New York State Department of Health
Congenital Malformation Registry

ICD-10 Reportable Congenital Malformations Coding Manual

List of all Reportable Malformations

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Reporting Requirements and Instructions

Children to Report:

Hospitals and physicians are required to report children born or living in New York State who have been diagnosed before two years of age with congenital malformations, chromosomal anomalies, or persistent metabolic defects. For purposes of this registry, a congenital malformation is defined as any structural, functional, or biochemical abnormality determined genetically or induced during gestation and not due to birthing events. This includes newborns and pediatric patients with a birth defect even if they are transferred to or from another hospital. Any child with a congenital malformation who dies in the hospital (inpatient or emergency room) is also included.

What to Report:

This handbook contains a complete list of anomalies that are reportable to New York State's Congenital Malformations Registry (CMR). Rows with bold text and grey backgrounds represent non-billable categories or groups of defects as identified by the ICD-10 nomenclature. When reporting syndromes that have several defects, report each individual defect as well as the associated syndrome. Tables contain the following three columns:

1st Column called ICD-10 – ICD-10 codes that correspond to the child's condition.

2nd Column called Description of malformation – Lists the standard narrative descriptions of the birth defect and also gives specific directions or additional reporting information. For example, for congenital iodine-deficiency syndrome, do not report if infant's birth weight is less than 1500 grams because transient congenital hypothyroidism due to iodine deficiency is more common in premature infants. We have included additional text in blue which provides additional and/or more specific diagnoses that apply to that particular ICD-10 code. Please include as much detail as possible for each defect (i.e., indicate the laterality and try not to use codes with "not otherwise specified (NOS)"). As in the past, minor anomalies, those listed with an *(m)* in description, are only reported if the child also has an accompanying major malformation. Some codes are dependent upon gestational age and infant sex. The list of conditions under each code is not necessarily all-inclusive of anything that could be reported under that code.

NOTE: Narrative descriptions are now required for all birth defects!

3rd Column called ICD-9 - ICD-9 code that corresponds as closely as possible to the respective ICD-10 code.

For all syndrome and chromosomal defects, please report the cytogenetic diagnoses or test results. These results are also required for defects under ICD-10 code Q56 (indeterminate sex or pseudohermaphroditism).

Common Acronyms:

(m) = minor defect
NOS = Not Otherwise Specified
NEC = Not Elsewhere Classified

Color Coding:

Black font: From ICD-10 Codebook
Blue font: Synonyms or descriptions of other conditions also included under this code.
Red font: Key instructions to follow

Common Notation:

Includes: Further defines, or gives examples of, the content of the code or category
Excludes 1 (Type 1 Excludes): Indicates the code identified under the 'excludes' statement and the respective ICD10 code cannot be reported together because the two conditions cannot appear together.
Excludes 2 (Type 2 Excludes): Condition excluded is not part of the condition it is excluded from but a patient may have both conditions at the same time. Both codes may be reported together if the patient has both conditions.

Congenital Malformations of the Nervous System (Q00-Q07)

ICD -10	Description	ICD-9
Q00	**Anencephaly and similar malformations**	
Q00.0	Anencephaly Acephaly Acrania Amyelencephaly Hemianencephaly Hemicephaly	740.0
Q00.1	Craniorachischisis	740.1
Q00.2	Iniencephaly Closed iniencephaly Open iniencephaly Unspecified iniencephaly	740.2
Q01	**Encephalocele** **Includes:** Arnold-Chiari syndrome, type III, encephalocystocele, encephalomyelocele, hydroencephalocele, hydromeningocele (cranial), meningocele (cerebral), meningoencephalocele **Excludes 1:** Meckel-Gruber syndrome (Q61.9)	
Q01.0	Frontal encephalocele Encephalocystocele, frontal Encephalomyelocele, frontal Hydroencephalocele, frontal Hydromeningocele, frontal Meningocele, frontal Meningoencephalocele, frontal	742.0
Q01.1	Nasofrontal encephalocele Encephalocystocele, nasofrontal Encephalomyelocele, nasofrontal Hydroencephalocele, nasofrontal Hydromeningocele, nasofrontal Meningocele, nasofrontal Meningoencephalocele, nasofrontal	742.0
Q01.2	Occipital encephalocele Encephalocystocele, occipital Encephalomyelocele. Occipital Hydroencephalocele, occipital Hyd romeningocele, occipital Meningocele, occipital Menigoencephalocele, occipital	742.0
Q01.8	Encephalocele of other sites: cranial, cerebral, nasal, parietal, other specified site Encephalocystocele, cranial, cerebral, nasal, parietal or other specified site Encephalomyelocele, cranial, cerebral, nasal, parietal, other specified site Hydroencephalocele, cranial, cerebral, nasal, parietal, other specified site Hydromeningocele, cranial, cerebral, nasal, parietal, other specified site Meningocele, cranial, cerebral, nasal, parietal, other specified site Meningoecephalocele, cranial, cerebral, nasal, parietal, other specified site	742.0
Q01.9	Encephalocele, unspecified Encephalocystocele, unspecified Encephalomyelocele, unspecified Hydroencephalocele, unspecified Hydromeningocele, cranial, unspecified Meningocele, cerebral, unspecified Meningoencephalocele, unspecified	742.0

	Congenital cerebral meningocele Arnold-Chiari syndrome, Type 3 Chiari malformation type III	
Q02	**Microcephaly** **Includes:** hydromicrocephaly, micrencephalon **Excludes 1:** Meckel-Gruber syndrome (Q61.9)	
Q02	Microencephaly Brain hypoplasia, congenital Congenital brain hypoplasia Congenital hypoplasia of brain Hydromicrocephaly Microcephalus Primary microcephaly Micrencephalon Micrencephaly Galloway syndrome	742.1
Q03	**Congenital hydrocephalus** **Excludes 1:** Arnold-Chiari syndrome, type II (Q07.0-), acquired hydrocephalus (G91.-), hydrocephalus due to congenital toxoplasmosis (P37.1), hydrocephalus with spina bifida (Q05.0-Q05.4)	
Q03.0	Malformations of aqueduct of Sylvius Anomaly of aqueduct of Sylvius Obstruction of aqueduct of Sylvius, congenital Stenosis of aqueduct of Sylvius	742.3
Q03.1	Atresia of foramina of Magendie and Luschka Dandy-Walker syndrome	742.3
Q03.8	Other congenital hydrocephalus Communicating hydrocephaly/enlarged Cisterna Magna Other specified hydrocephaly (do not code if Intraventricular hemorrhage/bleed) Ventriculomegaly of the Head (verify not ventriculomegaly of the heart, do not report/code if Mild dilatation)	742.3
Q03.9	Congenital hydrocephalus, unspecified Unspecified hydrocephaly, NOS	742.3
Q04	**Other congenital malformations of the brain** **Excludes 1:** cyclopia (Q87.0), macrocephaly (Q75.3)	
Q04.0	Congenital malformations of corpus callosum Agenesis of corpus callosum Partial agenesis of the corpus callosum Other anomalies of the corpus callosum Absent corpus callosum Congenital malformation of corpus callosum Corpus callosum anomaly, congenital	742.2
Q04.1	Arhinencephaly	742.2
Q04.2	Holoprosencephaly Holoprosencephaly sequence	742.2
Q04.3	Other reduction deformities of the brain Absence of part of brain Agenesis of part brain Agyria Aplasia of part of brain Hydranencephaly Hypoplasia of part of brain Lissencephaly Microgyria Pachygyria	742.2

	Unspecified reduction defect of brain (try not to use) **Excludes 1**: congenital malformations of corpus callosum (Q04.0)	
Q04.4	Septo-optic dysplasia of brain	742.4
Q04.5	Megalencephaly Enlarged brain and/or head Macrocephaly Macroencephaly	742.4
Q04.6	Congenital cerebral cysts Porencephaly Schizencephaly Single cerebral cyst Porencephalic cysts **Excludes 1**: acquired porencephalic cyst (G93.0)	742.4
Q04.8	Other specified congenital malformation of brain Arnold-Chiari syndrome, type IV Macrogyria Cortical atrophy Cranial nerve defects Cystic encephalomalacia Encephalomalacia Anomalies of hypothalamus Anomalies of cerebellum, includes atrophy Other specified anomalies of brain Ulegyria Ventricular cysts Small brain	742.4
Q04.9	Congenital malformation of brain, unspecified Congenital anomaly NOS of brain Congenital deformity NOS of brain Congenital disease or lesion NOS of brain Multiple anomalies NOS of brain, congenital	742.9
Q05	**Spina bifida** **Includes:** hydromeningocele (spinal), meningocele (spinal), meningomyelocele, myelocele, myelomeningocele, rachischisis, spina bifida (aperta)(cystica), syringomyelocele **Excludes 1:** Arnold-Chiari syndrome, type II (Q07.0-), spina bifida occulta (Q76.0)	
Q05.0	Cervical spina bifida with hydrocephalus Cervical spina bifida with hydrocephalus and: • hydromeningocele • meningocele • meningomyelocele • myelocele • myelomeningocele • rachischisis • syringohydromyelia • lipomeningomyelocele • lipomyelomeningocele • lipomeningocele • spina bifida (aperta)(cystica)	741.01
Q05.1	Thoracic spina bifida with hydrocephalus Dorsal spina bifida Thoracolumbar spina bifida with hydrocephalus Thoracic/dorsal/thoracolumbar spina bifida with hydrocephalus and: • hydromeningocele • meningocele	741.02

	• meningomyelocele • myelocele • myelomeningocele • rachischisis • syringohydromyelia • lipomeningomyelocele • lipomyelomeningocele • lipomeningocele • spina bifida (aperta)(cystica)	
Q05.2	Lumbar spina bifida with hydrocephalus Lumbosacral spina bifida with hydrocephalus Lumbar/lumbosacral spina bifida with hydrocephalus and: • hydromeningocele • meningocele • meningomyelocele • myelocele • myelomeningocele • rachischisis • syringohydromyelia • lipomeningomyelocele • lipomyelomeningocele • lipomeningocele • spina bifida (aperta)(cystica)	741.03
Q05.3	Sacral spina bifida with hydrocephalus Sacral spina bifida with hydrocephalus and: • hydromeningocele • meningocele • meningomyelocele • myelocele • myelomeningocele • rachischisis • syringohydromyelia • lipomeningomyelocele • lipomyelomeningocele • lipomeningocele • spina bifida (aperta)(cystica)	741.03
Q05.4	Unspecified spina bifida with hydrocephalus Unspecified spina bifida with hydrocephalus and: • hydromeningocele • meningocele • meningomyelocele • myelocele • myelomeningocele • rachischisis • syringohydromyelia • lipomeningomyelocele • lipomyelomeningocele • lipomeningocele • spina bifida (aperta)(cystica)	741.00
Q05.5	Cervical spina bifida without hydrocephalus Cervical spina bifida without hydrocephalus, with: • hydromeningocele • meningocele	741.91

	• meningomyelocele • myelocele • myelomeningocele • rachischisis • syringohydromyelia • lipomeningomyelocele • lipomyelomeningocele • lipomeningocele • spina bifida (aperta)(cystica)	
Q05.6	Thoracic spina bifida without hydrocephalus Dorsal spina bifida, NOS Thoracolumbar spina bifida, NOS Thoracic/dorsal/thoracolumbar spina bifida without hydrocephalus, with: • hydromeningocele • meningocele • meningomyelocele • myelocele • myelomeningocele • rachischisis • syringohydromyelia • lipomeningomyelocele • lipomyelomeningocele • lipomeningocele • spina bifida (aperta)(cystica)	741.92
Q05.7	Lumbar spina bifida without hydrocephalus Lumbosacral spina bifida, NOS Lumbar/lumbosacral spina bifida without hydrocephalus, with: • hydromeningocele • meningocele • meningomyelocele • myelocele • myelomeningocele • rachischisis • syringohydromyelia • lipomeningomyelocele • lipomyelomeningocele • lipomeningocele • spina bifida (aperta)(cystica)	741.93
Q05.8	Sacral spina bifida without hydrocephalus Sacral spina bifida without hydrocephalus, with: • hydromeningocele • meningocele • meningomyelocele • myelocele • myelomeningocele • rachischisis • syringohydromyelia • lipomeningomyelocele • lipomyelomeningocele • lipomeningocele • spina bifida (aperta)(cystica)	741.90
Q05.9	Spina bifida unspecified Unspecified spina bifida without hydrocephalus, with:	741.90

	• hydromeningocele • meningocele • meningomyelocele • myelocele • myelomeningocele • rachischisis • syringohydromyelia • lipomeningomyelocele • lipomyelomeningocele • lipomeningocele • spina bifida (aperta)(cystica)	
Q06	**Other congenital malformations of spinal cord**	
Q06.0	Amyelia	742.59
Q06.1	Hypoplasia and dysplasia of spinal cord Atelomyelia Myelatelia Myelodysplasia of spinal cord	742.59
Q06.2	Diastematomyelia	742.51
Q06.3	Other congenital cauda equina malformations	742.59
Q06.4	Hydromyelia Hydrorachis	742.53
Q06.8	Other specified congenital malformations of spinal cord Congenital tethered cord Tethered filum terminale	742.59
Q06.9	Congenital malformation of spinal cord, unspecified Congenital anomaly NOS of spinal cord Congenital deformity NOS of spinal cord Congenital disease or lesion NOS of spinal cord	742.9
Q07	**Other congenital malformations of nervous system** **Excludes 2:** congenital central alveolar hypoventilation syndrome (G47.35), familial dysautonomia [Riley-Day] (G90.1), neurofibromatosis (nonmalignant) (Q85.0-)	
Q07.0	Arnold-Chiari syndrome (type II) **Excludes 1**: Arnold-Chiari syndrome, type III (Q01.-), Arnold-Chiari syndrome, type IV (Q04.8)	
Q07.00	Arnold-Chiari syndrome (type II) without spina bifida or hydrocephalus Arnold-Chiari type 2 without hydrocephalus	741.90
Q07.01	Arnold-Chiari syndrome (type II) with spina bifida Arnold-Chiari syndrome (type II) with spina bifida and: • hydromeningocele • meningocele • meningomyelocele • myelocele • myelomeningocele • rachischisis • syringohydromyelia • lipomeningomyelocele • lipomyelomeningocele • lipomeningocele • spina bifida (aperta)(cystica) ○ Specify (cervical, thoracic, dorsal, thoracolumbar, lumbar, lumbosacral, sacral, unspecified) for all of the above	741.90 741.91 741.92 741.93
Q07.02	Arnold-Chiari syndrome (type II) with hydrocephalus Arnold-Chiari Type 2 with hydrocephalus	742.0
Q07.03	Arnold-Chiari syndrome (type II) with spina bifida and hydrocephalus	741.00

	Arnold-Chiari syndrome (type II) with spina bifida, hydrocephalus and: • hydromeningocele • meningocele • meningomyelocele • myelocele • myelomeningocele • rachischisis • syringohydromyelia • lipomeningomyelocele • lipomyelomeningocele • lipomeningocele • spina bifida (aperta)(cystica) ○ Specify (cervical, thoracic, dorsal, thoracolumbar, lumbar, lumbosacral, sacral, unspecified) for all of the above	741.01 741.02 741.03
Q07.8	**Other specified congenital malformations of nervous system** Agenesis of nerve Displacement of brachial plexus Jaw-winking syndrome Marcus Gunn's syndrome Congenital optic nerve pit	742.8
Q07.9	**Congenital malformation of nervous system, unspecified** Congenital anomaly NOS of nervous system Congenital deformity NOS of nervous system Congenital disease or lesion NOS of nervous system	742.9

Congenital Malformations of Eye, Ear, Face and Neck (Q10-Q18)

ICD-10	Description	ICD-9
Q10	**Congenital malformation of eyelid, lacrimal apparatus and orbit** **Excludes 1:** cryptophthalmos NOS (Q11.2), cryptophthalmos syndrome (Q87.0)	
Q10.0	Congenital ptosis Blepharoptosis	743.61
Q10.1	Congenital ectropion	743.62
Q10.2	Congenital entropion	743.62
Q10.3	Other congenital malformations of eyelid Ablepharon Blepharophimosis, congenital Coloboma of eyelid Congenital absence or agenesis of eyelid Congenital accessory eyelid Congenital accessory eye muscle Telecanthus *(m) Congenital absence or agenesis of cilia* *(m) Congenital malformation of eyelid NOS* *(m) Small or narrow palpebral fissures* *(m) Absence of eyelashes* *(m) Fused eyelids (if birth weight greater than 1000gms)* *(m) Weakness of eyelids* *(m) Epicanthal folds* *(m) Slanted eyes* *(m) Upward eyeslant*	743.62 743.63
Q10.4	*(m) Absence and agenesis of lacrimal apparatus* *(m) Congenital absence of punctum lacrimal* *(m) Nasolacrimal duct obstruction*	743.65
Q10.5	*(m) Congenital stenosis and stricture of lacrimal duct* *(m) Congenital blocked tear duct* *(m) Congenital dacryostenosis*	743.65
Q10.6	*(m) Other congenital malformations of lacrimal apparatus* *(m) Congenital malformation of lacrimal apparatus NOS* *(m) Accessory lacrimal canal*	743.65
Q10.7	Congenital malformation of orbit Congenital anomaly of orbit	743.66
Q11	**Anophthalmos, microphthalmos and macrophthalmos**	
Q11.0	Cystic eyeball	743.03
Q11.1	Other anophthalmos Anophthalmos, NOS Agenesis of eye Aplasia of eye Congenital absence of the eye	743.00
Q11.2	Microphthalmos Cryptophthalmos Cryptophthalmos, NOS Dysplasia of eye Hypoplasia of eye Rudimentary eye Simple microphthalmos Microphthalmos associated with other anomalies of eye and adnexa **Excludes 1**: cryptophthalmos syndrome (Q87.0)	743.06 743.10 743.11 743.12
Q11.3	Macrophthalmos **Excludes 1**: macrophthalmos in congenital glaucoma (Q15.0)	743.8

Q12	Congenital lens malformations	
Q12.0	Congenital cataract **Please specify right, left or bilateral Cataract, NOS Cataract, anterior polar Capsular and subcapsular cataract Cortical and zonular cataract Anterior and posterior axial embryonal cataract Nuclear cataract Total and subtotal cataract, congenital Combined form cataract Cataract, other specified	 743.30 743.31 743.32 743.33 743.34
Q12.1	Congenital displaced lens Congenital ectopic lens Ectopia lentis	743.37
Q12.2	Coloboma of lens	743.36
Q12.3	Congenital aphakia Absence of lens	743.35
Q12.4	Spherophakia	743.36
Q12.8	Other congenital lens malformations Microphakia Anomalies of lens shape Other specified lens anomaly	743.36
Q12.9	Congenital lens malformation, unspecified Unspecified lens anomaly	743.39
Q13	**Congenital malformations of anterior segment of eye**	
Q13.0	Coloboma of iris Coloboma, NOS	743.46
Q13.1	Absence of iris Aniridia	743.45
Q13.2	Other congenital malformations of iris *(m) Anisocoria, congenital* *(m) Atresia of pupil* *(m) Congenital malformation of iris, NOS* *(m) Corectopia* *(m) Congenital anomaly of pupil* *(m) Dyscoria* *(m) Congenital heterochromia iridis* *(m) Ectopic pupil* *(m) Iridial remnants* *(m) Polycoria* *(m) Other specified anomalies of ciliary body* *(m) Brushfield spots*	743.46
Q13.3	Congenital corneal opacity Congenital arcus juvenilis Corneal opacities, interfering with vision, congenital	743.42
Q13.4	Other congenital corneal malformations (size and shape) Congenital malformations of cornea, NOS Microcornea Peter's anomaly Irido-corneo-trabecular dysgenesis *(m) Megalocornea (without glaucoma)* *(m) Congenital megalocornea (without glaucoma)* *(m) Enlarged cornea (without glaucoma)*	 743.22 743.41
Q13.5	*(m) Blue sclera*	743.47

Q13.8	**Other congenital malformations of anterior segment of eye**	
Q13.81	Rieger's anomaly Irido-trabecular dysgenesis Reigers anomaly Rieger eye malformation sequence	743.44
Q13.89	Other congenital malformations of anterior segment of eye Multiple and combined anomaly of anterior segment Other specified colobomas and anomalies of anterior segments *(m) Specified anomalies of sclera*	743.47 743.48 743.49
Q13.9	Congenital malformations of anterior segment of eye, unspecified Unspecified anomalies of anterior segments or anterior chamber	743.49
Q14	**Congenital malformations of posterior segment of eye** **Excludes 2:** optic nerve hypoplasia (H47.03-)	
Q14.0	Congenital malformation of vitreous humor Congenital vitreous opacity	743.51
Q14.1	Congenital malformations of retina Congenital retinal aneurysm Congenital macular changes Other retinal changes, congenital Specified anomaly of retina	743.55 743.56 743.58
Q14.2	Congenital malformations of optic disc Coloboma of optic disc Congenital optic disc coloboma Optic nerve pit, congenital Pit of optic disc Specified anomaly of optic disc	743.57
Q14.3	Congenital malformation of choroid Chorioretinal degeneration, congenital Coloboma of choroid Congenital choroidal folds	743.53
Q14.8	Other congenital malformations of posterior segment of eye Coloboma of the fundus Congenital folds and cysts of posterior segment Vascular anomalies	743.52 743.58
Q14.9	Congenital malformation of posterior segment of eye, unspecified Unspecified anomaly of posterior segment of eye	743.59
Q15	**Other congenital malformations of eye** **Excludes 1:** congenital nystagmus (H55.01), ocular albinism (E70.31-), optic nerve hypoplasia (H47.03-), retinitis pigmentosa (H35.52)	
Q15.0	Congenital glaucoma Axenfeld's anomaly Buphthalmos Glaucoma of childhood Glaucoma of newborn Hydrophthamos, simple, unspecified Keratoglobus, congenital, with glaucoma Macrocornea with glaucoma Macrophthalmos in congenital glaucoma Megalocornea with glaucoma Enlarged eye, NOS Simple buphthalmos Buphthalmos associated with other ocular anomalies Congenital glaucoma, simple	743.20 743.21 743.22 743.44
Q15.8	Other specified congenital malformations of eye Epibulbar dermoid cyst	743.8
Q15.9	Congenital malformation of eye, unspecified	743.9

