS

染色體異常，他處未歸類者 (Q90-Q99)

Chromosomal abnormalities, not elsewhere classified (Q90-Q99)

Q90 唐(氏)症候群

Q90.0 (常染色體)21 三染色體(性)，減數分裂不分離

Q90.1 (常染色體)21 三染色體(性)，嵌合型(有絲分裂不分離)

Q90.2 (常染色體)21 三染色體(性)，易位

Q90.9 唐(Down)(氏)症候群，未特定者
(常染色體)21 三染色體(性)，其他未特定者

Q91 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 (氏)症候群及 Patau (氏)症候群

Q91.0 (常染色體)18 三染色體(性)，減數分裂不分離

Q91.1 (常染色體)18 三染色體(性)，嵌合型(有絲分裂不分離)

Q91.2 (常染色體)18 三染色體(性)，易位

Q91.3 Edwards(氏)症候群，未特定者

Q91.4 (常染色體)13 三染色體(性)，減數分裂不分離

Q91.5 (常染色體)13 三染色體(性)，嵌合型(有絲分裂不分離)

Q91.6 (常染色體)13 三染色體(性)，易位

Q91.7 Patau(氏)症候群，未特定者

Q91 Edwards' 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 Edwards' 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 常染色體其他三染色體(性)及部份三染色體(性)，他處未歸類者

Q92 Other trisomies and partial trisomies of the autosomes, not elsewhere classified

包 含：不平衡易位及嵌入

Includes: unbalanced translocations and insertions

排 除：常染色體 13、18、21 的三染色體(性) (Q90-Q91)

Excludes: trisomies of chromosomes 13, 18, 21(Q90-Q91)

Q92.0 全部染色體的三染色體(性)，減數分裂不分離

Q92.0 Whole chromosome trisomy, meiotic nondisjunction

Q92.1 全部染色體的三染色體(性)，嵌合型(有絲分裂不分離)

Q92.1 Whole chromosome trisomy, mosaicism (mitotic nondisjunction)

Q92.2 大(主要)部份三染色體(性)
全部染色體臂或更多的重複(複製)

Q92.2 Major partial trisomy
Whole arm or more duplicated.

Q92.3 小(次要)部份三染色體(性)
少於全部染色體臂的重複(複製)

Q92.3 Minor partial trisomy
Less than whole arm duplicated.

Q92.4 只出現在前中期的重複(複製)

Q92.4 Duplications seen only at prometaphase

Q92.5 重複(製)伴有(合併或併發)其他複雜性的重新排列

Q92.5 Duplications with other complex rearrangements

Q92.6 額外標記的染色體

Q92.6 Extra marker chromosomes

Q92.7 三套染色體(性)及多套染色體(性)

Q92.7 Triploidy and polyploidy

Q92.8 其他特定的三染色體(性)及部份三染色體(性)

Q92.8 Other specified trisomies and partial trisomies of autosomes

Q92.9 三染色體(性)及部份三染色體(性)，未特定者

Q92.9 Trisomy and partial trisomy of autosomes, unspecified

Q93 常染色體的單染色體(性)及脫損，他處未歸類者

Q93 Monosomies and deletions from the autosomes, not elsewhere classified

Q93.0 全部染色體的單染色體(性)，減數分裂不分離

Q93.0 Whole chromosome monosomy, meiotic nondisjunction

Q93.1 全部染色體的單染色體(性)，嵌合型(有絲分裂不分離)

Q93.1 Whole chromosome monosomy, mosaicism (mitotic nondisjunction)