	Congenital anomaly of eye Congenital deformity of eye	
Q16	**Congenital malformations of ear causing impairment of hearing** **Excludes 1:** congenital deafness (H90.-)	
Q16.0	Congenital absence of (ear) auricle Absence of external ear	744.01
Q16.1	Congenital absence, atresia and stricture of auditory canal (external) Congenital atresia or stricture of osseous meatus Absence of auditory canal Absence of external auditory meatus *(m) Narrow external auditory meatus*	744.02
Q16.2	Absence of Eustachian tube	744.24
Q16.3	Congenital malformation of ear ossicles Congenital fusion of ear ossicles	744.04
Q16.4	Other congenital malformations of middle ear Congenital malformations of middle ear, NOS	744.03
Q16.5	Congenital malformation of inner ear Congenital anomaly of membranous labyrinth Congenital anomaly of organ of Corti	744.05
Q16.9	Congenital malformation of ear causing impairment of hearing, unspecified Congenital absence of ear, NOS	744.00
Q17	**Other congenital malformations of ear Excludes 1:** congenital malformations of ear with impairment of hearing (Q16.0-Q16.9), preauricular sinus (Q18.1)	
Q17.0	*(m) Accessory auricle* *(m) Accessory tragus* *(m) Polyotia* *(m) Preauricular appendage or tag* *(m) Supernumerary ear* *(m) Supernumerary lobule* *(m) Ear, double lobule* *(m) Other appendage, tag, or lobule* *(m) Papillomas, ear tags* *(m) Skin tag of pinna*	744.1
Q17.1	*(m) Macrotia (enlarged pinna)*	744.22
Q17.2	Microtia (hypoplastic pinna)	744.23
Q17.3	*(m) Other misshapen ear* *(m) Pointed ear* *(m) Lop ear* *(m) Absent or decreased cartilage* *(m) Cauliflower ear* *(m) Cauliflower ears* *(m) Cleft ear* *(m) Cupped ears* *(m) Ears, absent tragus* *(m) Ears, bridged concha* *(m) Ears, lack of helical fold* *(m) Ears, thickened or over folded helix* *(m) Elfin* *(m) Elfin ears* *(m) Extra cartilage* *(m) Malformed ear* *(m) Notch in ear* *(m) Pixie like* *(m) Protruding ears* *(m) Ridge ear*	744.29

	(m) Posteriorly rotated ears *(m) Crumpled ears* *(m) Darwin's tubercle*	
Q17.4	*(m) Misplaced ear* *(m) Low-set ears* **Excludes 1**: cervical auricle (Q18.2)	744.29
Q17.5	*(m) Prominent ear* *(m) Bat ear*	744.29
Q17.8	Other specified anomalies of Eustachian tube *(m) Other specified congenital malformations of ear* *(m) Congenital absence of lobe of ear* *(m) Congenital split ear lobes* *(m) Other specified anomaly of ear without impairment of hearing*	744.21 744.24 743.29
Q17.9	*(m) Congenital malformation of ear, unspecified* *(m) Congenital anomaly of ear, NOS*	744.3
Q18	**Other congenital malformations of face and neck Excludes 1:** 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] (M26.-), malformation syndromes affecting facial appearance (Q87.0), persistent thyroglossal duct (Q89.2)	
Q18.0	*(m) Sinus, fistula and cyst of branchial cleft* *(m) Branchial vestage* *(m) Branchial cleft fistula* *(m) Branchial cleft sinus* *(m) Branchial cleft cyst* *(m) Branchial cleft pit*	744.41
Q18.1	*(m) Preauricular sinus and cyst* *(m) Fistula of auricle, congenital* *(m) Cervicoaural fistula* *(m) Ear pit* *(m) Preauricalar fistula* *(m) Preauricular pits* *(m) Preauricular sinus*	744.46 744.47
Q18.2	*(m) Other branchial cleft malformations* *(m) Branchial cleft malformations, NOS* *(m) Cervical auricle* *(m) Otocephaly* *(m) Dermal sinus of head* *(m) Other branchial cleft anomaly*	744.43 744.49
Q18.3	*(m) Webbing of neck* *(m) Pterygium colli* *(m) Redundant neck skin folds*	744.5
Q18.4	Macrostomia	744.83
Q18.5	Microstomia	744.84
Q18.6	*(m) Macrocheilia (large lips)* *(m) Hypertrophy of lip, congenital*	744.81
Q18.7	*(m) Microcheilia*	744.82
Q18.8	Other specified congenital malformations of face and neck Medial cyst of face and neck Medial fistula of face and neck Medial sinus of face and neck Cleft face (excluding cleft lip and palate) *(m) Short neck*	744.89

Q18.9	Congenital malformations of face and neck, unspecified Congenital anomaly NOS of face and neck *(m) Abnormal facies* *(m) Craniofacial dysmorphosis* *(m) Dysmorphic facies*	744.89 744.9

Congenital Malformations of the Circulatory System (Q20-Q28)

ICD-10	Descriptions	ICD-9
Q20	**Congenital malformations of cardiac chambers and connections** **Excludes 1:** dextrocardia with situs inversus (Q89.3), mirror-image atrial arrangement with situs inversus (Q89.3)	
Q20.0	Common arterial trunk Persistent truncus arteriosus Absent septum between aorta and pulmonary artery Common aortopulmonary trunk **Excludes 1:** aortic septal defect (Q21.4)	745.0
Q20.1	Double outlet right ventricle Taussig-Bing syndrome Origin of both great vessels from right ventricle	745.11
Q20.2	Double outlet left ventricle	745.19
Q20.3	Discordant ventriculoarterial connection Dextro-transposition of aorta Transposition of great vessels (complete) D-transposition	745.10
Q20.4	Double inlet ventricle (right or left) Common ventricle Cor triloculare biatriatum Single ventricle	745.3
Q20.5	Discordant atrioventricular connection Corrected transposition Levo-transposition Ventricular inversion L-transposition, ventricular inversion Other specific transposition of great vessels Unspecified transposition of great vessels	745.12
Q20.6	Isomerism of atrial appendages Isomerism of atrial appendages with asplenia or polysplenia	745.8 759.0
Q20.8	Other congenital malformations of cardiac chambers and connections Cor binoculare Absence of atrial and ventricular septa Two chambered heart	745.19 745.7 745.8
Q20.9	Congenital malformation of cardiac chambers and connections, unspecified	746.9
Q21	**Congenital malformations of cardiac septa** **Excludes 1:** acquired cardiac septal defect (I51.0)	
Q21.0	Ventricular septal defect (VSD) Roger's disease Other specified ventricular septal defect (see VSD synonym list following section Q28.9) Perimembranous type II (Gerbode) Muscular, type VI Malalignment type I (subarterial, subaortic, subpulmonic (see VSD synonym list following section Q28.9) Interventricular septal defect Ventricular septal defect, NOS	745.4

Q21.1	Atrial septal defect Coronary sinus defect Patent or persistent ostium secundum defect (type II) Atrial septal defect, NOS Auricular septal defect, NOS Other specified atrial septal defect Secundum atrial septal defect *(m) Patent or persistent foraman ovale* *(m) Patent or persistent sinus venosus defect* *(m) Fossa ovalis defect* *(m) Nonclosure of foramen ovale, NOS* *(m) Foramen ovale* *(m) Partial foramen ovale*	745.5
Q21.2	Atrioventricular septal defect Common atrioventricular canal Endocardial cushion defect Ostium primum atrial septal defect (ASD) (type I) Ostium primum ASD Ostium primum defects Persistent ostium primum Absence of atrial septum Single common atrium Artrioventricular canal type VSD Common atrioventricular canal with VSD Other endocardial cushion defects Type III VSD, Canal-type VSD	745.60 745.61 745.69
Q21.3	Tetralogy of Fallot (ventricular septal defect with pulmonary stenosis or atresia, dextroposition of aorta and hypertrophy of right ventricle)	745.2
Q21.4	Aortopulmonary septal defect Aortic septal defect Aortopulmonary window Abnormal communication between aorta and pulmonary artery	745.0
Q21.8	Other congenital malformations of cardiac septa Eisenmenger's defect Pentalogy of Fallot Fallot's tetralogy plus atrial septal defect **Excludes 1:** Eisenmenger's complex (I27.8), Eisenmenger's syndrome (I27.8)	745.8 745.2
Q21.9	Congenital malformations of cardiac septum, unspecified Septal (heart) defect, NOS	745.9
Q22	**Congenital malformations of pulmonary and tricuspid valves**	
Q22.0	Pulmonary valve atresia Congenital absence of pulmonary valve	746.01
Q22.1	Congenital pulmonary valve stenosis	746.02
Q22.2	Congenital pulmonary valve insufficiency (do not code if mild, trivial or physiologic) Congenital pulmonary valve regurgitation (do not code if mild, trivial or physiologic) Trilogy of Fallot	746.09
Q22.3	Other congenital malformations of pulmonary valve Congenital malformation of pulmonary valve, NOS Supernumerary cusps of pulmonary valve Quadricuspid pulmonary valve Unicuspid pulmonary valve Pulmonary valve hypoplasia Bicuspid pulmonary valve	746.00 746.09

	Other specified anomaly of pulmonary valve	
Q22.4	Congenital tricuspid stenosis Congenital tricuspid atresia Absence of tricuspid valve Tricuspid hypoplasia	746.1
Q22.5	Ebstein's anomaly Ebstein's anomaly of tricuspid valve	746.2
Q22.6	Hypoplastic right heart syndrome	756.89
Q22.8	Other congenital malformations of tricuspid valve Congenital insufficiency of tricuspid valve Congenital tricuspid valve regurgitation	746.89
Q22.9	Congenital malformations of tricuspid valve, unspecified	746.89
Q23	**Congenital malformations of aortic and mitral valves**	
Q23.0	Congenital stenosis of aortic valve Congenital aortic atresia Congenital aortic stenosis, NOS	746.3
Q23.1	Congenital insufficiency of aortic valve (do not code if mild, trivial or physiologic) Bicuspid aortic valve Congenital aortic insufficiency Aortic valve regurgitation, congenital	746.4
Q23.2	Congenital mitral stenosis Congenital mitral atresia Congenital supravalvular mitral stenosis Absence of mitral valve Atresia of mitral valve Hypoplasia of mitral valve/cleft mitral valve	746.5
Q23.3	Congenital mitral insufficiency (do not code if mild, trivial or physiologic) Mitral valve prolapse Mitral valve regurgitation	746.6
Q23.4	Hypoplastic left heart syndrome	746.7
Q23.8	Other congenital malformations of aortic and mitral valves Cleft leaflet of mitral valve Fused commissure of mitral valve Parachute deformity of mitral valve Supernumerary cusps Aortic valve atresia/absence Other specified anomaly of aortic valves	746.89
Q23.9	Congenital malformation of aortic and mitral valves, unspecified Unspecified anomaly of the aortic valves	746.89
Q24	**Other congenital malformations of heart** **Excludes 1:** endocardial fibroelastosis (I42.4)	
Q24.0	Dextrocardia Dextrocardia with no mention of situs inversus Dextrocardia without situs inversus (situs solitus) **Excludes 1:** 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)	746.87
Q24.1	Levocardia	746.87
Q24.2	Cor triatriatum	746.82
Q24.3	Pulmonary infundibular stenosis Subvalvular pulmonic stenosis	746.83
Q24.4	Congenital subaortic stenosis Congenital subvalvular aortic stenosis	746.81
Q24.5	Malformation of coronary vessels Congenital coronary (artery) aneurysm Congenital coronary artery fistula	746.85

	Absence of coronary artery Anomalies of coronary artery or sinus Anomalous origin or communication of coronary artery Coronary artery arising from aorta or pulmonary trunk Single coronary artery	
Q24.6	Congenital heart block Complete or incomplete atrioventricular (AV) block	746.86
Q24.8	Other specified congenital malformations of heart Congenital diverticulum of left ventricle Congenital malformation of myocardium Congenital malformation of pericardium Malposition of heart Uhl's disease (hypoplastic right ventricle) Shones syndrome (Complex) = 1) Parachute Mitral Valve; 2) supravalvular ring of L atrium; 3) subaortic stenosis; 4) coarctation of aorta [Code the stenosis and coarctation separately] Hypoplastic left ventricle Hypoplastic ventricle, NOS Obstructive anomalies of heart, NEC Abdominal heart Ectopia (ectopic) cordis (mesocordia) Anomalies of pericardium Congenital pericardial defect Anomalies of myocardium Cardiomyopathy, congenital Other specified anomaly of heart Ventricular hypertrophy (right or left) Other defects of the atria Atresia of cardiac vein Hypoplasia of cardiac vein Absence of papillary muscle of luthisis Anomaly of papillary muscle Conduction defects, NOS *(m) Cardiomyopathy, hypertrophic* *(m) Congenital cardiomegaly*	746.84 746.87 746.89
Q24.9	Congenital malformations of heart, unspecified Congenital anomaly of heart Congenital disease of heart	746.9
Q25	**Congenital malformations of great arteries**	
Q25.0	Patent ductus arteriosus (PDA) (do not report if birthweight less than 1500 grams) Patent ductus Botallo Persistent ductus arteriosus Patent ductus arteriosis closed with medication	747.0
Q25.1	Coarctation of aorta Coarctation of aorta (preductal) (postductal) Unspecified coarctation of aorta	747.10
Q25.2	Atresia of aorta Interrupted aortic arch Interrupted aortic arch. Type A Interrupted aortic arch. Type B	747.11
Q25.3	Supravalvular aortic stenosis Stenosis of aorta Stricture of aorta Small aorta, narrowing of aorta	747.22

	Excludes 1: congenital aortic stenosis NOS (Q23.0); congenital stenosis of aortic valve (Q23.0)	
Q25.4	Other congenital malformations of aorta Absence of aorta Aneurysm of sinus of Valsalva (ruptured) Aplasia of aorta Congenital aneurysm of aorta Congenital malformations of aorta Congenital dilation of aorta Double aortic arch [vascular ring of aorta] Hypoplasia of aorta Persistent convolutions of aortic arch Persistent right aortic arch Hypoplastic aortic isthmas Tubular hypoplasia of aorta Vascular ring compression of trachea Dextroposition of aorta Overridding aorta Kommerell's diverticulum Aberrant subclavian artery Anomalous origin, right subclavian artery Pseudotruncus arteriousus **Excludes 1:** hypoplasia of aorta in hypoplastic left heart syndrome (Q23.4)	747.20 747.21 747.29
Q25.5	Atresia of pulmonary artery Pulmonary artery absence Pulmonary artery agenesis Pulmonary artery atresia	747.31
Q25.6	Stenosis of pulmonary artery Supravalvular pulmonary stenosis *(m) Peripheral pulmonary artery stenosis, do not code if only defect or preterm infant*	747.31 747.39
Q25.7	Other congenital malformations of pulmonary artery	
Q25.71	Coarctation of pulmonary artery	747.31
Q25.72	Congenital pulmonary arteriovenous malformation Congenital pulmonary arteriovenous aneurysm Aneurysm of pulmonary artery	747.32
Q25.79	Other congenital malformations of pulmonary artery Aberrant pulmonary artery Agenesis of pulmonary artery Congenital aneurysm of pulmonary artery Congenital anomaly of pulmonary artery Hypoplasia of pulmonary artery Dilatation of pulmonary artery Unspecified anomaly of pulmonary artery	747.39
Q25.8	Other congenital malformations of other great arteries	
Q25.9	Congenital malformations of great arteries, unspecified	747.29
Q26	**Congenital malformations of great veins**	
Q26.0	Congenital stenosis of vena cava Congenital stenosis of vena cava (inferior) (superior)	747.49
Q26.1	Persistent left superior vena cava	747.49
Q26.2	Total anomalous pulmonary venous connection Total anomalous pulmonary venous return [TAPVR], subdiaphragmatic Total anomalous pulmonary venous return [TAPVR], supradiaphragmatic	747.41
Q26.3	Partial anomalous pulmonary venous connection Partial anomalous pulmonary venous return	747.42

Q26.4	Anomalous pulmonary venous connection, unspecified	747.42
Q26.5	Anomalous portal venous connection	747.61
Q26.6	Portal vein-hepatic artery fistula	747.61
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 Stenosis of inferior vena cava Stenosis of superior vena cava Anomalous portal vein termination Portal vein-hepatic artery fistula Transposition of pulmonary veins, NOS	747.49
Q26.9	Congenital malformation of great vein, unspecified Congenital anomaly of vena cava (inferior) (superior), NOS Anomaly NOS of pulmonary veins	747.40
Q27	**Other congenital malformations of peripheral vascular system** **Excludes 2:** anomalies of cerebral and precerebral vessels (Q28.0-Q28.3), anomalies of coronary vessels (Q24.5), anomalies of pulmonary artery (Q25.5-Q25.7), congenital retinal aneurysm (Q14.1), hemangioma and lymphangioma (D18.-)	
Q27.0	*(m) Congenital absence and hypoplasia of umbilical artery* *(m)* Single umbilical artery *(m) Two vessels in umbilical cord*	747.5
Q27.1	Congenital renal artery stenosis	747.62
Q27.2	Other congenital malformations of renal artery Congenital malformation of renal artery, NOS Multiple renal arteries Other anomaly of renal artery Renal vessel anomaly	747.62
Q27.3	Arteriovenous malformation (peripheral) Arteriovenous aneurysm	747.69
Q27.30	Arteriovenous malformation, site unspecified	747.69
Q27.31	Arteriovenous malformation of vessel of upper limb	747.63
Q27.32	Arteriovenous malformation of vessel of lower limb	747.64
Q27.33	Arteriovenous malformation of digestive system vessel	747.61
Q27.34	Arteriovenous malformation of renal vessel	747.62
Q27.39	Arteriovenous malformation, other site	747.69
Q27.4	Congenital phlebectasia	747.89
Q27.8	Other specified congenital malformations of peripheral vascular system Absence of peripheral vascular system Atresia of peripheral vascular system Congenital aneurysm (peripheral) Congenital stricture, artery Congenital varix Absence of artery, NEC Atresia of artery Other anomaly of peripheral arteries Other anomaly of peripheral veins Anomaly of spinal vessel Gastrointestinal vessel anomaly **Excludes 1:** arteriovenous malformation (Q27.3-)	747.69
Q27.9	Congenital malformation of peripheral vascular system, unspecified Anomaly of artery or vein, NOS	747.60
Q28	**Other congenital malformations of circulatory system**	

	Excludes 1: congenital aneurysm NOS (Q27.8), congenital coronary aneurysm (Q24.5), ruptured cerebral arteriovenous malformation (I60.8), ruptured malformation of precerebral vessels (I72.0) **Excludes 2:** congenital peripheral aneurysm (Q27.8), congenital pulmonary aneurysm (Q25.79), congenital retinal aneurysm (Q14.1)	
Q28.0	Arteriovenous malformation of precerebral vessels Congenital arteriovenous precerebral aneurysm (non-ruptured)	747.89
Q28.1	Other malformations of precerebral vessels	747.89
Q28.2	Arteriovenous malformation of cerebral vessels Arteriovenous malformation of brain, NOS Congenital arteriovenous cerebral aneurysm (nonruptured) Anomalies of cerebrovascular system Other anomaly of cerebral vessels Anomalies of vein of Galen	747.81
Q28.3	Other malformations of cerebral vessels Congenital cerebral aneurysm (nonruptured) Congenital malformation of cerebral vessels, NOS Developmental venous anomaly	747.81
Q28.8	Other specified congenital malformations of circulatory system Congenital aneurysm, specified site NEC Spinal vessel anomaly	747.89 747.82
Q28.9	Congenital malformation of circulatory system, unspecified	747.9

VSD Synonyms List

- *VSD, Malalignment (type I, subarterial, subaortic, subpulmonic)*
  - *Synonyms: outlet, supracristal*
- *VSD, Perimembranous (type II, membranous)*
  - *Synonyms: membranous, infracristal, subaortic, Gerbode*
- *VSD, Endocardial cushion defect (type III)*
  - *Synonyms: inlet, inflow type, subtricuspid, canal-type*
- *VSD, Muscular (type IV)*
  - *Synonyms: travecular, swiss cheese, central, apical, marginal, Roger's disease*

Congenital Malformations of the Respiratory System (Q30-Q34)

ICD-10	Description	ICD-9
Q30	**Congenital malformations of nose Excludes 1:** congenital deviation of nasal septum (Q67.4)	
Q30.0	Choanal atresia Atresia of nares (anterior) (posterior) Congenital stenosis of nares (anterior) (posterior) Choanal stenosis	748.0
Q30.1	Agenesis and underdevelopment of nose Congenital absent of nose Agenesis of nose Congenital absence of nose	748.1
Q30.2	Fissured, notched and cleft nose Cleft nose anomaly Congenital cleft nose Congenital cleft nose anomaly Midline fissured, notched and cleft nose	748.1
Q30.3	*(m) Congenital perforated nasal septum*	748.1
Q30.8	Other congenital malformations of nose Accessory nose *(m) Congenital anomaly of nasal sinus wall* *(m) Congenital malformation of nasal septum* *(m) Absent nasal septum* *(m) Anteverted nares* *(m) Flat or wide nasal bridge or other minor nose malformation* *(m) Notched or hypoplastic alae nasi* *(m) Other anomalies of nose* *(m) Small nares* *(m) Small nose and nostril* *(m) Upturned nose* *(m) Wide nasal bridge* *(m) Tubular nose, single nostril, proboscis*	748.1
Q30.9	*(m) Congenital malformation of nose, unspecified* *(m) Congenital anomaly of the nose*	748.1
Q31	**Congenital malformation of larynx Excludes 1:** congenital laryngeal stridor, NOS (P28.89)	
Q31.0	Web of larynx Glottic web of larynx Subglottic web of larynx Web of larynx, NOS Laryngeal web Subglottic web	748.2
Q31.1	Congenital subglottic stenosis	748.3
Q31.2	Laryngeal hypoplasia	748.3
Q31.3	*(m) Laryngocele*	748.3
Q31.5	*(m) Congenital laryngomalacia*	748.3
Q31.8	Other congenital malformations of larynx Absence of larynx Agenesis of larynx Atresia of larynx Congenital cleft thyroid cartilage Congenital cleft larynx Laryngeal cleft Congenital fissure of epiglottis	748.3

	Congenital stenosis of larynx, NEC Posterior cleft of cricoid cartilage Cleft larynx, laryngotracheoesophageal cleft Cleft thyroid, cartilage, congenital Anomalies of larynx and supporting cartilage Anomaly of epiglottis Atresia of epiglottis Atresia glottis Anomaly of thyroid cartilage	
Q31.9	Congenital malformation of larynx, unspecified	748.3
Q32	**Congenital malformations of trachea and bronchus** **Excludes 1:** congenital bronchiectasis (Q33.4)	
Q32.0	*(m) Congenital tracheomalacia*	748.3
Q32.1	Other congenital malformations of trachea Atresia of trachea Congenital anomaly of tracheal cartilage Congenital dilation of trachea Congenital malformation of trachea Congenital stenosis of trachea Congenital tracheocele Congenital laryngotracheal anomaly Diverticulum of trachea Rudimentary tracheal bronchus	748.3
Q32.2	Congenital bronchomalacia	748.3
Q32.3	Congenital stenosis of bronchus	748.3
Q32.4	Other congenital malformations of bronchus Absence of bronchus Agenesis of bronchus Atresia of bronchus Congenital diverticulum of bronchus Congenital malformations of bronchus, NOS	748.3
Q33	**Congenital malformations of lung**	
Q33.0	Congenital cystic lung Congenital cystic lung disease Congenital honeycomb lung Congenital polycystic lung disease Bronchogenic cyst Lung cyst, NOS Single cyst, lung Cystic lung, NOS Multiple cysts, lung Polycystic lung (do not use this code for broncopulmonary dysplasia) CCAM (congenital cystic adenomatoid malformation) Cystic adenomatoid dysplasia of lung Cystic adenomatoid lung Lobar adenomatosis Other specified congenital cystic lung **Excludes 1:** Cystic Fibrosis (E84.0), cystic lung disease, acquired or unspecified (J98.4)	748.4
Q33.1	Accessory lobe of lung Azygos lobe (fissured), lung Accessory lung	748.69
Q33.2	Sequestration of lung Congenital sequestration of lung Pulmonary sequestration	748.5

Q33.3	Agenesis of lung Congenital absence of lung (lobe) Congenital absence of lung	748.5
Q33.4	Congenital bronchiectasis	748.61
Q33.5	Ectopic tissue in lung	748.69
Q33.6	Congenital hypoplasia and dysplasia of lung Congenital pulmonary dysplasia Hypoplasia of lobe of lung Hypoplasia of lung Pulmonary hypoplasia Other specified dysplasia of lung Unspecified dysplasia of lung **Excludes 1:** pulmonary hypoplasia associated with short gestation (P28.0)	748.5
Q33.8	Other congenital malformations of lung Bilobar right lung Right lung with left lung bronchial pattern Trilobal left lung Aplasia of lung Fusion of lobes of lung Congenital lobar emphysema Lymphangiectasia of lungs	748.69
Q33.9	Congenital malformations of lung, unspecified Congenital anomaly of lung	748.60
Q34	**Other congenital malformations of respiratory system** **Excludes 2:** congenital central alveolar hypoventilation syndrome (G47.35)	
Q34.0	Anomaly of pleura Congenital anomaly of pleural folds Abnormal communication between pericardial/pleural sacs	748.8
Q34.1	Congenital cyst of mediastinum	748.8
Q34.8	Other specified congenital malformation of respiratory system Atresia of nasopharynx Immotile cilia syndrome Primary ciliary dyskinesia	748.8
Q34.9	Congenital malformation of respiratory system, unspecified Congenital absence of respiratory system Congenital anomaly of respiratory system, NOS	748.9

Congenital Malformations of the Cleft Lip and Cleft Palate (Q35-Q37)

ICD-10	Description	ICD-9
Q35	**Cleft palate Excludes 1:** cleft palate with cleft lip (Q37.-)	
Q35.1	Cleft hard palate Cleft hard palate, central Cleft hard palate, NOS Cleft hard palate, unilateral, left Cleft hard palate, unilateral, right Cleft hard palate, unilateral Cleft hard palate, bilateral	749.00
Q35.3	Cleft soft palate Cleft palate, submucous Submucous cleft palate Cleft soft palate alone, central Cleft soft palate alone, NOS Cleft soft palate alone, unilateral, left Cleft soft palate alone, unilateral, right Cleft soft palate alone, unilateral Cleft soft palate alone, bilateral	749.00
Q35.5	Cleft hard palate with cleft soft palate Complete cleft palate, unilateral, right Complete cleft palate, unilateral Complete cleft palate, bilateral Complete cleft palate, unilateral, left Complete cleft palate, central	749.01 749.03
Q35.7	*(m) Cleft uvula* *(m) Bifid uvula*	749.02
Q35.9	Cleft palate, unspecified Cleft palate, NOS Palatoschisis	749.00 749.02 749.04
Q36	**Cleft lip** **Includes:** cheiloschisis, congenital fissure of lip, harelip, labium leporinum **Excludes 1:** cleft lip with cleft palate (Q37.-)	
Q36.0	Cleft lip, bilateral Complete cleft lip, bilateral Incomplete cleft lip, bilateral	749.13 749.14
Q36.1	Cleft lip, median Central cleft lip	749.10
Q36.9	Cleft lip, unilateral Cleft lip, NOS Cleft lip, unilateral, right Cleft lip, unilateral, left Cleft gingiva Cleft gum Cleft lip, unspecified Incomplete form of cleft lip (microform) Complete cleft lip, unilateral, left Complete cleft lip, unilateral, right Complete cleft lip, unilateral Incomplete cleft lip, unilateral, left Incomplete cleft lip, unilateral, right Incomplete cleft lip, unilateral	749.10 749.11 complete 749.12 incomplete
Q37	**Cleft palate with cleft lip** **Includes:** Cheilopalatoschisis	

Q37.0	Cleft hard palate with bilateral cleft lip	749.24 749.25
Q37.1	Cleft hard palate with unilateral cleft lip Cleft hard palate with cleft lip, NOS	749.22 749.25
Q37.2	Cleft soft palate with bilateral cleft lip	749.24 749.25
Q37.3	Cleft soft palate with unilateral cleft lip Cleft soft palate with cleft lip, NOS	749.22 749.25
Q37.4	Cleft hard and soft palate with bilateral cleft lip Complete cleft palate and bilateral cleft lip	749.23 749.24
Q37.5	Cleft hard and soft palate with unilateral cleft lip Cleft hard and soft palate with cleft lip, NOS Complete cleft palate and cleft lip, unilateral, left Complete cleft palate and cleft lip, unilateral, right Complete cleft palate and cleft lip, unilateral Complete cleft palate and cleft lip, NOS	749.21 749.22
Q37.8	Unspecified cleft palate with bilateral cleft lip Incomplete cleft palate and bilateral cleft lip Bilateral Cleft lip, with any cleft palate	749.24
Q37.9	Unspecified cleft palate with unilateral cleft lip Cleft palate with cleft lip, NOS Cheilopalatoschisis Cleft palate with cleft lip Cleft palate with cleft lip, unspecified Other combinations cleft palate and lip	749.20

Congenital Malformations of the Digestive System (Q38-Q45)

ICD-10	Descriptions	ICD-9
Q38	**Other congenital malformations of tongue, mouth and pharynx** **Excludes 1:** dentofacial anomalies (M26.-), macrostomia (Q18.4), microstomia (Q18.5)	
Q38.0	Congenital malformations of lips, NEC Van der Woude syndrome *(m) Congenital malformation of lip, NOS* *(m) Congenital fistula of lip or pits* *(m) Long philtrum (code if with 760.71 or 760.79)* *(m) Notched lip (code if with 760.71 or 760.79)* *(m) Other lip anomaly (code if with 760.71 or 760.79)* *(m) Prominent philtrum (code if with 760.71 or 760.79)* **Excludes 1:** cleft lip (Q36.-), cleft lip with cleft palate (Q37.-), macrocheilia (Q18.6), microcheilia (Q18.7)	750.25
Q38.1	*(m) Ankyloglossia* *(m) Tongue tie* *(m) Short frenulum of tongue* *(m) Aberrant frenula*	750.0
Q38.2	*(m) Macroglossia (large tongue)* *(m) Congenital hypertrophy of tongue*	750.15
Q38.3	Other congenital malformations of tongue Aglossia (absence of tongue) *(m) Bifid tongue* *(m) Congenital adhesion of tongue* *(m) Congenital fissure of tongue* *(m) Congenital malformation of tongue, NOS* *(m) Double tongue* *(m) Hypoglossia (small tongue)* *(m) Hypoplasia of tongue* *(m) Microglossia* *(m) Congenital abnormal tongue position anomaly* *(m) Congenital anomaly of tongue* *(m) Dislocation or displacement of tongue* *(m) Glossoptosis* *(m) Cleft tongue or split tongue*	750.10 750.11 750.12 750.13 750.16 750.19
Q38.4	Congenital malformations of salivary glands and ducts Atresia of salivary glands and ducts Congenital absence of salivary glands and ducts Congenital accessory salivary glands and ducts Congenital fistula of salivary gland Congenital ranula Congenital salivary gland anomaly Imperforate salivary duct Other anomaly of salivary glands or ducts	750.21 750.22 750.23 750.24
Q38.5	Congenital malformations of palate, NEC *(m) Congenital absence of uvula* *(m) Congenital malformation of palate, NOS* *(m) Congenital high arched palate* **Excludes 1:** cleft palate (Q35.-), cleft palate with cleft lip (Q37.-)	750.26
Q38.6	*(m) Other congenital malformations of mouth* *(m) Congenital malformations of mouth, NOS* *(m) Fordyce spots of mouth* *(m) Fordyce's disease*	750.26
Q38.7	Congenital pharyngeal pouch	750.27

	Congenital diverticulum of pharynx **Excludes 1:** pharyngeal pouch syndrome (D82.1)	
Q38.8	Other congenital malformations of pharynx Congenital malformation of pharynx, NOS Imperforate pharynx Congenital anomaly of pharynx Congenital velopharyngeal dysfunction Congenital velopharyngeal impairment Congenital velopharyngeal incompetence	750.29
Q39	**Congenital malformations of esophagus**	
Q39.0	Atresia of esophagus without fistula Atresia of esophagus, NOS Imperforate esophagus	750.3
Q39.1	Atresia of esophagus with tracheoesophageal fistula Atresia of esophagus with broncho-esophageal fistula Esophageal atresia with tracheoesophageal fistula	750.3
Q39.2	Congenital trachea-esophageal fistula without atresia Congenital trachea-esophageal fistula, NOS T E fistula without esophageal atresia Tracheoesophageal fistula “h” type	750.3
Q39.3	Congenital stenosis and stricture of esophagus Congenital esophageal ring Congenital Schatzkis ring Congenital stenosis of esophagus	750.3
Q39.4	Esophageal web	750.3
Q39.5	Congenital dilatation of esophagus Congenital cardiospasm Congenital achalasia Giant esophagus	750.4
Q39.6	Congenital diverticulum of esophagus Congenital esophageal pouch	750.4
Q39.8	Other congenital malformations of esophagus Congenital absence of esophagus Congenital displacement of esophagus Congenital duplication of esophagus Esophageal duplication Other tracheoesophageal anomaly	750.4
Q39.9	Congenital malformation of esophagus, unspecified	750.4
Q40	**Other congenital malformations of upper alimentary tract**	
Q40.0	Congenital hypertrophic pyloric stenosis Congenital or infantile constriction Congenital or infantile hypertrophy Congenital or infantile spasm Congenital of infantile stenosis Congenital or infantile stricture Other congenital pyloric obstruction	750.5
Q40.1	Congenital hiatus hernia Congenital hiatal hernia Congenital displacement of cardia through esophageal hiatus **Excludes 1**: congenital diaphragmatic hernia (Q79.0)	750.6
Q40.2	Other specified congenital malformations of stomach Congenital displacement of stomach Congenital diverticulum of stomach Congenital hourglass stomach Congenital duplication of stomach	750.7

	Megalogastria Microgastria Displacement or transposition of stomach	
Q40.3	Congenital malformation of stomach, unspecified Congenital gastric anomaly	750.7
Q40.8	Other specified congenital malformations of upper alimentary tract	750.8
Q40.9	Congenital malformation of upper alimentary tract, unspecified Congenital anomaly of upper alimentary tract Congenital deformity of upper alimentary tract Congenital upper GI tract anomaly	750.9
Q41	**Congenital absence, atresia and stenosis of small intestine** **Includes:** congenital obstruction, occlusion or stricture of small intestine or intestine, NOS **Excludes 1:** cystic fibrosis with intestinal manifestation (E84.11), meconium ileus NOS (without cystic fibrosis) (P76.0)	
Q41.0	Congenital absence, atresia and stenosis of duodenum	751.1
Q41.1	Congenital absence, atresia and stenosis of jejunum Apple peel syndrome Imperforated jejunum	751.1
Q41.2	Congenital absence, atresia and stenosis of ileum	751.1
Q41.8	Congenital absence, atresia and stenosis of other specified parts of small intestine	751.1
Q41.9	Congenital absence, atresia and stenosis of small intestine, part unspecified Absence of small intestine with fistula Atresia of small intestine with fistula Stenosis of small intestine with fistula	751.1
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	751.2
Q42.1	Congenital absence, atresia and stenosis of rectum without fistula Imperforated rectum	751.2
Q42.2	Congenital absence, atresia and stenosis of anus with fistula Imperforated anus with fistula Congenital or infantile occlusion of anus with fistula Congenital infantile stricture of anus with fistula	751.2
Q42.3	Congenital absence, atresia and stenosis of anus without fistula Imperforated anus Anal atresia Congenital or infantile occlusion of anus without fistula Congenital infantile stricture of anus without fistula	751.2
Q42.8	Congenital absence, atresia and stenosis of other parts of large intestine Absence of appendix Atresia of appendix	751.2
Q42.9	Congenital absence, atresia and stenosis of large intestine, part unspecified	751.2
Q43	**Other congenital malformations of intestine**	
Q43.0	*(m) Meckel's diverticulum (displaced) (hypertrophic)* *(m) Persistent vitelline duct* *(m) Persistent omphalomesenteric duct*	751.0
Q43.1	Hirschsprung's disease Aganglionosis Congenital (aganglionic) megacolon Megacolon, congenital Long segment Hirschsprung's disease Short segment Hirschsprung's-Recto-Sigmoid Hirschsprung's disease, NOS	751.3
Q43.2	Other congenital functional disorders of colon	751.3

	Congenital dilation of colon Congenital megacolon, not aganglionic	
Q43.3	Congenital malformations of intestinal fixation Congenital omentum, anomalous adhesions [bands] Congenital peritoneal adhesions [bands] Incomplete rotation of cecum and colon Insufficient rotation of cecum and colon Jackson's membrane Intestinal malrotation Malrotation of colon Rotation failure of cecum and colon Universal mesentery Ladd's bands Malrotation of cecum Anomalies of mesentery Other specified malrotation Unspecified malrotation Malrotation of small intestine alone	751.4
Q43.4	Duplication of intestine Duplication of cyst ileum Duplication of anus Duplication of appendix Duplication of pylorus Duplication of cecum	751.5
Q43.5	Ectopic anus	751.5
Q43.6	Congenital fistula of rectum and anus **Excludes 1:** congenital fistula of anus with absence, atresia and stenosis (Q42.2), congenital fistula of rectum with absence, atresia and stenosis (Q42.0), congenital rectovaginal fistula (Q52.2), congenital urethrorectal fistula (Q64.73), pilonidal fistula or sinus (L05.-)	751.5
Q43.7	Persistent cloaca Cloaca, NOS	751.5
Q43.8	Other specified congenital malformations of intestine Congenital blind loop syndrome Congenital diverticulitis, colon Congenital diverticulum, intestine Dolichocolon Megaloduodenum Microcolon Transposition of appendix Transposition of colon Transposition of intestine Congenital anal fissure Congenital rectal fissure Enterogenous cyst Volvus Duodenal web *(m) Megaloappendix*	751.5
Q43.9	Congenital malformation of intestine, unspecified	751.5
Q44	**Congenital malformations of gallbladder, bile ducts and liver**	
Q44.0	Agenesis, aplasia and hypoplasia of gallbladder Congenital absence of gallbladder	751.69
Q44.1	Other congenital malformations of gallbladder Congenital malformation of gallbladder, NOS Intrahepatic gallbladder	751.69 751.60

	Duplication gallbladder Floating gallbladder	
Q44.2	Atresia of bile ducts Biliary atresia	751.61
Q44.3	Congenital stenosis and stricture of bile ducts Congenital hypoplasia of bile duct (common) or passage Congenital obstruction of bile duct (common) or passage	751.61
Q44.4	Choledochal cyst	751.69
Q44.5	Other congenital malformations of bile ducts Accessory hepatic duct Biliary duct duplication Congenital malformation of bile duct, NOS Cystic duct duplication Congenital dialated bile duct Agenesis or atresia of hepatic ducts Other anomaly of hepatic or bile ducts Anomalies of biliary tract, NEC	751.69 751.60
Q44.6	Cystic disease of liver Fibrocystic disease of liver	751.62
Q44.7	Other congenital malformation of liver Accessory liver Alagille's syndrome Congenital absence of liver Congenital malformation of liver, NOS Absence or agenesis of liver, partial Absence or agenesis of liver, total Floating liver Other anomaly of liver *(m) Congenital hepatomegaly*	751.60 751.69
Q45	**Other congenital malformations of digestive system** **Excludes 2:** congenital diaphragmatic hernia (Q79.0), congenital hiatus hernia (Q40.1)	
Q45.0	Agenesis, aplasia and hypoplasia of pancreas Congenital absence of pancreas	751.7
Q45.1	Annular pancreas	751.7
Q45.2	Congenital pancreatic cyst	751.7
Q45.3	Other congenital malformations of pancreas and pancreatic duct Accessory pancreas Congenital malformation of pancreas or pancreatic duct, NOS Ectopic pancreas Ectopic pancreatic tissue Pancreatic heterotopia **Excludes 1:** congenital diabetes mellitus (E10.-), cystic fibrosis (E84.0-E84.9), fibrocystic disease of pancreas (E84.-), neonatal diabetes mellitus (P70.2)	751.7
Q45.8	Other specified congenital malformations of digestive system Absence(complete) (partial) of alimentary tract, NOS Duplication of digestive system Malposition, congenital of digestive system Cyst of mesenteric remnant Duplication of alimentary tract Congenital malposition of digestive organs, NOS Ectopic digestive organs, NOS Other specified anomaly of digestive system	751.8
Q45.9	Congenital malformation of digestive system, unspecified Congenital anomaly of digestive system Congenital deformity of digestive system	751.9

Congenital Malformations of Genital Organs (Q50-Q56)

ICD-10 codes 56.0 to 56.4 require chromosome results.