Q93.2 環狀或兩中心粒取代(替換)的染色體

Q93.2 Chromosome replaced with ring or dicentric

Q93.3 第 4 對染色體短臂脫損
Wolff-Hirschorn (二氏)症候群

Q93.3 Deletion of short arm of chromosome 4
Wolff-Hirschorn syndrome

Q93.4 第 5 對染色體短臂脫損
貓啼症候群

Q93.4 Deletion of short arm of chromosome 5
Cri-du-chat syndrome

Q93.5 其他部份染色體脫損

Q93.5 Other deletions of part of a chromosome

Q93.6 只出現在前中期的脫損

Q93.6 Deletions seen only at prometaphase

Q93.7 脫損伴有(合併或併發)其他複雜性的重新排列

Q93.7 Deletions with other complex rearrangements

Q93.8 常染色體其他脫損

Q93.8 Other deletions from the autosomes

Q93.9 常染色體脫損，未特定者

Q93.9 Deletion from autosomes, unspecified

Q95 平衡重新排列及結構標記，他處未歸類者

包含：Robertsonian (氏)及平衡交互易位及嵌入

Q95.0 正常個體平衡易位及嵌入

Q95.1 正常個體常染色體倒位

Q95.2 異常個體平衡的常染色體重新排列

Q95.3 異常個體平衡的性 / 常染全體重新排列

Q95.4 伴有(合併或併發)異染色質標記的個體

Q95.5 伴有(合併或併發)常染色體脆折處個體

Q95.8 其他平衡重新排列及結構標記

Q95.9 平衡重新排列及結構標誌，未特定者

Q96 Turner (氏)症候群

排除：Noonan (氏)症候群 (Q87.1)

Q96.0 核型 45 X

Q96.1 核型 46 X 同種(Xq)

Q96.2 核型 46 X，伴有(合併或併發)性染色體異常，同種(Xq)除外

Q96.3 嵌合型，45 X / 46 XX 或 XY

Q96.4 嵌合型，45 X / 其他細胞株伴有(合併或併發)異常的性染色體

Q96.8 Turner (氏)症候群其他變異型

Q96.9 Turner (氏)症候群，未特定者

Q97 其他性染色體異常，女性表現型，他處未歸類者

排除：Turner (氏)症候群 (Q96.-)

Q97.0 核型 47 XXX

Q97.1 女性伴有(合併或併發)多於3個X染色體

Q97.2 嵌合型，細胞株伴有(合併或併發)不同數目的X-染色體

Q97.3 女性伴有(合併或併發)46 XY 核型

Q95 Balanced rearrangements and structural markers, not elsewhere classified

Includes: 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

Excludes: Noonan's syndrome (Q87.1)

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

Excludes: Turner's syndrome (Q96.-)

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

Q97.8 其他特定的性染色體異常，女性表現型

Q97.9 性染色體異常，女性表現型，未特定者

Q98 其他性染色體異常，男性表現型，他處未歸類者

Q98.0 Klinefelter (氏)症候群，47 XXY 核型

Q98.1 Klinefelter (氏)症候群、男性伴有(合併或併發)多於兩個的 X-染色體[先天性睪丸發育不全]

Q98.2 Klinefelter (氏)症候群，男性伴有(合併或併發) 46 XX 核型[先天性睪丸發育不全]

Q98.3 其他男性伴有(合併或併發)46 XX 核型

Q98.4 Klinefelter (氏)症候群，未特定者

Q98.5 47 XYY 核型

Q98.6 男性伴有(合併或併發)結構異常的性染色體

Q98.7 男性伴有(合併或併發)性染色體嵌合型

Q98.8 其他特定的性染色體異常，男性表現型

Q98.9 性染色體異常，男性表現型，未特定者

Q99 其他染色體異常，他處未歸類者

Q99.0 嵌合體 46 XX / 46 XY

嵌合體 46XX / 46XY 真性陰陽人
[兩性同體]

Q99.1 46 XX 真性陰陽人[兩性同體]

46 XX 伴有(合併或併發)條紋性腺

46 XY 伴有(合併或併發)條紋性腺
單純性腺形成或發育不良

Q99.2 脆折的 X-染色體

脆折的 X-染色體症候群

Q99.8 其他特定的染色體異常

Q99.9 染色體異常，未特定者

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