ICD- 10	Description	ICD-9
Q50	**Congenital malformations of ovaries, fallopian tubes and broad ligaments**	
Q50.0	**Congenital absence of ovary** **[Only applicable to female patients.]** **Excludes 1:** Turner's Syndrome (Q96.-)	
Q50.01	Congenital absence or agenesis of ovary, unilateral	752.0
Q50.02	Congenital absence or agenesis of ovary, bilateral	752.0
Q50.1	Developmental ovarian cyst Cystic ovary Multiple ovarian cysts	752.0
Q50.2	Congenital torsion of ovary	752.0
Q50.3	**Other congenital malformations of ovary** **[Only applicable to female patients.]**	
Q50.31	Accessory ovary	752.0
Q50.32	Ovarian streak 46.XX with streak gonads	752.0
Q50.39	Other congenital malformation of ovary Congenital malformation of ovary, NOS Ectopic ovary	752.0
Q50.4	Embryonic cyst of fallopian tube Fimbrial cyst	752.11
Q50.5	Embryonic cyst of broad ligament Epoophoron cyst Parovarian cyst Cyst of hydatid of Morgagni	752.11
Q50.6	Other congenital malformations of fallopian tube and broad ligament Absence of fallopian tube and broad ligament Accessory fallopian tube and broad ligament Atresia of fallopian tube and broad ligament Congenital malformation of fallopian tube or board ligament, NOS	752.10 752.19
Q51	**Congenital malformations of uterus and cervix** **[Only applicable to female patients.]**	
Q51.0	Agenesis and aplasia of uterus Congenital absence of uterus	752.31
Q51.1	**Doubling of uterus with doubling of cervix and vagina** **[Only applicable to female patients.]**	
Q51.10	Doubling of uterus with doubling of cervix and vagina without obstruction Doubling of uterus with doubling of cervix and vagina, NOS	752.2
Q51.11	Doubling of uterus with doubling of cervix and vagina with obstruction	752.2
Q51.2	Other doubling of uterus Doubling of uterus, NOS Septate uterus, complete or partial Congenital duplication of uterus	752.2 752.35
Q51.3	Bicornate uterus Bicornate uterus, complete or partial	752.34
Q51.4	Unicornate uterus Unicornate uterus with or without a separate uterine horn Uterus with only one functioning horn Uterus unicornis	752.33
Q51.5	Agenesis and aplasia of cervix Congenital absence of cervix	752.43
Q51.6	*(m) Embryonic cyst of cervix*	752.41
Q51.7	Congenital fistula between uterus and digestive and urinary tracts Congenital uterointestinal fistula	752.49 752.39

Q51.8	**Other congenital malformations of uterus and cervix [Only applicable to female patients.]**	
Q51.81	**Other congenital malformations of uterus [Only applicable to female patients.]**	
Q51.810	Arcuate uterus Arcuatus uterus	752.36
Q51.811	Hypoplasia of uterus	752.32
Q51. 818	Other congenital malformations of uterus Müllerian anomaly of uterus, NEC	752.39
Q51.82	**Other congenital malformations of cervix [Only applicable to female patients.]**	
Q51.820	Cervical duplication Congenital duplication of cervix	752.44
Q51.821	Hypoplasia of cervix	752.49
Q51.828	Other congenital malformations of cervix Congenital imperforate cervix Congenital stenosis of cervical canal Congenital stenosis of uterine cervix Congenital stenosis or stricture of cervical canal	752.49
Q51.9	Congenital malformation of uterus and cervix, unspecified Congenital uterine anomaly	752.39 752.49
Q52	**Other congenital malformations of female genitalia [Only applicable to female patients.]**	
Q52.0	Congenital absence of vagina Vaginal agenesis, total or partial	752.45
Q52.1	**Doubling of vagina [Only applicable to female patients.]** **Excludes 1:** doubling of vagina with doubling of uterus and cervix (Q51.1-)	
Q52.10	Doubling of vagina, unspecified Septate vagina, NOS Congenital duplication of vagina Congenital vaginal septum	752.47
Q52.11	Transverse vaginal septum	752.46
Q52.12	Longitudinal vaginal septum Longitudinal vaginal septum with or without obstruction	752.47
Q52.2	Congenital rectovaginal fistula **Excludes 1:** cloaca (Q43.7)	752.49
Q52.3	(m) *Imperforate hymen*	752.42
Q52.4	Other congenital malformation of vagina Congenital malformation of vagina, NOS *(m) Canal of Nuck cyst, congenital* *(m) Embryonic vaginal cyst* *(m) Gartner's duct cyst* *(m) Cyst of vagina, vulva* *(m)Congenital stenosis or stricture of vagina*	752.41 752.49
Q52.5	Fusion of labia *(m) Labial adhesions*	752.49
Q52.6	Congenital malformation of clitoris Absence of clitoris Agenesis of clitoris *(m) Clitoromegaly* *(m) Enlarged clitoris* *(m) Other anomaly of clitoris* *(m) Prominent clitoris*	752.49
Q52.7	**Other and unspecified congenital malformations of vulva [Only applicable to female patients.]**	

Q52.70	Unspecified congenital malformations of vulva Congenital malformation of vulva, NOS Fusion of vulva	752.49
Q52.71	Congenital absence of vulva	752.49
Q52.79	Other congenital malformations of vulva *(m) Congenital cyst of vulva*	752.49
Q52.8	Other specified congenital malformation of female genitalia *(m) Vaginal or hymenal tags* *(m) Hypoplastic majora*	752.49
Q52.9	Congenital malformation of female genitalia, unspecified	752.9
Q53	**Undescended and ectopic testicle [Only applicable to male patients.]**	
Q53.0	**Ectopic testis [Only applicable to male patients.]**	
Q53.00	Ectopic testis, unspecified	752.51
Q53.01	Ectopic testis, unilateral Ectopic testis, left only Ectopic testis, right only	752.51
Q53.02	Ectopic testes, bilateral	752.51
Q53.1	**Undescended testicle, unilateral [Only applicable to male patients.]**	
Q53.10	Unspecified undescended testicle, unilateral Cryptorchidism, left only (birth weight greater than 2,500 grams) Left undescended testicle (birth weight greater than 2,500 grams) Cryptorchidism, right only (birth weight greater than 2,500 grams) Right undescended testicle (birth weight greater than 2,500 grams) Cryptorchidism, unilateral (birth weight greater than 2,500 grams) Undescended testicle, unilateral (birth weight greater than 2,500 grams)	752.51
Q53.11	Abdominal testis, unilateral	752.51
Q53.12	Ectopic perineal testis, unilateral	752.51
Q53.2	**Undescended testicle, bilateral [Only applicable to male patients.]**	
Q53.20	Undescended testicle, unspecified, bilateral Cryptorchidism, bilateral (birth weight greater than 2,500 grams) Undescended testicle, bilateral (birth weight greater than 2,500 grams)	752.51
Q53.21	Abdominal testis, bilateral	752.51
Q53.22	Ectopic perineal testis, bilateral	752.51
Q53.9	Undescended testicle, unspecified (birth weight greater than 2,500 grams) Cryptorchidism, NOS	752.51
Q54	**Hypospadias PLEASE SPECIFY DEGREE OR SITE OF HYPOSPADIAS [Only applicable to male patients.]** **Excludes 1:** epispadias (Q64.0)	
Q54.0	Hypospadias, balanic Hypospadias, coronal Hypospadias, glandular Hypospadias 1 degree, glandular, coronal	752.61
Q54.1	Hypospadias, penile Hypospadias 2 degree, penile, subcoronal	752.61
Q54.2	Hypospadias, penoscrotal Hypospadias 3 degree, scrotal, penile scrotal	752.61
Q54.3	Hypospadias 3 degree, perineal, scrotal, penile scrotal	752.61
Q54.4	Congenital chordee (report only with hypospadias) *(m) Chordee without hypospadias*	752.63
Q54.8	Other hypospadias Hypospadias with intersex state	752.61
Q54.9	Hypospadias, unspecified Paraspadias	752.61
Q55	**Other congenital malformations of male genital organs [Only applicable to male patients.]**	

	Excludes 1: congenital hydrocele (P83.5), hypospadias (Q54.-)	
Q55.0	Absence and aplasia of testis Monorchism Congenital absence of testicle	752.89
Q55.1	Hypoplasia of testis and scrotum Fusion of testes	752.89
Q55.2	**Other and unspecified congenital malformations of testis and scrotum [Only applicable to male patients.]**	
Q55.20	Unspecified congenital malformations of testis and scrotum Congenital malformation of testis or scrotum, NOS	752.9
Q55.21	Polyorchidism	752.89
Q55.22	*(m) Retractile testis*	752.52
Q55.23	Scrotal transposition Penoscrotal transposition	752.81
Q55.29	Other congenital malformations of testis and scrotum	752.89
Q55.3	Atresia of vas deferens Code first any associated cystic fibrosis (E84)	752.89
Q55.4	Other congenital malformations of vas deferens, epididymis, seminal vesicles and prostate Absence or aplasia of prostate Absence or aplasia of spermatic cord Congenital malformation of vas deferens, epididymis, seminal vesicles or prostate, NOS Atresia of ejaculatory duct Appendix testis cyst Cysts of embryonic remnants Hydatid of morgagni cyst Wolffian duct cyst	752.89
Q55.5	Congenital absence and aplasia of penis	752.69
Q55.6	Other congenital malformations of penis	752.69
Q55.61	Curvature of penis (lateral)	752.69
Q55.62	Hypoplasia of penis Micropenis	752.64
Q55.63	Congenital torsion of penis **Excludes 1:** acquired torsion of penis (N48.82)	752.69
Q55.64	Concealed penis Webbed penis *(m) Hidden penis* *(m) Buried penis* **Excludes 1:** acquired buried penis (N48.83)	752.65
Q55.69	Other congenital malformation of penis Congenital malformation of penis, NOS Absent or hooded foreskin Incomplete foreskin	752.69
Q55.7	Congenital vasocutaneous fistula	752.89
Q55.8	Other specified congenital malformations of male genital organs	752.89
Q55.9	Congenital malformations of male genital organ, unspecified Congenital anomaly of male genital organ Congenital deformity of male genital organ	752.9
Q56	**Indeterminate sex and pseudohermaphroditism** **PLEASE REPORT CHROMOSOME TEST RESULTS** **Excludes 1:** 46,XX true hermaphrodite (Q99.1), androgen insensitivity syndrome (E34.5-), chimera 46,XXX/46,XY true hermaphrodite (Q99.0), female pseudohermaphroditism with adrenocortical disorder (E25.-), pseudohermaphroditism with specified chromosomal anomaly (Q96-Q99),pure gonadal dysgenesis (Q99.1)	

Q56.0	Hermaphroditism, NEC Ovotestis True hermaphroditism Gynandrism	752.7
Q56.1	Male pseudohermaphroditism, NEC 46, XY with streak gonads Male pseudohermaphroditism, NOS	752.7
Q56.2	Female pseudohermaphroditism, NEC Female pseudohermaphroditism, NOS	752.7
Q56.3	Pseudohermaphroditism, unspecified	752.7
Q56.4	Indeterminate sex, unspecified Ambiguous genitalia Disorder of sexual differentiation	752.7

Congenital Malformations of the Urinary System (Q60-Q64)

ICD-10	Description	ICD-9
Q60	**Renal agenesis and other reduction defects of kidney** **Includes:** congenital absence of kidney, congenital atrophy of kidney, infantile atrophy of kidney	
Q60.0	Renal agenesis, unilateral Single kidney Solitary kidney	753.0
Q60.1	Renal agenesis, bilateral	753.0
Q60.2	Renal agenesis, unspecified	753.0
Q60.3	Renal hypoplasia, unilateral	753.0
Q60.4	Renal hypoplasia, bilateral	753.0
Q60.5	Renal hypoplasia, unspecified	753.0
Q60.6	Potter's syndrome Oligohydramnios sequence	753.0
Q61	**Cystic kidney disease** **Excludes 1:** acquired cyst of kidney (N28.1), Potter's syndrome (Q60.6)	
Q61.0	Congenital renal cyst	
Q61.00	Congenital renal cyst, unspecified Cyst of kidney, NOS (congenital)	753.10
Q61.01	Congenital single renal cyst	753.11
Q61.02	Congenital multiple renal cysts	753.19
Q61.1	Polycystic kidney, infantile type Polycystic kidney, autosomal recessive	
Q61.11	Cystic dilation of collecting ducts	753.14
Q61.19	Other polycystic kidney, infantile type Autosomal recessive infantile polycystic kidney disease Polycystic kidney, autosomal recessive	753.14
Q61.2	Polycystic kidney, adult type Polycystic kidney, autosomal dominant	753.13
Q61.3	Polycystic kidney, unspecified	753.12
Q61.4	Renal dysplasia Multicystic dysplastic kidney Multicystic kidney (development) Multicystic kidney disease Multicystic renal dysplasia Congenital renal dysplasia Dysplastic kidney, bilateral Renal dysplasia, bilateral Unilateral dysplasia of kidney Renal dysplasia, NOS; dysplastic kidney, NOS **Excludes 1:** polycystic kidney disease (Q61.11-Q61.3)	753.15 753.19
Q61.5	Medullary cystic kidney Medullary cystic kidney, juvenile type Medullary cystic kidney, adult type Medullary sponge kidney Nephronopthisis Sponge kidney, NOS	753.16 753.17
Q61.8	Other cystic kidney diseases Fibrocystic kidney Fibrocystic renal degeneration of disease	753.19
Q61.9	Cystic kidney disease, unspecified Meckel-Gruber syndrome Cystic kidney disease	753.10
Q62	**Congenital obstructive defects of renal pelvis and congenital malformations of ureter**	

Q62.0	Congenital hydronephrosis Congenital hydronephrosis, NOS Congenital hydronephrosis, left only Congenital hydronephrosis, right only Congenital hydronephrosis, unilateral Congenital hydronephrosis, bilateral Pyelectasis Pelvicaliectasis/Pelviectasis	753.29
Q62.1	Congenital occlusion of ureter Atresia and stenosis of ureter	
Q62.10	Congenital occlusion of ureter, unspecified Stenosis of ureter Atresia, stricture, or stenosis of ureter, NOS Impervious ureter, NOS Ureteral valves, NOS Atresia, structure, or stenosis of ureter, left Congenital occlusion of ureter, left Impervious ureter, left Ureteral valves, left Atresia, structure, or stenosis of ureter, right Congenital occlusion of ureter, right Impervious ureter, right Ureteral valves, right Atresia, structure, or stenosis of ureter, unilateral Congenital occlusion of ureter, unilateral Impervious ureter, unilateral Ureteral valves, unilateral Atresia, structure, or stenosis of ureter, bilateral Congenital occlusion of ureter, bilateral Impervious ureter, bilateral Ureteral valves, bilateral	753.29
Q62.11	Congenital occlusion of ureteropelvic junction Stricture of pelviureteric junction Ureteropelvic junction obstruction/stenosis, left Ureteropelvic junction obstruction/stenosis, right Ureteropelvic junction obstruction/stenosis, unilateral Ureteropelvic junction obstruction/stenosis, bilateral	753.21
Q62.12	Congenital occlusion of ureterovesical orifice Ureterovesical junction obstruction/stenosis, NOS Ureterovesical junction obstruction/stenosis, left Ureterovesical junction obstruction/stenosis, right Ureterovesical junction obstruction/stenosis, unilateral Ureterovesical junction obstruction/stenosis, bilateral	753.22
Q62.2	Congenital megaureter Congenital dilation of ureter Congenital hydroureter, NOS Megaloureter, NOS Congenital hydroureter, left Megaloureter, left Congenital hydroureter, right Megaloureter, right Congenital hydroureter, unilateral Megaloureter, unilateral Congenital hydroureter, bilateral Megaloureter, bilateral	753.22

Q62.3	Other obstructive defects of renal pelvis and ureter	753.22
Q62.31	Congenital ureterocele, orthotopic Ureterocele is reportable; Hydrocele is NOT reportable	753.23
Q62.32	Cecoureterocele Ectopic ureterocele	753.23
Q62.39	Other obstructive defects of renal pelvis ureter Ureteropelvic junction obstruction, NOS	753.20
Q62.4	Agenesis of ureter Congenital absence ureter	753.4
Q62.5	Duplication of ureter Accessory ureter Double ureter, duplex collecting system Duplication of collecting system	753.4
Q62.6	Malposition of ureter	
Q62.60	Malposition of ureter, unspecified	753.4
Q62.61	Deviation of ureter	753.4
Q62.62	Displacement of ureter	753.4
Q62.63	Anomalous implantation of ureter Ectopia of ureter Ectopic ureter Displaced ureteric orifice	753.4
Q62.69	Other malposition of ureter	753.4
Q62.7	Congenital vesico-uretero-renal reflux Ureteral reflux Variations of vesicoureteral reflux	753.4
Q62.8	Other congenital malformations of ureter Anomaly of ureter, NOS Unspecified obstructive defects of renal pelvis and ureter Other specified anomalies of ureter Unspecified anomaly of ureter Hypoplastic ureter, NOS Hypoplastic ureter, left Hypoplastic ureter, right Hypoplastic ureter, bilateral	753.4
Q63	**Other congenital malformations of kidney** **Excludes 1:** congenital nephrotic syndrome (N04.-)	
Q63.0	Accessory kidney Double kidney and pelvis, pyelon duplex Trifid kidney (pelvis) Triple kidney and pelvis, pyelon triplex	753.3
Q63.1	Lobulated, fused and horseshoe kidney	753.3
Q63.2	Ectopic kidney/Pelvic Kidney Congenital displaced kidney Malrotation of kidney	753.3
Q63.3	Hyperplastic and giant kidney Compensatory hypertrophy of kidney	753.3
Q63.8	Other specified congenital malformations of kidney Congenital renal caluli Discoid kidney Congenital calculus of kidney	753.3
Q63.9	Congenital malformation of kidney, unspecified	753.3
Q64	**Other congenital malformations of urinary system**	
Q64.0	Epispadias **Excludes 1:** hypospadias (Q54.-) Anaspadias Ventral meatus	752.62

	[Q64.0 is only applicable to male patients.]	
Q64.10	Exstrophy of urinary bladder, unspecified Ectopia vesicae	753.5
Q64.11	Supravesical fissure of urinary bladder	753.8
Q64.12	Cloacal exstrophy of urinary bladder Exstrophy of cloaca sequence	753.5
Q64.19	Other exstrophy of urinary bladder Estroversion of bladder	753.5
Q64.2	Congenital posterior urethral valves	753.6
Q64.3	Other atresia and stenosis of urethra and bladder neck	
Q64.31	Congenital bladder neck obstruction Congenital obstruction of vesicourethral orifice	753.6
Q64.32	Congenital stricture of urethra	753.6
Q64.33	Congenital stricture of urinary meatus	753.6
Q64.39	Other atresia and stenosis of urethra and bladder neck Atresia and stenosis of urethra and bladder neck, NOS	753.6
Q64.4	Malformation of urachus Cyst of urachus Patent urachus Prolapse of urachus Fistula of urachus	753.7
Q64.5	Congenital absence of bladder and urethra	753.8
Q64.6	Congenital diverticulum of bladder	753.8
Q64.7	Other and unspecified congenital malformations of bladder and urethra **Excludes 1:** congenital prolapse of bladder (mucosa) (Q79.4)	
Q64.70	Unspecified congenital malformations of bladder and urethra Malformation of bladder or urethra, NOS	753.9
Q64.71	Congenital prolapse of urethra	753.8
Q64.72	Congenital prolapse of urinary meatus	753.8
Q64.73	Congenital urethrorectal fistula	753.8
Q64.74	Double urethra	753.8
Q64.75	Double urinary meatus	753.8
Q64.79	Other congenital malformations of bladder and urethra	753.8
Q64.8	Other specified congenital malformations of urinary system	753.8
Q64.9	Congenital malformation of urinary system, unspecified Congenital anomaly NOS of urinary system Congenital deformity NOS of urinary system	753.9

Congenital Malformations of the Musculoskeletal System (Q65-Q79)

ICD 10	Description	ICD 9
Q65	**Congenital deformities of hip Excludes 1:** clicking hip (R29.4)	
Q65.0	Congenital dislocation of hip, unilateral	754.30
Q65.00	Congenital dislocation of unspecified hip, unilateral	
Q65.01	Congenital dislocation of right hip, unilateral	754.30
Q65.02	Congenital dislocation of left hip, unilateral	754.30
Q65.1	Congenital dislocation of hip, bilateral	754.31
Q65.2	Congenital dislocation of hip, unspecified	754.30
Q65.3	Congenital partial dislocation of hip, unilateral	
Q65.30	Congenital partial dislocation of unspecified hip, unilateral Congenital subluxation of unspecified hip, unilateral	754.32
Q65.31	Congenital partial dislocation of right hip, unilateral Congenital subluxation of right hip	754.32
Q65.32	Congenital partial dislocation of left hip, unilateral Congenital subluxation of left hip	754.32
Q65.4	Congenital partial dislocation of hip, bilateral Congenital subluxation of hip, bilateral	754.33
Q65.5	Congenital partial dislocation of hip, unspecified Congenital subluxation of hip	754.32
Q65.6	Congenital unstable hip Congenital dislocatable hip Predislocation status of hip at birth, unilateral, left Preluxation of hip, congenital, left Predislocation status of hip at birth, unilateral, right Preluxation of hip, congenital, right Unstable hip, unilateral, right Predislocation status of hip at birth, unilateral Preluxation of hip, congenital, unilateral Unstable hip, unilateral Unstable hip, unilateral, left	754.32
Q65.8	Other congenital deformities of hip	755.61
Q65.81	Congenital coxa valga	755.61
Q65.82	Congenital coxa vara	755.62
Q65.89	Other specified congenital deformities of hip Anteversion of femoral neck Congenital acetabular dysplasia Hip dysplasia, NOS Unilateral hip dysplasia Bilateral hip dysplasia Congenital flexion deformity, hip or thigh, left Congenital flexion deformity, hip or thigh, right	755.63
Q65.9	Congenital deformity of hip, unspecified	755.63
Q66	**Congenital deformities of feet** **Excludes 1:** reduction defects of feet (Q72.-), valgus deformities (acquired) (M21.0-), varus deformities (acquired) (M21.1-)	
Q66.0	Congenital talipes equinovarus Talipes equinovarus, NOS Talipes equinovarus, left Talipes equinovarus, right Talipes equinovarus, unilateral Talipes equinovarus, bilateral	754.51
Q66.1	Congenital talipes calcancovarus	754.59
Q66.2	Congenital metatarsus (primus) varus	754.52

	Metatarsus varus or metatarsus adductus	
Q66.3	Other congenital varus deformities of feet Hallux varus, congenital Talipes varus	754.59
Q66.4	Congenital talipes calcancovalgus Talipes calcaneovalgus, left Talipes calcaneovalgus, right Talipes calcaneovalgus, unilateral Talipes calcaneovalgus, bilateral	754.62
Q66.5	Congenital pes planus Congenital flat foot Congenital rigid flat foot Congenital spastic (everted) flat foot *(m) Congenital rocker bottom foot* **Excludes 1:** pes planus, acquired (M21.4)	754.61
Q66.50	Congenital pes planus, unspecified foot	754.61
Q66.51	Congenital pes planus, right foot	754.61
Q66.52	Congenital pes planus, left foot	754.61
Q66.6	Other congenital valgus deformities of feet Congenital metatarsus valgus Talipes valgus Talipes equinovalgus Talipes planovalgus Clawfoot or lobster claw foot	754.69
Q66.7	Congenital pes cavus Cavas foot Talipes cavas	754.71
Q66.8	Other congenital deformities of feet	754.61
Q66.80	Congenital vertical talus deformity, unspecified foot	754.61
Q66.81	Congenital vertical talus deformity, right foot	754.61
Q66.82	Congenital vertical talus deformity, left foot	754.61

Q66.89	Other specified congenital deformities of feet Congenital asymmetric talipes Congenital talipes, NOS Congenital tarsal coalition Congenital clubfoot, NOS Clubfoot, NOS Dorsiflexion of foot (fixed only) Other specified deformities of toes Anomalies of foot Calcaneonavicular bar Coalition of calcaneus Plantar furrow Prominent heel Talonavicular synostosis Tarsal coalitions Astragalo-scaphoid synostosis Talipes calcaneus Talipes equinus Congenital hallux valgus Congenital hallux varus *(m) Hammer toe, congenital* *(m) Gap between toes (1-2)* *(m) Other anomalies of toes* *(m) Overlapping toes* *(m) Recessed toes (4,5)* *(m) Wide spaced first and second toes* *(m) Long toes* *(m) Short or broad great toe*	754.79 755.67
Q66.9	Congenital deformity of feet, unspecified	754.79
Q67	**Congenital musculoskeletal deformities of head, face, spine and chest** **Excludes 1:** congenital malformation syndromes classified to Q87.-, Potter's syndrome (Q60.6)	
Q67.0	*(m) Congenital facial asymmetry* *(m) Asymmetry of face* *(m) Facial asymmetry* *(m) Head asymmetry*	754.0
Q67.1	*(m) Congenital compression facies*	754.0
Q67.2	*(m) Dolichocephaly*	754.0
Q67.3	*(m) Plagiocephaly* *(m) Flat occiput*	754.0
Q67.4	Other congenital deformities of skull, face and jaw Congenital hemifacial atrophy or hypertrophy *(m) Congenital depressions in skull* *(m) Deviation of nasal septum, congenital* *(m) Squashed or bent nose, congenital* *(m) Metopic suture open to bregma* *(m) Large or small fontanelles* *(m) Third fontanel* **Excludes 1:** dentofacial anomalies [including malocclusion] (M26.-), syphilitic saddle nose (A50.5)	748.1 754.0
Q67.5	Congenital deformity of spine Congenital postural scoliosis Congenital scoliosis, NOS **Excludes 1:** infantile idiopathic scoliosis (M41.0), scoliosis due to congenital bony malformation (Q76.3)	754.2
Q67.6	Pectus excavatum	754.81

	Congenital funnel chest	
Q67.7	Pectus carinatum Congenital pigeon chest	754.82
Q67.8	Other congenital deformities of chest Congenital deformity of chest wall, NOS Congenital anomaly of thoracic cage Shield chest *(m) Bell shaped chest*	754.89
Q68	**Other congenital musculoskeletal deformities** **Excludes 1:** reduction defects of limb(s) (Q71-Q73) **Excludes 2:** congenital myotonic chondrodystrophy (G71.13)	
Q68.0	Congenital deformity of sternocleidomastoid muscle Congenital contracture of sternocleidomastoid (muscle) Congenital anomaly of sternocleidomastoid muscle Congenital (sternomastoid) torticollis Sternomastoid tumor (congenital) Absent or hypoplastic sternocleidomastoid muscle Sternomastoid tumor Congenital wryneck	754.1
Q68.1	Congenital deformity of finger(s) and hand Congenital club finger Spade-like hand (congenital) Anthrogryposis Flexion contractures of individual joints of the hand	754.89
Q68.2	Congenital deformity of knee Congenital dislocation of knee Congenital genu recurvatum	755.64 754.41 754.40
Q68.3	Congenital bowing of femur **Excludes 1:** anteversion of femur (neck) (Q65.89)	754.42
Q68.4	Congenital bowing of tibia and fibula	754.43
Q68.5	Congenital bowing of long bones of leg, unspecified	754.44
Q68.6	Discoid meniscus Discoid meniscus of knee	717.5
Q68.8	Other specified congenital musculoskeletal deformities Congenital deformity of clavicle Congenital deformity of elbow Congenital deformity of forearm Congenital deformity of scapula Congenital deformity of wrist Congenital dislocation of elbow Congenital dislocation of shoulder Congenital dislocation of wrist Congenital dislocation of radial head Congenital elevation of scapula Generalized flexion contractures of lower limb joints	756.9 755.51 755.52
Q69	**Polydactyly**	
Q69.0	Accessory finger(s) Accessory digits hand, NOS Polydactyly of fingers	755.01
Q69.1	Accessory thumb(s)	755.01
Q69.2	Accessory toe(s), do not report if skin tag Polydactyly of toes Accessory hallux Accessory toes (postaxial) Accessory big toe (preaxial) Accessory digits foot, NOS	755.02

Q69.9	Polydactyly, unspecified Supernumerary digit(s), NOS Postaxial polydactyly, NOS Accessory digits, NOS (hand/foot not specified)	755.00
Q70	**Syndactyly**	
Q70.0	**Fused fingers** **Complex syndactyly of fingers and/or thumb with synostosis**	
Q70.00	Fused fingers and/or thumb, unspecified hand Syndactyly of fingers with fusion of bones	755.12
Q70.01	Fused fingers and/or thumb, right hand Syndactyly of fingers of right hand with fusion of bones	755.12
Q70.02	Fused fingers and/or thumb, left hand Syndactyly of fingers of left hand with fusion of bones	755.12
Q70.03	Fused fingers and/or thumb, bilateral Syndactyly of fingers, bilateral, with fusion of bones	755.12
Q70.1	***(m) Webbed fingers*** ***(m) Simple syndactyly of fingers without synostosis***	
Q70.10	*(m) Webbed fingers, unspecified hand*	755.11
Q70.11	*(m) Webbed fingers, right hand*	755.11
Q70.12	*(m) Webbed fingers, left hand*	755.11
Q70.13	*(m) Webbed fingers, bilateral*	755.11
Q70.2	**Fused toes** **Complex syndactyly of toes with synostosis**	
Q70.20	Fused toes, unspecified foot Syndactyly of toes with fusion of bones	755.14
Q70.21	Fused toes, right foot Syndactyly of toes of right foot, with fusion of bones	755.14
Q70.22	Fused toes, left foot Syndactyly of toes of left foot, with fusion of bones	755.14
Q70.23	Fused toes, bilateral Syndactyly of toes of both feet, with fusion of bones	755.14
Q70.3	***(m) Webbed toes*** ***(m) Simple syndactyly of toes without synostosis***	
Q70.30	*(m) Webbed toes, unspecified foot*	755.13
Q70.31	*(m) Webbed toes, right foot*	755.13
Q70.32	*(m) Webbed toes, left foot*	755.13
Q70.33	*(m) Webbed toes, bilateral*	755.13
Q70.4	Polysyndactyly, unspecified **Excludes 1:** specified syndactyly of hand and feet - code to specified conditions (Q70.0- -Q70.3-)	755.00 755.10
Q70.9	Syndactyly, unspecified Symphalangy, NOS Syndactyly of multiple and unspecified sites	755.10
Q71	**Reduction defects of upper limb**	
Q71.0	**Congenital complete absence of upper limb** **Transverse deficiency of upper limb**	
Q71.00	Congenital complete absence of unspecified upper limb Congenital amputation of upper limb, NOS Transverse hemimelia of upper limb	755.21
Q71.01	Congenital complete absence of right upper limb	755.21
Q71.02	Congenital complete absence of left upper limb	755.21
Q71.03	Congenital complete absence of upper limb, bilateral	755.21
Q71.1	Congenital absence of upper arm and forearm with hand present	
Q71.10	Congenital absence of unspecified upper arm and forearm with hand present Phocomelia, complete, of upper limb	755.23

	Proximal phocomelia of upper limb Distal phocomelia of upper limb (forearm missing. upper arm/hand present) Total absence of forearm only (radius and ulna)	
Q71.11	Congenital absence of right upper arm and forearm with hand present	755.23
Q71.12	Congenital absence of left upper arm and forearm with hand present	755.23
Q71.13	Congenital absence of upper arm and forearm with hand present, bilateral Complete phocomelia of bilateral upper limbs	755.23
Q71.2	**Congenital absence of both forearm and hand** **Transverse deficiency, radioulnar, complete or partial**	
Q71.20	Congenital absence of both forearm and hand, unspecified upper limb	755.25
Q71.21	Congenital absence of both forearm and hand, right upper limb	755.25
Q71.22	Congenital absence of both forearm and hand, left upper limb	755.25
Q71.23	Congenital absence of both forearm and hand, bilateral	755.25
Q71.3	**Congenital absence of hand and finger**	
Q71.30	Congenital absence of unspecified hand and finger Absence of finger(s), congenital (includes individual missing fingers/phalanges) Absence of preaxial fingers Absence of thumb Absence of fingers (postaxial) Hypoplasia of fingers Hypoplasia of thumb	755.29
Q71.31	Congenital absence of right hand and finger	755.29
Q71.32	Congenital absence of left hand and finger	755.29
Q71.33	Congenital absence of hand and finger, bilateral	755.29
Q71.4	**Longitudinal reduction defect of radius (congenital)** **Radial clubhand**	
Q71.40	Longitudinal reduction defect of unspecified radius Preaxial (longitudinal) reduction deficiency of upper limb, radial ray defect Absence of radius Congenital absence of radius with or without absence of some distal elements Longitudinal deficiency, radial, complete or partial	755.26
Q71.41	Longitudinal reduction defect of right radius	755.26
Q71.42	Longitudinal reduction defect of left radius	755.26
Q71.43	Longitudinal reduction of radius, bilateral	755.26
Q71.5	**Longitudinal reduction defect of ulna**	
Q71.50	Longitudinal reduction defect of unspecified ulna Absence of ulna Congenital absence ulna, with or without absence of some distal elements Longitudinal deficiency ulna, complete or partial Postaxial (longitudinal) reduction deficiency of upper limb, ulna ray defect	755.27
Q71.51	Longitudinal reduction defect of right ulna	755.27
Q71.52	Longitudinal reduction defect of left ulna	755.27
Q71.53	Longitudinal reduction defect of ulna, bilateral	755.27
Q71.6	**Lobster-claw hand** **Cleft hand, congenital**	
Q71.60	Lobster-claw hand, unspecified hand Cleft hand	755.58
Q71.61	Lobster-claw right hand Cleft hand, right hand	755.58
Q71.62	Lobster-claw left hand Cleft hand, left hand	755.58
Q71.63	Lobster-claw hand, bilateral Cleft hand, bilateral	755.58
Q71.8	**Other reduction defect of upper limb**	

Q71.81	**Congenital shorting of upper limb**	
Q71.811	Congenital shortening of right upper limb	755.20
Q71.812	Congenital shortening of left upper limb	755.20
Q71.813	Congenital shortening of upper limb, bilateral	755.20
Q71.819	Congenital shortening of unspecified upper limb	755.20
Q71.89	**Other reduction defects of upper limb**	
Q71.891	Other reduction defects of right upper limb	755.20
Q71.892	Other reduction defects of left upper limb	755.20
Q71.893	Other reduction defects of upper limb, bilateral	755.20
Q71.899	Other reduction defects of unspecified upper limb Ectromelia NOS of upper limb Hemimelia of upper limb, NOS Longitudinal reduction deficiency of upper limb, unspecified ray defect	755.20
Q71.9	**Unspecified reduction defect of upper limb**	
Q71.90	Unspecified reduction defect of unspecified upper limb	755.20
Q71.91	Unspecified reduction defect of right upper limb	755.20
Q71.92	Unspecified reduction defect of left upper limb	755.20
Q71.93	Unspecified reduction defect of upper limb, bilateral	755.20
Q72	**Reduction defects of lower limb**	
Q72.0	Congenital complete absence of lower limb Amelia of lower limb Transverse hemimelia of lower leg Congenital amputation of lower limbs, NOS	
Q72.00	Congenital complete absence of unspecified lower limb	755.31
Q72.01	Congenital complete absence of right lower limb	755.31
Q72.02	Congenital complete absence of left lower limb	755.31
Q72.03	Congenital complete absence of lower limb, bilateral	755.31
Q72.1	Congenital absence of thigh and lower leg with foot present	
Q72.10	Congenital absence of unspecified thigh and lower leg with foot present Phocomelia, complete, of lower limb (upper and lower leg missing, foot present) Absence of lower leg only (femur and foot present) Proximal phocomelia of lower limb (femur absent, lower leg, foot present) Distal phocomelia of lower limb (lower leg absent, femur/foot present)	755.33
Q72.11	Congenital absence of right thigh and lower leg with foot present	755.33
Q72.12	Congenital absence of left thigh and lower leg with foot present	755.33
Q72.13	Congenital absence of thigh and lower leg with foot present, bilateral	755.33
Q72.2	**Congenital absence of both lower leg and foot**	
Q72.20	Congenital absence of both lower leg and foot, unspecified lower limb Transverse deficiency lower limb - knee level	755.35
Q72.21	Congenital absence of both lower leg and foot, right lower limb	755.35
Q72.22	Congenital absence of both lower leg and foot, left lower limb	755.35
Q72.23	Congenital absence of both lower leg and foot, bilateral	755.35
Q72.3	**Congenital absence of foot and toe(s)** **Aphalangia of lower limb, terminal, complete or partial**	
Q72.30	Congenital absence of unspecified foot and toe(s)	755.38
Q72.31	Congenital absence of right foot and toe(s)	755.38
Q72.32	Congenital absence of left foot and toe(s)	755.38 755.39
Q72.33	Congenital absence of foot and toe(s), bilateral	755.38 755.39
Q72.4	**Longitudinal reduction defect of femur** **Proximal femoral focal deficiency** **Congenital absence of femur with absence of distal elements** **Congenital absence of femur, without presence of distal elements**	

Q72.40	Longitudinal reduction defect of unspecified femur	755.34
Q72.41	Longitudinal reduction defect of right femur	755.34
Q72.42	Longitudinal reduction defect of left femur	755.34
Q72.43	Longitudinal reduction defect of femur, bilateral	755.34
Q72.5	**Longitudinal reduction defect of tibia** **Absence of tibia, preaxial longitudinal** **Agenesis of tibia, tibial ray defect** **Congenital absence tibia with absence of some distal elements, tibial ray defect** **Longitudinal deficiency, tibia, complete or partial**	
Q72.50	Longitudinal reduction defect of unspecified tibia	755.36
Q72.51	Longitudinal reduction defect of right tibia	755.36
Q72.52	Longitudinal reduction defect of left tibia	755.36
Q72.53	Longitudinal reduction defect of tibia, bilateral	755.36
Q72.6	**Longitudinal reduction defect of fibula** **Congenital absence of fibula** **Absence of fibula, postaxial longitudinal, fibular ray defect** **Agenesis of fibula** **Congenital absence of fibula, with absence of some distal elements** **Longitudinal deficiency, fibula, complete or partial, fibular ray defect**	
Q72.60	Longitudinal reduction defect of unspecified fibula	755.37
Q72.61	Longitudinal reduction defect of right fibula	755.37
Q72.62	Longitudinal reduction defect of left fibula	755.37
Q72.63	Longitudinal reduction defect of fibula, bilateral	755.37
Q72.7	**Split foot** **Cleft foot** **Clawfoot or lobster claw foot**	
Q72.70	Split foot, unspecified lower limb	755.67
Q72.71	Split foot, right lower limb	755.67
Q72.72	Split foot, left lower limb	755.67
Q72.73	Split foot, bilateral	755.67
Q72.8	**Other reduction defects of lower limb**	
Q72.81	Congenital shorting of lower limb	755.30
Q72.811	Congenital shorting of right lower limb	755.30
Q72.812	Congenital shorting of left lower limb	755.30
Q72.813	Congenital shorting of lower limb, bilateral	755.30
Q72.819	Congenital shorting od unspecified lower limb	755.30
Q72.89	Other reduction defects of lower limb	755.30
Q72.891	Other reductions defects of right lower limb	755.30
Q72.892	Other reductions defects of left lower limb	755.30
Q72.893	Other reduction defects of lower limb, bilateral	755.30
Q72.899	Other reduction defects of unspecified lower limb	755.30
Q72.9	**Unspecified reduction defect of lower limb**	
Q72.90	Unspecified reduction defect of unspecified lower limb	755.32
Q72.91	Unspecified reduction defect of right lower limb	755.32
Q72.92	Unspecified reduction defect of left lower limb	755.32
Q72.93	Unspecified reduction defect of lower limb, bilateral	755.32
Q73	**Reduction defects of unspecified limb (complete or partial)**	
Q73.0	Congenial absence of unspecified limb(s) Amelia, NOS	755.4
Q73.1	Phocomelia, unspecified limb(s) Phocomelia, NOS	755.4
Q73.8	Other reduction defects of unspecified limb(s) Longitudinal reduction deformity of unspecified limb(s) Ecromelia of limb, NOS	755.4

	Hemimelia of limb, NOS Reduction defect of limb, NOS Amputation of unspecified limb Absent digits, NOS Unspecified reduction defect of unspecified limb	
Q74	**Other congenital malformations of limb(s)** **Excludes 1:** 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 Madelung's deformity Radioulnar synostosis Sprengel's deformity Triphalangeal thumb Bifid thumb Anomalies of hand Other specified deformity of hands Anomalies of wrist Radioulnar dysostosis Anomalies of elbow and upper arm Cubitus valgus, congenital Cubitus varus, congenital Anomalies of shoulder Hyperextensibilty of upper limb Other specified anomaly of upper limb Hypoplasia of arms Hypoplasia of upper limb *(m) Macrodactylia (fingers)* *(m) Anomalies of finger* *(m) Brachydactyly* *(m) Camptodactyly* *(m) Clinodactyly* *(m) Incurving fingers* *(m) Long fingers* *(m) Long fingers and toes* *(m) Overlapping fingers* *(m) Tapered fingers*	755.53 755.54 755.56 755.57 755.59
Q74.1	Congenital malformation of knee (and joint) Congenital absence of patella Congenital dislocation of patella Congenital genu valgum (knock-knee) Congenital genu varum (bowleg) Rudimentary patella Bifid patella **Excludes 1**: congenital dislocation of knee (Q68.2), congenital genu recurvatum (Q68.2), nail patella syndrome (Q87.2)	755.64
Q74.2	Other congenital malformations of lower limb(s), including pelvic girdle Congenital fusion of sacroiliac joint Congenital malformation of ankle joint Congenital malformation of sacroiliac joint Deformity of leg, NOS Hypoplasia of feet Dorsiflexion of foot Anomalies of ankle Anomalies of upper leg	755.69

	Anomalies of pelvis Congenital deformity of sacroiliac (joint) Other specified anomaly of lower limb Femoral hypoplasia syndrome *(m) Anomalies of lower leg* *(m) Congenital angulation of tibia* *(m) Tibial torsion* *(m) Macrodactylia of toes* **Excludes 1:** anteversion of femur (neck) (Q65.89)	
Q74.3	Arthrogryposis multiplex congenita	754.89
Q74.8	Other specified congenital malformations of limb(s) Larsen's syndrome	755.8
Q74.9	Unspecified congenital malformations of limb(s) Congenital anomaly of limb(s), NOS Anomalies of forearm, NOS Other anomaly of whole arm Unspecified anomalies of upper limb	755.9
Q75	**Other congenital malformations of skull and face bones** **Excludes 1:** congenital malformation of face NOS (Q18.-), congenital malformation syndromes classified to Q87.-, dentofacial anomalies [including malocclusion] (M26.-), 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.-), and hydrocephalus (Q03.-)microcephaly (Q02)	
Q75.0	Craniosynostosis Complex craniosynostosis Simple craniosynostosis Craniosynostosis, NOS Acrocephaly Imperfect fusion of skull Oxycephaly Trigonocephaly Closed skull sutures, NOS Premature closure of cranial sutures Sagittal craniosynostosis Scaphocephaly (if due to craniosynostosis) Metopic craniosynostosis Coronal craniosynostosis Lambdoidal Basilar craniosynostosis Other types of craniosynostosis Tower skull (if due to crainosynostosis) *(m) Trigonocephaly, other head deformations without synostosis*	756.0
Q75.1	Craniofacial dysostosis Crouzon's disease	756.0
Q75.2	Hypertelorism Wide set eyes	756.0
Q75.3	Macrocephaly	756.0
Q75.4	Mandibulofacial dysostosis Franceschetti syndrome Treacher Collins syndrome	756.0
Q75.5	Oculomandibular Dysostosis	756.0
Q75.8	Other specified congenital malformations of skull and face bones Absence of skull bone, congenital Congenital deformity of forehead Platybasia	756.0

	Prominent occiput Hallermann-Streiff syndrome Oculomandibulofacial syndrome Other craniofacial syndromes Goldenhar syndrome Oculoauriculovertebral (OAV) dysplasia Hemifacial microsomia Cleft mandible malar hypoplasia Cloverleaf skull (Keeblatt) Kleeblattschädel *(m) Bony occipital spur* *(m) Frontal bossing* *(m) Hypotelorism* *(m) Localized skull defects* *(m) Midfacial hypoplasia*	
Q75.9	Congenital malformation of skull and face bones, unspecified Congenital anomaly of face bones, NOS Congenital anomaly of skull, NOS	756.0
Q76	**Congenital malformations of spine and bony thorax** **Excludes 1:** congenital musculoskeletal deformities of spine and chest (Q67.5-Q67.8)	
Q76.0	Spina bifida occulta **Excludes 1:** meningocele (spinal) (Q05.-), spina bifida (aperta) (cystica) (Q05.-)	756.17
Q76.1	Klippel-Feil syndrome Klippel feil sequence Cervical fusion syndrome Wildervanck syndrome Cervicooculaoaccoustic syndrome	756.16
Q76.2	Congenital spondylolisthesis Congenital spondylolysis Congenital cervical spondylolisthesis Congenital lumbar spondylolisthesis Congenital lumbosacral spondylolysis Prespondylolisthesis, lumbosacral Spondyloysis, lumbosacral **Excludes 1:** spondylolisthesis (acquired) (M43.1-), spondylolysis (acquired) (M43.0-)	756.12
Q76.3	Congenital scoliosis due to congenital bony malformation Hemivertebra fusion or failure of segmentation with scoliosis	754.2
Q76.4	Other congenital malformations of spine, not associated with scoliosis	754.2
Q76.41	Congenital kyphosis	756.19
Q76.411	Congenital kyphosis, occipito-atlanto-axial region	756.19
Q76.412	Congenital kyphosis, cervical region	756.19
Q76.413	Congenital kyphosis, cervicothoracic region	756.19
Q76.414	Congenital kyphosis, thoracic region	756.19
Q76.415	Congenital kyphosis, thoracolumbar region	756.19
Q76.419	Congenital kyphosis, unspecified region	756.19
Q76.42	Congenital lordosis	754.2
Q76.425	Congenital lordosis, thoracolumbar region	754.2
Q76.426	Congenital lordosis, lumbar region	754.2
Q76.427	Congenital lordosis, lumbosacral region	754.2
Q76.428	Congenital lordosis, sacral and sacrococcygeal region	754.2
Q76.429	Congenital lordosis, unspecified region	754.2
Q76.49	Other congenital malformations of spine, not associated with scoliosis Congenital absence of vertebra, NOS Congenital fusion of spine, NOS Congenital malformation of lumbosacral (joint) (region), NOS	756.19 756.13 756.15 756.14

	Congenital malformation of spine, NOS Hemivertebrae, NOS Malformation of spine, NOS Platyspondylisis, NOS Supernumerary vertebra, NOS Sacrococcygeal anomaly Sacral mass, NOS Other specified vertebral anomaly Agenesis of cervical vertebrae Agenesis of thoracic vertebrae Agenesis of lumbar vertebrae Agenesis of sacrum Hemivertebrae, cervical vertebrae Hemivertebrae, thoracic vertebrae Hemivertebrae, lumbar vertebrae	
Q76.5	*(m) Cervical rib* *(m) Supernumerary rib in cervical region*	756.2
Q76.6	Other congenital malformations of ribs Accessory rib Congenital absence of rib Congenital fusion of ribs Congenital malformation of ribs, NOS **Excludes 1:** short rib syndrome (Q77.2)	756.3
Q76.7	Congenital malformations of sternum Congenital absence of sternum Sternum bifidum Congenital fissure of sternum Double ossification center in the manubrium Other anomaly of sternum Short sternum Anomalies of thoracic cage, unspecified	756.3
Q76.8	Other congenital malformations of bony thorax	756.3
Q76.9	Congenital malformation of bony thorax, unspecified	756.3
Q77	**Osteochondrodysplasia with defects of growth of tubular bones and spine** **Excludes 1:** mucopolysaccharidosis (E76.0-E76.3) **Excludes 2:** congenital myotonic chondrodystrophy (G71.13)	
Q77.0	Achondrogenesis Hypochondrogenesis	756.4
Q77.1	Thanatophoric short stature Thanatophoric dysplasia	756.4
Q77.2	Short rib syndrome Asphyxiating thoracic dysplasia (Jeune) Short rib dysplasia	756.3
Q77.3	Chondrodysplasia punctate Chondrodysplasia punctata (stippled epiphyses) group Conradi syndrome **Excludes 1:** Rhizomelic chondrodysplasia punctata (E71.43)	756.59
Q77.4	Achondroplasia Hypochondroplasia Osteosclerosis congenital	756.4
Q77.5	Diastrophic dysplasia	756.4
Q77.6	Chondroectodermal dysplasia Ellis-van Creveld syndrome	756.55
Q77.7	Spondyloepiphyseal dysplasia	756.4
Q77.8	Other osteochondrodysplasia with defects of growth of tubular bones and spine	756.4

Q77.9	Osteochondrodysplasia with defects of growth of tubular bones and spine, unspecified	756.4
Q78	**Other osteochondrodysplasias** **Excludes 2:** congenital myotonic chondrodystrophy (G71.13)	
Q78.0	Osteogenesis imperfecta Fragilitas ossium Osteopsathyrosis Brittle bones disease	756.51
Q78.1	Polyostotic fibrous dysplasia Albright (McCune) (Sternberg) syndrome Osteitis fibrosa disseminata	756.54
Q78.2	Osteopetrosis Albers-Schonberg syndrome Osteosclerosis, NOS Marble bones	756.52
Q78.3	Progressive diaphyseal dysplasia (multiple epiphyseal dysplasias) Camurati-Engelmann syndrome Engelmann syndrome	756.56
Q78.4	Enchondromatosis Maffucci's syndrome Ollier's disease	756.4
Q78.5	Metaphyseal dysplasia Pyle's syndrome	756.59
Q78.6	Multiple congenital exostoses Diaphyseal aclasis	756.6
Q78.8	Other specified osteochondrodysplasia Osteopoikilosis Melnick-needles syndrome	756.59
Q78.9	Osteochondrodysplasia, unspecified Chondrodystrophy, NOS Osteodystrophy, NOS	756.50
Q79	**Congenital malformations of musculoskeletal system, NEC** **Excludes 2:** congenital (sternomastoid) torticollis (Q68.0)	
Q79.0	Congenital diaphragmatic hernia **Excludes 1:** congenital hiatus hernia (Q40.1)	756.6
Q79.1	Other congenital malformations of diaphragm Absence of diaphragm Congenital malformation of diaphragm, NOS Eventration of diaphragm	756.6
Q79.2	Exomphalos Omphalocele **Excludes 1:** umbilical hernia (K42.-)	756.72
Q79.3	Gastroschisis	756.73
Q79.4	Prune belly syndrome Congenital prolapse of bladder mucosa Eagle-Barrett syndrome Early urethral obstruction sequence	756.71
Q79.5	Other congenital malformations of abdominal wall **Excludes 1:** umbilical hernia (K42.-)	756.79
Q79.51	Congenital hernia of bladder	756.71
Q79.59	Other congenital malformations of abdominal wall (specify) *(m) Diastasis recti* *(m) Unspecified anomaly of abdominal wall*	756.79
Q79.6	Ehlers-Danlos syndrome Cutis laxa hyperelastica	756.83
Q79.8	Other congenital malformations of musculoskeletal system	756.9

	Absence of muscle Absence of tendon Accessory muscle Amyotrophia congenital Congenital constricting bands Congenital shortening of tendon Poland syndrome Absent pectoralis major Other absent or hypoplastic muscle Other specified anomaly muscle, tendon, fascia, connective tissue Hypoplastic Depressor Angularis Muscle (face)	756.81 756.81 756.82
Q79.9	Congenital malformations of musculoskeletal system, unspecified Congenital anomaly of musculoskeletal system, NOS Congenital deformity of musculoskeletal system, NOS Unspecified anomaly of muscle Unspecified anomaly of tendon Unspecified anomaly of bone Unspecified anomaly of cartilage Unspecified anomaly of connective tissue Unspecified anomaly of musculoskeletal system (try not to use these codes)	756.9

Other Congenital Malformations (Q80-Q89)

ICD-10	Description	ICD-9
Q80	**Congenital ichthyosis Excludes 1:** Refsum's disease (G60.1)	
Q80.0	Ichthyosis vulgaris	757.1
Q80.1	X-linked ichthyosis	757.1
Q80.2	Lamellar ichthyosis Collodion baby	757.1
Q80.3	Congenital bullous ichthyosiform erythroderma	757.1
Q80.4	Harlequin fetus	757.1
Q80.8	Other congenital ichthyosis	757.1
Q80.9	Congenital ichthyosis, unspecified	757.1
Q81	**Epidermolysis bullosa**	
Q81.0	Epidermolysis bullosa simplex **Excludes 1:** Cockayne's syndrome (Q87.1)	757.39
Q81.1	Epidermolysis bullosa letalis Herlitz' syndrome	757.39
Q81.2	Epidermolysis bullosa dystrophica	757.39
Q81.8	Other epidermolysis bullosa	757.39
Q81.9	Epidermolysis bullosa, unspecified	757.39
Q82	**Other congenital malformations of skin** **Excludes 1:** acrodermatitis enteropathica (E83.2), congenital erythropoietic porphyria E80.0), pilonidal cyst or sinus (L05.-), Sturge-Weber (-Dimitri) syndrome (Q85.8)	
Q82.0	Hereditary lymphedema Congenital lymphedema Hereditary edema of legs Hereditary trophedema Milroy's disease	757.0
Q82.1	Xeroderma pigmentosum	757.33
Q82.2	Mastocytosis Urticaria pigmentosa **Excludes 1:** malignant mastocytosis (C96.2)	757.33
Q82.3	Incontinentia pigmenti	757.33
Q82.4	Ectodermal dysplasia (anhidrotic) X-linked type ectodermal dysplasia Other specified ectodermal dysplasias **Excludes 1:** Ellis-van Creveld syndrome (Q77.6)	757.31
Q82.5	*(m) Congenital non-neoplastic nevus* *(m) Birthmark, NOS* *(m) Flammeus Nevus* *(m) Portwine Nevus* *(m) Sanguineous Nevus* *(m) Strawberry Nevus* *(m) Vascular Nevus, NOS* *(m) Verrucous Nevus (m) Vascular hamartomas* *(m) Angel bite or kiss* *(m) Salmon patch* *(m) Stork bites* **Excludes 2:** Café au lait spots (L81.3), lentigo (L81.4), nevus NOS (D22.-), araneus nevus (I78.1), melanocytic nevus (D22.-), pigmented nevus (D22.-), spider nevus (I78.1), stellar nevus (I78.1)	757.32

Q82.8	Other specified congenital malformations of skin Benign familial pemphigus [Hailey-Hailey] Congenital poikiloderma Cutis laxa (hyperelastica) Inherited keratosis palmaris et plantaris Keratosis follicularis [Darier-White] Absence of skin Cutis aplasia Goltz syndrome *(m) Abnormal palmar creases* *(m) Accessory skin tags* *(m) Dermatoglyphic anomalies* *(m) Extra or absent creases* *(m) Sidney line* *(m) Simian creases, transverse palmar creases* *(m) Single transverse palmar crease* *(m) Sole crease* *(m) Unusual dermatoglyphics* *(m) Anal tags* *(m) Skin tags* *(m) Congenital scar* *(m) Cutis marmorata* *(m) Hypoplastic dermal patterns* *(m) Congenital keraderma* *(m) Mongolian spot* *(m) Depigmented or hyperpigmented spot* *(m) Hyperpigmented areas* **Excludes 1:** Ehlers-Danlos syndrome (Q79.6)	757.39
Q82.9	Congenital malformation of skin, unspecified	757.39
Q83	**Congenital malformations of breast** **Excludes 2:** absence of pectoral muscle (Q79.8), hypoplasia of breast (N64.82), micromastia (N64.82)	
Q83.0	Congenital absence of breast with absent nipple	757.6
Q83.1	*(m) Accessory breast* *(m) Supernumerary breast*	757.6
Q83.2	Absent nipple	757.6
Q83.3	*(m) Accessory nipple* *(m) Supernumerary nipple*	757.6
Q83.8	Other congenital malformations of breast Hypoplastic breast with hypoplastic nipple *(m) Asymmetry of breasts, congenital* *(m) Widely spaced nipples* *(m) Small nipple (hypoplastic)*	757.6
Q83.9	Congenital malformation of breast, unspecified	757.6
Q84	**Other congenital malformations of integument**	
Q84.0	*(m) Congenital alopecia* *(m) Congenital atrichosis*	757.4
Q84.1	*(m) Congenital morphological disturbances of hair, NEC* *(m) Beaded hair* *(m) Monilethrix* *(m) Pili annulati* *(m) Pili torti* *(m) Twisted hair* *(m) Taenzer's hair* **Excludes 1:** Menkes' kinky hair syndrome (E83.0)	757.4

Q84.2	*(m) Other congenital malformations of hair* *(m) Congenital hypertrichosis* *(m) Congenital malformation of hair, NOS* *(m) Persistent lanugo* *(m) Hirsutism* *(m) Aberrant scalp hair patterning* *(m) Low hair line* *(m) Other specified anomaly of hair*	757.4
Q84.3	*(m) Anonychia* *(m) Absent nails* **Excludes 1:** nail patella syndrome (Q87.2)	757.5
Q84.4	*(m) Congenital leukonychia*	757.5
Q84.5	*(m) Enlarged and hypertrophic nails* *(m) Congenital onychauxis* *(m) Pachyonychia* *(m) Hyperconvex or thickened toenails*	757.5
Q84.6	Hypoplastic, hyperconvex, or duplicated fingernails *(m) Other congenital malformations of nails* *(m) Congenital clubnail* *(m) Congenital koilonychias* *(m) Congenital malformations of nail, NOS* *(m) Hypoplastic (small) fingernails and/or toenails*	757.5
Q84.8	Other specified congenital malformations of integument Aplasia cutis congenital *(m) Scalp defects*	757.8
Q84.9	*(m) Congenital malformation of integument, unspecified* *(m) Congenital anomaly of integument, NOS* *(m) Congenital deformity of integument, NOS* *(m) Unspecified anomaly of hair* *(m) Unspecified anomaly of nail* *(m) Unspecified anomaly of the integument*	757.9
Q85	**Phakomatoses, NEC** **Excludes 1:** ataxia telangiectasia [Louis-Bar] (G11.3), familial dysautonomia [Riley-Day] (G90.1)	
Q85.0	**Neurofibromatosis (nonmalignant)**	
Q85.00	Neurofibromatosis, unspecified	237.70
Q85.01	Neurofibromatosis, Type 1 Von Recklinghausen disease	237.71
Q85.02	Neurofibromatosis, Type 2 Acoustic neurofibromatosis	237.72
Q85.03	Schwannomatosis	237.73
Q85.09	Other neurofibromatosis	237.79
Q85.1	Tuberous sclerosis Bourneville's disease Epiloia	759.5
Q85.8	Other phakomatoses, NEC Peutz-Jeghers Syndrome Studge-Weber (-Dimitri) syndrome Von Hippel-Lindau syndrome Neuro-cutaneous syndrome **Excludes 1:** Meckel-Gruber syndrome (Q61.9)	759.6
Q85.9	Phakomatosis, unspecified Hamartosis, NOS	759.6
Q86	**Congenital malformations syndromes due to known exogenous causes, NEC** **Excludes 2:** iodine-deficiency-related hypothyroidism (E00-E02), nonteratogenic effects of substances transmitted via placenta or breast milk (P04.-)	

Q86.0	Fetal alcohol syndrome (dysmorphic)	760.71
Q86.1	Fetal hydantoin syndrome Meadow's syndrome	760.77
Q86.2	Dysmorphism due to warfarin	760.79
Q86.8	Other congenital malformation syndromes due to known exogenous causes	760.76
Q87	**Other specified congenital malformation syndromes affecting multiple systems** ****Use Additional code(s) to identify all associated manifestations**	
Q87.0	Congenital malformation syndromes predominantly affecting facial appearance Acrocephalopolysyndactyly Acrocephalosyndactyly [Apert] Cryptophthalmos syndrome Cyclopia Goldenhar syndrome Moebius syndrome Oro-facial-digital syndrome Robin syndrome Whistling face Hallerman-Streiff syndrome Pierre Robin syndrome Goldenhar syndrome (oculo-auriculovertebral) Carpenter's syndrome Antley-Bixler	755.55 756.0 759.89
Q87.1	Congenital malformation syndromes predominantly associated w/ short stature Aarskog syndrome Cockayne syndrome De Lange syndrome Dubowitz syndrome Noonan syndrome Prader-willi syndrome Robinow-Silverman-Smith syndrome Russell-Silver syndrome Seckel syndrome Bonnevie-Ullrich syndrome Cornelia de Lange syndrome Sjogren-Larsson syndrome **Excludes 1:** Ellis-van Creveld syndrome (Q77.6), Smith-Lemli-Opitz syndrome (E78.72)	759.81 759.89
Q87.2	Congenital malformation syndromes predominantly involving limbs Holt-Oram syndrome Klippel-Trenaunay-Weber syndrome Nail patella syndrome Rubinstein-Taybi syndrome Sirenomelia syndrome Thrombocytopenia with absent radius [TAR] syndrome VATER syndrome	759.89
Q87.3	Congenital malformation syndromes involving early overgrowth Beckwith-Wiedemann syndrome Sotos syndrome Weaver syndrome Proteus syndrome	759.89
Q87.4	Marfan's syndrome	
Q87.40	Marfan's syndrome, unspecified	759.82
Q87.41	Marfan's syndrome with cardiovascular manifestations	
Q87.410	Marfan's syndrome with aortic dilation	759.82
Q87.418	Marfan's syndrome with other cardiovascular manifestations	759.82

Q87.42	Marfan's syndrome with ocular manifestations	759.82
Q87.43	Marfan's syndrome with skeletal manifestation	759.82
Q87.5	Other congenital malformation syndromes with other skeletal changes	759.89
Q87.8	Other specified congenital malformation syndromes, NEC **Excludes 1:** Zellweger syndrome (E71.510)	
Q87.81	Alport syndrome Hereditary nephritis Use Additional code to identify stage of chronic kidney disease (N18.1-N18.6)	759.89
Q87.89	Other specified congenital malformation syndromes, NEC Laurence-Moon (-Bardet)-Biedl syndrome Basal cell nevus syndrome Gorlin syndrome	759.89
Q89	**Other congenital malformations, NEC**	
Q89.0	Congenital absence and malformations of spleen **Excludes 1:** isomerism of atrial appendages (with asplenia or polysplenia) (Q20.6)	
Q89.01	Asplenia (congenital)	759.0
Q89.09	Congenital malformations of spleen Congenital splenomegaly	759.0
Q89.1	Congenital malformations of adrenal gland **Excludes 1:** adrenogenital disorders (E25.-), congenital adrenal hyperplasia (E25.0)	759.1
Q89.2	Congenital malformations of other endocrine glands Congenital malformation of parathyroid or thyroid gland Persistent thyroglossal duct Thyroglossal cyst **Excludes 1:** congenital goiter (E03.0), congenital hypothyroidism (E03.1)	759.2
Q89.3	Situs inversus Dextrocardia with situs inversus Mirror-image atrial arrangement with situs inversus Situs inversus or transversus abdominalis Situs inversus or transversus thoracis Transposition of abdominal viscera Transposition of thoracic viscera **Excludes 1:** dextrocardia NOS (Q24.0)	759.3
Q89.4	Conjoined twins Craniopagus Cicephaly Pygopagus Thoracopagus	759.4
Q89.7	Multiple congenital malformations, NEC Multiple congenital anomalies, NOS Multiple congenital deformities, NOS **Excludes 1:** congenital malformation syndromes affecting multiple systems (Q87.-)	759.7
Q89.8	Other specified congenital malformations Use additional code(s) to identify all associated manifestations	759.89
Q89.9	Congenital malformation, unspecified Congenital anomaly, NOS Congenital deformity, NOS	759.9

Chromosomal Abnormalities, Not Elsewhere Classified (NEC) (Q90-Q99)

**** Please report karyotype (chromosome test results) in addition to the ICD code!**

ICD-10	Description	ICD-9
Q90	**Trisomy 21, Down Syndrome**	
Q90.0	Trisomy 21, nonmosaicism (meiotic nondisjunction) Karyotype trisomy 21 (47,XX or XY,+21)	758.0
Q90.1	Trisomy 21, mosaicism (mitotic nondisjunction) (46,XX/47,XX+21 or 46,XY/47,XY+21)	758.0
Q90.2	Trisomy 21, translocation t(13:21), t(14:21), t(15:21), t(21:22) Translocation trisomy duplication of a 21[t(21:21)]	758.0
Q90.9	Down syndrome, unspecified Trisomy 21, NOS	758.0
Q91	**Trisomy 18 and Trisomy 13**	
Q91.0	Trisomy 18, nonmosaicism (meiotic nondisjunction) Edwards syndrome, karyotype trisomy 18(47,XX or XY+18)	758.2
Q91.1	Trisomy 18, mosaicism (mitotic nondisjunction) (46,XX/47,XX+18 or 46,XY/47,XY+18)	758.2
Q91.2	Trisomy 18, translocation	758.2
Q91.3	Trisomy 18, unspecified Edwards syndrome, NOS	758.2
Q91.4	Trisomy 13, nonmosaicism (meiotic nondisjunction) Patau syndrome, karyotype trisomy 13(47,XX or XY,+13)	758.1
Q91.5	Trisomy 13, mosaicism nondisjunction) (46,XX/47, XX+13 or 46,XY/47,XY+13)	758.1
Q91.6	Trisomy 13, (translocation) t(13:14), t(13:21), t(13:15), t(13:22), t(13:13)	758.1
Q91.7	Trisomy 13, unspecified Patau syndrome, NOS	758.1
Q92	**Other trisomies and partial trisomies of the autosome, NEC** **Includes:** unbalanced transolocations and insertions **Excludes 1:** trisomies of chromosomes 13, 18, 21 (Q90-Q91)	
Q92.0	Whole chromosomes trisomy, nonmosaicism (meiotic nondisjunction) • Trisomy 1 • Trisomy 2 • Trisomy 3 • Trisomy 4 • Trisomy 5 • Trisomy 6 • Trisomy 7 • Trisomy 8 • Trisomy 9 • Trisomy 10 • Trisomy 11 • Trisomy 12 • Trisomy 14 • Trisomy 15 • Trisomy 16 • Trisomy 17 • Trisomy 19 • Trisomy 20 • Trisomy 22	758.5
Q92.1	Whole chromosome trisomy, mosaicism (mitotic nondisjunction) Mosaic trisomy of any chromosome except 21, 13 or 18	758.5
Q92.2	Partial trisomy Less than whole arm duplicated Whole arm or more duplicated **Excludes 1:** partial trisomy due to unbalanced translocation (Q92.5)	758.2
Q92.5	Duplications with other complex rearrangements Partial trisomy due to unbalanced translocations Partial trisomy syndromes (unbalanced, not defined) **Also code any associated deletions due to unbalanced translocations, inversions and insertions	758.5
Q92.6	Marker chromosomes Trisomies due to dicentrics	758.5

	Trisomies due to extra rings Trisomies due to isochromosomes Individual with marker heterochromatin	
Q92.61	Marker chromosomes in normal individual	758.5
Q92.62	Marker chromosomes in abnormal individual	758.5
Q92.7	Triploidy and polyploidy Triploidy, 69 chromosomes Other Polyploidy (96 chromosomes or other multiples of 23, except 46,69)	758.5
Q92.8	Other specified trisomies and partial trisomies of autosomes (chromosomes 1-22) Duplication identified by fluorescence in situ hybridization (FISH) Duplications identified by in situ hybridization (ISH) Duplications seen only at prometaphase	758.5
Q92.9	Trisomy and partial trisomy of autosomes, unspecified Trisomy, NOS	758.5
Q93	**Monosomies and deletions from the autosomes, NEC**	
Q93.0	Whole chromosomes monosomy, nonmosaicism (meiotic nondisjunction) E.g. (45, XX, -21) or (45,XY,-21) for any chromosome #1-22	758.5
Q93.1	Whole chromosome monosomy, mosaicism (mitotic nondisjunction) Monosomy 21 mosaicism: e.g.: (46,XX/45,XX,-21) or (46,XY/45,XY,-21) Monosomy of other whole chromosomes #1-22 in mosaic form	758.5
Q93.2	Chromosome replaced with ring, dicentric or isochromosomes	758.5
Q93.3	Deletion of short arm of chromosome 4 Wolff-Hirschorn syndrome 4p deletion syndrome del4p15 Deletion 4p15-16	758.39
Q93.4	Deletion of short arm of chromosome 5 Cri-du-chat syndrome (5p-)	758.31
Q93.5	Other specified deletions of part of any chromosome #1-22 such as: Specific deletion of chromosome 15 resulting in Angelman syndrome Antimongolism syndrome: partial or total deletion of 21 [del (21)] Deletion of long arm 13: (del(13q)) or 13q- Deletion of long arm 17: (del(17q)) or 17q- Deletion of long arm 18: (del(18q)) or 18q- Deletion of short arm 17: (del(17p)) or 17p- Deletion of short arm 18: (del(18p)) or 18p-	758.39
Q93.7	Deletions with other complex rearrangements Deletions due to unbalanced translocations, inversions and insertions **Code also any associated duplications due to unbalanced translocations, inversions and insertions	758.39
Q93.8	**Other deletions from the autosomes**	
Q93.81	Velo-cardio-facial syndrome Deletion 22q11.2	758.32
Q93.88	Other microdeletions Miller-Dieker syndrome Smith-Magenis syndrome	758.33
Q93.89	Other deletions from the autosomes (chromosomes 1-22) Deletions identified by fluorescence in situ hybridization (FISH) Deletions identified by in situ hybridization (IS) Deletions seen only at prometaphase	758.39
Q93.9	Deletion from autosomes, unspecified Autosomal deletion, NOS	758.39
Q95	**Balanced rearrangements and structural markers, NEC** **Includes:** Robertsonian and balanced reciprocal translocations and insertions	
Q95.0	Balanced translocation and insertion in normal individual	758.4

Q95.1	Chromosome inversion in normal individual	758.4
Q95.2	Balanced autosomal rearrangement in abnormal individual	758.5
Q95.3	Balanced sex/autosomal rearrangement in abnormal individual	758.5
Q95.5	Individual with autosomal fragile site Autosomal fragile site [fra (_)]	758.4
Q95.8	Other balanced rearrangements and structural markers	758.4
Q95.9	Balanced rearrangement and structural marker, unspecified	758.4
Q96	**Turner's syndrome** **XO syndrome** **Bonnevie-Ullrich syndrome** **[Only applicable to female patients.]** **Excludes 1:** Noonan syndrome (Q87.1), pure gonadal dysgenesis (Q99.1)	
Q96.0	Karyotype 45,X Turner's phenotype, karyotype 45,X	758.6
Q96.1	Karyotype 46, X iso (Xq) Karyotype 46, isochromosome Xq	758.6
Q96.2	Karyotype 46, X with abnormal sex chromosome, except iso (Xq) Karyotype 46, X with abnormal sex chromosome, except isochromosome Xq Turner syndrome variant, [46,X,r(X)] Turner syndrome variant, [46,Xdel(Xq)] Turner syndrome variant, [46,X,del(Xp)]	758.6
Q96.3	Mosaicism, 45, X/46, XX or XY	758.6
Q96.4	Mosaicism, 45, X/other cell line(s) with abnormal sex chromosome	756.6
Q96.8	Other variants of Turner's syndrome	758.6
Q96.9	Turner's syndrome, unspecified Turner Syndrome, karyotype unspecified	758.6
Q97	**Other sex chromosome abnormalities, female phenotype, NEC** **[Only applicable to female patients.]** **Excludes 1:** Turner's syndrome (Q96.-)	
Q97.0	Karyotype 47, XXX	758.81
Q97.1	Female with more than three X chromosomes	758.81
Q97.2	Mosaicism, lines with various numbers of X chromosomes	758.81
Q97.3	Female with 46, XY karyotype	758.81
Q97.8	Other specified sex chromosome abnormalities, female phenotype	758.81
Q97.9	Sex chromosome abnormalities, female phenotype unspecified	758.81
Q98	**Other sex chromosome abnormalities, male phenotype, NEC [Only applicable to male patients.]**	
Q98.0	Klinefelter syndrome karyotype 47, XXY	758.7
Q98.1	Klinefelter syndrome, male with more than two X chromosome 48,XXXY 48,XXYY 49,XXXXY	758.7
Q98.3	Other male with 46, XX karyotype	758.7
Q98.4	Klinefelter syndrome, unspecified	758.7
Q98.5	Karyotype 47, XYY	758.81
Q98.6	Male with structurally abnormal sex chromosome	758.81
Q98.7	Male with sex chromosome mosaicism Klinefelter syndrome mosaic (46,XY/47,XXY) Mosaic X/XY, 45X/46XY Mosaic XY/XXY, 46XY/47XXY Mosaic including XXXXY,49XXXXY Mosaic XYY male[46,XY/47XYY] Sex chromosomes mosaicism	758.81
Q98.8	Other specified sex chromosome abnormalities, male phenotype	758.81
Q98.9	Sex chromosome abnormality, male phenotype unspecified	758.81

Q99	Other chromosome abnormalities, NEC	
Q99.0	Chimera 46, XX/46, XY Chimera 46, XX/46, XY true hermaphrodite	758.81
Q99.1	46, XX true hermaphrodite 46, XX with streak gonads 46, XY with streak gonads Pure gonadal dysgenesis	758.81
Q99.2	Fragile X chromosome Fragile X syndrome	759.83
Q99.8	Other specified chromosome abnormalities Mosaicism, NOS Additional chromosome(s), NOS Deletion of chromosome(s), NOS Duplication of chromosome(s), NOS Accessory autosomes, N Other specified anomaly of autosomes, includes satellites, stalks, excess Derivative chromosomes, NOS [not X or Y;der (_)] Double minute chromosomes (+dmidn) Unspecified sex chromosome anomaly Isochromosome, autosome [I (_)] Variable region, autosome [var (_)] Insertion, autosome [inv (_)] Inversion, autosome [inv (_)]	758.89
Q99.9	Chromosomal abnormality, unspecified (try not to use)	758.9

Other Selected Reportable Conditions

ICD-10	Notes & Description	ICD-9
D55	**Anemia due to enzyme disorders Excludes 1:** drug-induced enzyme deficiency anemia (D59.2)	
D55.0	Anemia due to glucose-6-phosphate dehydrogenase [G6PD] deficiency	282.2
D55.1	Anemia due to other disorders of glutathione metabolism Anemia (due to) enzyme deficiencies, except G6PD, related to the hexose monophosphate [HMP] shunt pathway Anemia (due to) hemolytic nonspherocytic (hereditary), type I	282.2
D55.2	Anemia due to disorders of glycolytic enzymes **Excludes 1:** disorders of glycolysis not associated with anemia (E74.8)	282.3
D55.3	Anemia due to disorders of nucleotide metabolism	282.3
D58	**Other hereditary hemolytic anemias Excludes 1:** hemolytic anemia of the newborn (P55.-)	
D58.0	Hereditary spherocytosis Acholuric (familial) jaundice Congenital (spherocytic) hemolytic icterus Minkowski-Chauffard syndrome	282
D58.1	Hereditary elliptocytosis Elliptocytosis (congenital) Ovalocytosis (congenital) (hereditary)	282.1
D58.2	Other hemoglobinopathies Abnormal hemoglobin NOS Congenital Heinz body anemia Hb-C disease Hb-D disease Hb-E disease Hemoglobinopathy NOS Unstable hemoglobin hemolytic disease **Excludes 1:** familial polycythemia (D75.0), Hb-M disease (D74.0), hemoglobin E-beta thalassemia (D56.5), hereditary persistence of fetal hemoglobin [HPFH] (D56.4), high-altitude polycythemia (D75.1), methemoglobinemia (D74.-), other hemoglobinopathies with thalassemia (D56.8)	282.7
D58.8	Other specified hereditary hemolytic anemias Stomatocytosis	282.8
D58.9	Hereditary hemolytic anemia, unspecified	282.9
D61	**Other aplastic anemias and other bone marrow failure syndromes Excludes 1:** neutropenia (D70.-)	
D61.0	Constitutional aplastic anemia	
D61.01	Constitutional (pure) red blood cell aplasia Blackfan-Diamond syndrome Congenital (pure) red cell aplasia Familial hypoplastic anemia Primary (pure) red cell aplasia Red cell (pure) aplasia of infants **Excludes 1:** acquired red cell aplasia (D60.9)	284.01
D61.09	Other constitutional aplastic anemia Fanconi's anemia Pancytopenia with malformations	284.09
D64	**Other anemias Excludes 1:** refractory anemia (D46.-), refractory anemia with excess blasts in transformation [RAEB T] (C92.0-)	
D64.0	Hereditary sideroblastic anemia Sex-linked hypochromic sideroblastic anemia	285
D64.4	Congenital dyserythropoietic anemia Dyshematopoietic anemia (congenital)	285.8

	Excludes 1: Blackfan-Diamond syndrome (D61.01), Di Guglielmo's disease (C94.0)	
D66	Hereditary factor VIII deficiency Classical hemophilia Deficiency factor VIII (with functional defect) Hemophilia NOS Hemophilia A **Excludes 1:** factor VIII deficiency with vascular defect (D68.0)	286
D67	Hereditary factor IX deficiency Christmas disease Factor IX deficiency (with functional defect) Hemophilia B Plasma thromboplastin component [PTC] deficiency	286.1
D68	**Other coagulation defects Excludes 1:** abnormal coagulation profile (R79.1), coagulation defects complicating abortion or ectopic or molar pregnancy (O00-O07, O08.1), coagulation defects complicating pregnancy, childbirth and the puerperium (O45.0, O46.0, O67.0, O72.3)	
D68.0	Von Willebrand's disease Angiohemophilia Factor VIII deficiency with vascular defect Vascular hemophilia **Excludes 1:** capillary fragility (hereditary) (D69.8), factor VIII deficiency NOS (D66), factor VIII deficiency with functional defect (D66)	286.4
D68.1	Hereditary factor XI deficiency Hemophilia C Plasma thromboplastin antecedent [PTA] deficiency Rosenthal's disease	286.2
D68.2	Hereditary deficiency of other clotting factors AC globulin deficiency Congenital afibrinogenemia Deficiency of factor I [fibrinogen] Deficiency of factor II [prothrombin] Deficiency of factor V [labile] Deficiency of factor VII [stable] Deficiency of factor X [Stuart-Prower] Deficiency of factor XII [Hageman] Deficiency of factor XIII [fibrin stabilizing] Dysfibrinogenemia (congenital) Hypoproconvertinemia Owren's disease Proaccelerin deficiency	286.3
D70	**Neutropenia** **Includes:** Agranulocytosis, decreased absolute neurophile count (ANC) **Excludes 1:** neutropenic splenomegaly (D73.81), transient neonatal neutropenia (P61.5)	
D70.0	Congenital agranulocytosis Congenital neutropenia Infantile genetic agranulocytosis Kostmann's disease	288.1
D72	**Other disorders of white blood cells** **Excludes 1:** basophilia (D72.824), immunity disorders (D80-D89), neutropenia (D70), preleukemia (syndrome) (D46.9)	
D72.0	Genetic anomalies of leukocytes Alder (granulation) (granulocyte) anomaly Alder syndrome Hereditary leukocytic hypersegmentation Hereditary leukocytic hyposegmentation Hereditary leukomelanopathy	288.2

	May-Hegglin (granulation) (granulocyte) anomaly May-Hegglin syndrome Pelger-Huet (granulation) (granulocyte) anomaly Pelger-Huet syndrome **Excludes 1:** Chediak (-Steinbrinck)-Higashi syndrome (E70.330)	
D72.1	Only report if hereditary eosinohilia – DO NOT REPORT if allergic **Excludes 1:** Leffker's syndrome (J82), pulmonary eosinophilia (J82)	288.3
D74	**Methemoglobinemia**	
D74.0	Congenital methemoglobinemia Congenital NADH-methemoglobin reductase deficiency Hemoglobin-M [Hb-M] disease Methemoglobinemia, hereditary	289.7
D80	**Immunodeficiency with predominantly antibody defects**	
D80.0	Hereditary hypogammaglobulinemia Autosomal recessive agammaglobulinemia (Swiss type) X-linked agammaglobulinemia [Bruton] (with growth hormone deficiency	279.04
D80.2	Selective deficiency of immunoglobulin A [IgA]	279.01
D80.3	Selective deficiency of immunoglobulin G [IgG] subclasses	279.03
D80.4	Selective deficiency of immunoglobulin M [IgM]	279.02
D80.5	Immunodeficiency with increased immunoglobulin M [IgM]	279.05
D80.6	Antibody deficiency with near-normal immunoglobulins or with hyperimmunoglobulinemia	279.19
D80.7	Transient hypogammaglobulinemia of infancy	279.09
D80.8	Other immunodeficiencies with predominantly antibody defects Kappa light chain deficiency	279.19
D80.9	Immunodeficiency with predominantly antibody defects, unspecified	279.19
D81	**Combined immunodeficiencies** **Excludes 1:** autosomal recessive agammaglobulinemia (Swiss type) (D80.0)	
D81.0	Severe combined immunodeficiency (SCID) with reticular dysgenesis	279.2
D81.1	Severe combined immunodeficiency (SCID) with low T- and B-cell numbers	279.2
D81.2	Severe combined immunodeficiency (SCID) with low or normal B-cell numbers	279.2
D81.4	Nezelof's syndrome	279.13
D81.6	Major histocompatibility complex class I deficiency	279.2
D81.7	Major histocompatibility complex class II deficiency	279.2
D81.810	Biotinidase deficiency	277.6
D81.89	Other combined immunodeficiency	279.2
D81.9	Combined immunodeficiency, unspecified Severe combined immunodeficiency disorder [SCID] NOS	279.2
D82	**Immunodeficiency associated with other major defects** **Excludes 1:** ataxia telangiectasia [Louis-Bar] (G11.3)	
D82.0	Wiskott-Aldrich syndrome Immunodeficiency with thrombocytopenia and eczema	279.12
D82.1	Di George's syndrome Also code 22q deletion separately if reported • Pharyngeal pouch syndrome • Thymic alymphoplasia • Thymic aplasia or hypoplasia with immunodeficiency	279.11
D82.2	Immunodeficiency with short-limbed stature	279.8
D82.3	X-linked lymphoproliferative disease	279.8
D82.4	Hyperimmunoglobulin E [IgE] syndrome	279.8
D82.8	Immunodeficiency associated with other specified major defects	279.8
D82.9	Immunodeficiency associated with major defect, unspecified	279.8
E00	**Congenital iodine-deficiency syndrome** **Excludes 1:** subclinical iodine-deficiency hypothyroidism (E02) ** Do not report if birth weight less than 1500 grams	

E00.0	Congenital iodine-deficiency syndrome, neurological type Endemic cretinism, neurological type	243
E00.1	Congenital iodine-deficiency syndrome, myxedematous type Endemic hypothyroid cretinism Endemic cretinism, myxedematous type	243
E00.2	Congenital iodine-deficiency syndrome, mixed type Endemic cretinism, mixed type	243
E00.9	Congenital iodine-deficiency syndrome, unspecified congenital iodine-deficiency Hypothyroidism NOS Endemic cretinism, NOS Hypothyroidism, congenital	243
E03	**Other hypothyroidism** ** Do not report if birth weight less than 1500 grams **Excludes 1:** iodine-deficiency related hypothyroidism (E00-E02), postprocedural hypothyroidism (E89.0)	
E03.0	Congenital hypothyroidism with diffuse goiter	243
E03.1	Congenital hypothyroidism without goiter	243
E07	**Other disorders of thyroid** ** Do not report if birth weight less than 1500 grams	
E07.1	Congenital dyshormogenetic goiter Familial dyshormogenetic goiter Pendred's syndrome **Excludes 1:** transitory congenital goiter with normal function (P72.0)	246.1
E22	**Hyperfunction of pituitary gland** **Excludes 1:** Cushing's syndrome (E24.-), Nelson's syndrome (E24.1), overproduction of ACTH not associated with Cushing's disease (E27.0), overproduction of pituitary ACTH (E24.0), overproduction of thyroid-stimulating hormone (E05.8-)	
E22.0	Acromegaly and pituitary gigantism Overproduction of growth hormone **Excludes 1:** constitutional gigantism (E34.4), constitutional tall stature (E34.4), increased secretion from endocrine pancreas of growth hormone-releasing hormone (E16.8)	253.0
E23	**Hypofunction and other disorders of the pituitary gland** Only report if present at birth or report if genetic-based cause **Includes** the listed conditions whether the disorder is in the pituitary or the hypothalamus **Excludes 1:** postprocedural hypopituitarism (E89.3)	
E23.0	Hypopituitarism Fertile eunuch syndrome Hypogonadotropic hypogonadism Idiopathic growth hormone deficiency Isolated deficiency of gonadotropin Isolated deficiency of growth hormone Isolated deficiency of pituitary hormone Kallmann's syndrome Lorain-Levi short stature Necrosis of pituitary gland (postpartum) Panhypopituitarism, congenital Pituitary cachexia Pituitary insufficiency NOS Pituitary short stature Sheehan's syndrome Simmonds' disease Pituitary dwarfism	253.2 253.3
E23.2	Diabetes insipidus **Excludes 1:** nephrogenic diabetes insipidus (N25.1)	253.5
E25	**Adrenogenital disorders**	

	Please report cytogenetic diagnosis (Congenital Adrenal Hyperplasia does not require cytogenetic diagnosis) **Includes:** adrenogenital syndromes, virilizing or feminizing, whether acquired or due to adrenal hyperplasia consequent on inborn enzyme defects in hormone synthesis, Female adrenal pseudohermaphroditism, Female heterosexual precocious pseudopuberty, Male isosexual precocious pseudopuberty, Male macrogenitosomia praecox, Male sexual precocity with adrenal hyperplasia, Male virilization (female) **Excludes 1:** indeterminate sex and pseudohermaphroditism (Q56), chromosomal abnormalities (Q90-Q99)	
E25.0	Congenital adrenogenital disorders associated with enzyme deficiency Congenital adrenal hyperplasia 21-Hydroxylase deficiency Salt-losing congenital adrenal hyperplasia	255.2
E25.8	Other adrenogenital disorders Idiopathic adrenogenital disorder	255.2
E25.9	Adrenogenital disorder, unspecified Adrenogenital syndrome NOS	255.2
E34	**Other endocrine disorders ** only report if congenital or genetic** **Excludes 1:** pseudohypoparathyroidism (E20.1)	
E34.3	Short stature due to endocrine disorder Laron-type short stature **Excludes 1:** achondroplastic short stature (Q77.4), hypochondroplastic short stature (Q77.4), nutritional short stature (E45), pituitary short stature (E23.0), progeria (E34.8), renal short stature (N25.0), Russell-Silver syndrome (Q87.1), short-limbed stature with immunodeficiency (D82.2), short stature in specific dysmorphic syndromes - code to syndrome, short stature, NOS (R62.52)	259.4
E34.5	**Androgen insensitivity syndrome**	
E34.50	Androgen insensitivity syndrome, unspecified	259.5
E34.51	Complete androgen insensitivity syndrome Complete androgen insensitivity de Quervain syndrome Goldberg-Maxwell syndrome Testicular feminization	259.5
E34.52	Partial androgen insensitivity syndrome Partial androgen insensitivity Reifenstein syndrome	259.5
E34.8	Other specified endocrine disorders Pineal gland dysfunction Progeria Hutchinson-Gilford syndrome **Excludes 2:** pseudohypoparathyroidism (E20.1)	259.8
E70	**Disorders of aromatic amino-acid metabolism**	
E70.0	Classical phenylketonuria	270.1
E70.1	Other hyperphenylalaninemias	270.1
E70.2	**Disorders of tyrosine metabolism** **Excludes 1:** transitory tyrosinemia of newborn (P74.5)	
E70.20	**Disorder of tyrosine metabolism, unspecified**	
E70.21	Tyrosinemia Hypertyrosinemia	270.2
E70.29	Other disorders of tyrosine metabolism Alkaptonuria Ochronosis	270.2
E70.3	**Albinism**	
E70.30	Albinism, unspecified	270.2
E70.31	**Ocular albinism**	
E70.310	X-lined ocular albinism	270.2

E70.311	Autosomal recessive ocular albinism	270.2
E70.318	Other ocular albinism	270.2
E70.319	Ocular albinism, unspecified	270.2
E70.32	**Oculocutaneous albinism** **Excludes 1:** Chediak-Higashi syndrome (E70.330), Hermansky-Pudlak syndrome (E70.331)	
E70.320	Tyrosinase negative oculocutaneous albinism Albinism I Oculocutaneous albinism ty-neg Tyrosinase-negative oculocutaneous albinism	270.2
E70.321	Tyrosinase positive oculocutaneous albinism Albinism II Oculocutaneous albinism ty-pos Tyrosinase-positive oculocutaneous albinism	270.2
E70.328	Other oculocutaneous albinism Cross syndrome	270.2
E70.329	Oculocutaneous albinism, unspecified	270.2
E70.33	**Albinism with hematologic abnormality**	
E70.330	Chediak-Higashi syndrome	270.2
E70.331	Hermansky-Pudlak syndrome	270.2
E70.338	Other albinism with hematologic abnormality	270.2
E70.339	Albinism with hematologic abnormality, unspecified	270.2
E70.39	Other specified albinism Piebaldism	270.2
E70.4	**Disorders of histidine metabolism**	
E70.40	Disorders of histidine metabolism, unspecified	270.5
E70.41	Histidinemia	270.5
E70.49	Other disorders of histidine metabolism	270.5
E70.5	Disorders of tryptophan metabolism	270.2
E70.8	Other disorders of aromatic amino-acid metabolism	270.2
E70.9	Disorder of aromatic amino-acid metabolism, unspecified	270.2
E71	**Disorders of branched-chain amino-acid metabolism and fatty-acid metabolism**	
E71.0	Maple-syrup-urine disease	270.3
E71.1	Other disorders of branched-chain amino-acid metabolism	270.3
E71.11	Branched-chain organic acidurias	
E71.110	Isovaleric acidemia	270.3
E71.111	3-methylglutaconic aciduria	270.3
E71.118	Other branched-chain organic acidurias	270.3
E71.12	Disorders of propionate metabolism	
E71.120	Methylmalonic acidemia	270.3
E71.121	Propionic acidemia	270.3
E71.128	Other disorders of propionate metabolism	270.3
E71.19	Other disorders of branched-chain amino-acid metabolism	270.3
E71.2	Disorders of branched-chain amino-acid metabolism, unspecified	270.3
E71.3	**Disorders of fatty-acid metabolism** **Excludes 1:** peroxisomal disorders (E71.5), Refsum's disease (G60.1), Schilder's disease (G37.0) **Excludes 2:** carnitine deficiency due to inborn error of metabolism (E71.42)	
E71.31	**Disorders of fatty-acid oxidation**	
E71.310	Long chain/very long chain acyl CoA dehydrogenase deficiency LCAD VLCAD	277.85
E71.311	Medium chain acyl CoA dehydrogenase deficiency MCAD	277.85
E71.312	Short chain acyl CoA dehydrogenase deficiency	277.85

	SCAD	
E71.313	Glutaric aciduria type II (A, B or C) **Excludes 1:** glutaric aciduria (type 1), NOS (E72.3)	277.85
E71.314	Muscle carnitine palmitoyltransferase deficiency	277.85
E71.318	Other disorders of fatty-acid oxidation	277.85
E71.32	Disorders of ketone metabolism	277.85
E71.39	Other disorders of fatty-acid metabolism	277.89
E71.4	**Disorders of carnitine metabolism** **Exclude 1:** Muscle carnitine palmitoyltransferase deficiency (E71.314)	
E71.41	Primary carnitine deficiency	277.81
E71.42	Carnitine deficiency due to inborn errors of metabolism Code also: associated inborn error or metabolism	277.82
E71.43	Iatrogenic carnitine deficiency Carnitine deficiency due to: hemodialysis Carnitine deficiency due to: Valproic acid therapy	277.83
E71.44	**Other secondary carnitine deficiency**	
E71.440	Ruvalcaba-Myhre-Smith syndrome	277.84
E71.448	Other secondary carnitine deficiency	277.84
E71.5	**Peroxisomal disorders** **Exclude 1:** Schilder's disease (G37.0)	
E71.50	Peroxisomal disorder, unspecified	277.86
E71.51	**Disorders of peroxisome biogenesis** **Group 1 peroxisomal disorders** **Excludes 1:** Refsum's disease (G60.1)	
E71.510	Zellweger syndrome	277.86
E71.511	Neonatal adrenoleukodystrophy **Excludes 1:** X-linked adrenoleukodystrophy (E71.42-) E71.511 is only applicable to newborns of age 0 years.	277.86
E71.518	Other disorders of peroxisome biogenesis	277.86
E71.52	**X-linked adrenoleukodystrophy**	
E71.520	Childhood cerebral X-linked adrenoleukodystrophy	277.86
E71.521	Adolescent X-linked adrenoleukodystrophy	277.86
E71.522	Adrenomyeloneuropathy	277.86
E71.528	Other X-linked adrenoleukodystrophy Addison only phenotype adrenoleukodystrophy Addison-Schilder adrenoleukodystrophy	277.86
E71.529	X-linked adrenoleukodystrophy, unspecified type	277.86
E71.53	Other group 2 peroxisomal disorders	277.86
E71.54	**Other peroxisomal disorders**	
E71.540	Rhizomelic chondrodysplasia punctate **Excludes 1:** chondrodysplasia punctata NOS (Q77.3)	277.86
E71.541	Zellweger-like syndrome	277.86
E71.542	Other group 3 peroxisomal disorders	277.86
E71.548	Other peroxisomal disorders	277.86
E72	**Other disorders of amino-acid metabolism** **Excludes 1:** disorders of:aromatic amino-acid metabolism (E70.-), branched-chain amino-acid metabolism (E71.0-E71.2), fatty-acid metabolism (E71.3), purine and pyrimidine metabolism (E79.-), gout (M1A.-, M10.-)	
E72.0	**Disorders of amino-acid transport** **Excludes 1:** disorders of tryptophan metabolism (E70.5)	
E72.00	Disturbances of amino acid transport, unspecified	270.0
E72.01	Cystinuria	270.0
E72.02	Hartnup's disease	270.0
E72.03	Lowe's syndrome	270.8

	Use additional code for associated glaucoma(H42)	
E72.04	Cystinosis Fanconi (-de Toni) (-Debré) syndrome with cystinosis **Excludes 1:** Fanconi (-de Toni) (-Debré) syndrome without cystinosis (E72.09)	270.0
E72.09	Other disorders of amino-acid transport Fanconi (-de Toni) (-Debré) syndrome, unspecified	270.0
E72.1	**Disorders of sulfur-bearing amino-acid metabolism** **Excludes 1:** cystinosis (E72.04), cystinuria (E72.01), transcobalamin II deficiency (D51.2)	
E72.10	Disorders of sulphur-bearing amino-acid metabolism, unspecified	270.4
E72.11	Homocystinuria Cystathionine synthase deficiency	270.4
E72.12	Methylenetetrahydrofolate reductase (MTHFR) deficiency	270.4
E72.19	Other disorders of sulfur-bearing amino-acid metabolism • Cystathioninuria • Methioninemia • Sulfite oxidase deficiency	270.4
E72.2	**Disorders of urea cycle metabolism** **Excludes 1:** disorders of ornithine metabolism (E72.4)	
E72.20	Disorders of urea cycle metabolism, unspecified Hyperammonemia **Excludes 1:** hyperammonemia-hyperornithinemia-homocitrullinemia syndrome E72.4, transient hyperammonemia of newborn (P74.6)	270.6
E72.21	Argininemia	270.6
E72.22	Arginosuccinic aciduria	270.6
E72.23	Citrullinemia	270.6
E72.29	Other disorders of urea cycle metabolism	270.6
E72.3	Disorders of lysine and hydroxylysine metabolism • Glutaric aciduria NOS • Glutaric aciduria (type I) • Hydroxylysinemia • Hyperlysinemia **Excludes 1:** glutaric aciduria type II (E71.313), Refsum's disease (G60.1), Zellweger syndrome (E71.510)	270.7
E72.4	Disorders of ornithine metabolism • Hyperammonemia-Hyperornithinemia-Homocitrullinemia syndrome • Ornithinemia (types I, II) • Ornithine transcarbamylase deficiency **Excludes 1:** hereditary choroidal dystrophy (H31.2-)	270.6
E72.5	Disorders of glycine metabolism	
E72.50	Disorder of glycine metabolism, unspecified	270.7
E72.51	Non-ketotic hyperglycinemia	270.7
E72.52	Trimethylaminuria	271.8
E72.53	Hyperoxaluria • Oxalosis • Oxaluria	271.8
E72.59	Other disorders of glycine metabolism • D-glycericacidemia • Hyperhydroxyprolinemia • Hyperprolinemia (types I, II) • Sarcosinemia	270.7
E72.8	Other specified disorders of amino-acid metabolism • Disorders of beta-amino-acid metabolism • Disorders of gamma-glutamyl cycle	270.8
E72.9	Disorder of amino-acid metabolism, unspecified	270.9
E74	**Other disorders of carbohydrate metabolism**	

	Excludes 1: diabetes mellitus (E08-E13), hypoglycemia NOS (E16.2), increased secretion of glucagon (E16.3), mucopolysaccharidosis (E76.0-E76.3)	
E74.0	**Glycogen storage disease**	
E74.00	Glycogen storage disease, unspecified	271.0
E74.01	von Gierke disease Type I glycogen storage disease	271.0
E74.02	Pompe disease Cardiac glycogenosis Type II glycogen storage disease	271.0
E74.03	Cori disease Forbes disease Type III glycogen storage disease	271.0
E74.04	McArdle disease Type V glycogen storage disease	271.0
E74.09	Other glycogen storage disease • Andersen disease • Hers disease • Tauri disease • Glycogen storage disease, types 0, IV, VI-XI • Liver phosphorylase deficiency • Muscle phosphofructokinase deficiency	271.0
E74.2	**Disorders of galactose metabolism**	
E74.20	Disorders of galactosetabolism, unspecified	271.1
E74.21	Galactosemia	271.1
E74.29	Other disorders of galactose metabolism Galactokinase deficiency	271.1
E74.4	Disorders of pyruvate metabolism and gluconeogenesis • Deficiency of phosphoenolpyruvate carboxykinase • Deficiency of pyruvate carboxylase • Deficiency of pyruvate dehydrogenase **Excludes 1:** disorders of pyruvate metabolism and gluconeogenesis with anemia (D55.-), Leigh's syndrome (G31.82)	271.0 271.8
E75	**Disorders of sphingolipid metabolism and other lipid storage disorders** **Excludes 1:** mucolipidosis, types I-III (E77.0-E77.1), Refsum's disease (G60.1)	
E75.0	**GM2 gangliosidosis**	
E75.00	GM2 gangliosidosis, unspecified	330.1
E75.01	Sandhoff disease	330.1
E75.02	Tay-Sachs disease	330.1
E75.09	Other GM2 gangliosidosis Adult GM2 gangliosidosis – **do not report** Juvenile GM2 gangliosidosis	330.1
E75.1	**Other and unspecified gangliosidosis**	
E75.10	Unspecified gangliosidosis Gangliosidosis NOS	330.1
E75.11	Mucolipidosis IV	330.1
E75.19	Other gangliosidosis GM1 gangliosidosis GM3 gangliosidosis	330.1
E75.2	**Other sphingolipidosis** **Excludes 1:** adrenoleukodystrophy [Addison-Schilder] (E71.528)	
E75.21	Fabry (-Anderson) disease	272.7
E75.22	Gaucher disease	272.7
E75.23	Krabbe disease	330.0
E75.24	**Niemann-Pick disease**	
E75.240	Niemann-Pick disease type A	272.7

E75.241	Niemann-Pick disease type B	272.7
E75.25	Metachromatic Leukodystrophy	330.0
E75.29	Other sphingolipidosis Farber's syndrome Sulfatase deficiency Sulfatide lipidosis	330.0
E75.4	Neuronal ceroid lipofuscinosis • Batten disease • Bielschowsky-Jansky disease • Kufs disease • Spielmeyer-Vogt disease	330.1
E75.5	Other lipid storage disorders • Cerebrotendinous cholesterosis [van Bogaert-Scherer-Epstein] • Wolman's disease	272.8
E76	**Disorders of glycosaminoglycan metabolism**	
E76.0	**Mucopolysaccharidosis, type I**	
E76.01	Hurler's syndrome Lipochondrodystrophy	277.5
E76.02	Hurler-Scheie syndrome	277.5
E76.03	Scheie's syndrome	277.5
E76.1	Mucopolysaccharidosis, type II Hunter's syndrome	277.5
E76.2	**Other mucopolysaccharidoses**	
E76.21	**Morquio mucopolysaccharidoses**	
E76.210	Morquio A mucopolysaccharidoses • Classic Morquio syndrome • Morquio syndrome A • Mucopolysaccharidosis, type IVA	277.5
E76.211	Morquio B mucopolysaccharidoses • Morquio-like mucopolysaccharidoses • Morquio-like syndrome • Morquio syndrome B • Mucopolysaccharidosis, type IVB	277.5
E76.219	Morquio mucopolysaccharidoses, unspecified • Morquio syndrome • Mucopolysaccharidosis, type IV	277.5
E76.22	Sanfilippo mucopolysaccharidoses • Mucopolysaccharidosis, type III (A) (B) (C) (D) • Sanfilippo A syndrome • Sanfilippo B syndrome • Sanfilippo C syndrome • Sanfilippo D syndrome	277.5
E76.29	Other mucopolysaccharidoses • Beta-Glucuronidase deficiency • Maroteaux-Lamy (mild) (severe) syndrome • Mucopolysaccharidosis, types VI, VII	277.5
E76.3	Mucopolysaccharidosis, unspecified	277.5
E76.8	Other disorders of glucosaminoglycan metabolism	277.5
E76.9	Glucosaminoglycan metabolism disorder, unspecified	277.5
E77	**Disorders of glycoprotein metabolism**	
E77.0	Defects in post-translational modification of lysosomal enzymes • Mucolipidosis II [I-cell disease] • Mucolipidosis III [pseudo-Hurler polydystrophy]	272.7
E77.1	Defects in glycoprotein degradation • Aspartylglucosaminuria	271.8 272.7

	• Fucosidosis • Mannosidosis • Sialidosis [mucolipidosis I]	
E77.8	Other disorders of glycoprotein metabolism	272.7
E77.9	Disorder of glycoprotein metabolism, unspecified	272.7
E78	Disorders of lipoprotein metabolism and other lipidemias **Excludes 1:** sphingolipidosis (E75.0-E75.3)	
E78.71	Barth syndrome	759.89
E78.72	Smith-Lemli-Opitz syndrome	759.89
E79	**Disorders of purine and pyrimidine metabolism** **Excludes 1:** Ataxia-telangiectasia (Q87.1), Bloom's syndrome (Q82.8), Cockayne's syndrome (Q87.1), calculus of kidney (N20.0), combined immunodeficiency disorders (D81.-), Fanconi's anemia (D61.09), gout (M1A.-, M10.-), orotaciduric anemia (D53.0), progeria (E34.8), Werner's syndrome (E34.8), and xeroderma pigmentosum (Q82.1)	
E79.1	Lesch-Nyhan syndrome HGPRT deficiency	277.2
E79.2	Myoadenylate deaminase deficiency	277.2
E79.8	Other disorders of purine and pyrimidine metabolism Hereditary xanthinuria	277.2
E79.9	Disorder of purine and pyrimidine metabolism, unspecified	277.2
E80	**Disorders of porphyrin and bilirubin metabolism** **Includes:** defects of catalase and peroxidase	
E80.0	Hereditary erythropoietic porphyria Congenital erythropoietic porphyria Erythropoietic protoporphyria	277.1
E80.1	Porphyria cutanea tarda	277.1
E80.2	Other and unspecified porphyria	
E80.20	Unspecified porphyria Porphyria NOS	277.1
E80.21	Acute intermittent (hepatic) porphyria	277.1
E80.29	Other porphyria Hereditary coproporphyria	277.1
E80.3	Defects of catalase and peroxidase Acatalasia [Takahara]	277.89
E80.4	Gilbert syndrome	277.4
E80.5	Crigler-Najjar syndrome	277.4
E80.6	Other disorders of bilirubin metabolism Dubin-Johnson syndrome Rotor's syndrome	277.4
E80.7	Disorder of bilirubin metabolism, unspecified	277.4
E83	**Disorders of mineral metabolism** **Excludes 1:** dietary mineral deficiency (E58-E61), parathyroid disorders (E20-E21), vitamin D deficiency (E55.-)	
E83.00	Disorder of copper metabolism, unspecified	275.1
E83.01	Wilson's disease Code also associated Kayser Fleischer ring (H18.04-)	275.1
E83.09	Other disorders of copper metabolism	275.1
E83.1	**Disorders of iron metabolism** **Excludes 1:** iron deficiency anemia (D50.-), sideroblastic anemia (D64.0-D64.3)	
E83.10	Disorder of iron metabolism, unspecified	275.09
E83.2	Disorders of zinc metabolism Acrodermatitis enteropathica	686.8
E83.3	Disorders of phosphorus metabolism and phosphatases Do not report if less than 1500 grams **Excludes 1:** adult osteomalacia (M83.-), osteoporosis (M80.-)	

E83.30	Disorder of phosphorus metabolism, unspecified	275.3
E83.31	Familial hypophosphatemia • Vitamin D-resistant osteomalacia • Vitamin D-resistant rickets **Excludes 1:** vitamin D-deficiency rickets (E55.0)	275.3
E83.32	Hereditary vitamin D-dependent rickets (type 1) (type 2) • 25-hydroxyvitamin D 1-alpha-hydroxylase deficiency • Pseudovitamin D deficiency • Vitamin D receptor defect	275.3
E84	**Cystic fibrosis** **Includes:** mucoviscidosis	
E84.0	Cystic fibrosis with pulmonary manifestations Use additional code to identify any infectious organism present, such as: Pseudomonas (B96.5)	277.02
E84.1	**Cystic fibrosis with intestinal manifestations**	
E84.11	Meconium ileus in cystic fibrosis **Excludes 1:** meconium ileus not due to cystic fibrosis (P76.0) E84.11 is only applicable to newborns of age 0 years.	277.01
E84.19	Cystic fibrosis with other intestinal manifestations Distal intestinal obstruction syndrome	277.03
E84.8	Cystic fibrosis with other manifestations	277.09
E84.9	Cystic fibrosis, unspecified	277
E85	**Amyloidosis** **Excludes 1:** Alzheimer's disease (G30.0-)	
E85.0	Non-neuropathic heredofamilial amyloidosis	277.31
E85.1	Neuropathic heredofamilial amyloidosis Amyloid polyneuropathy (Portuguese)	277.39
E85.2	Heredofamilial amyloidosis, unspecified	277.39
E88	**Other and unspecified metabolic disorders** Use Additional codes for associated conditions **Excludes 1:** histiocytosis X (chronic) (C96.6)	
E88.0	Disorders of plasma-protein metabolism, not elsewhere classified **Excludes 1:** disorder of lipoprotein metabolism (E78.-), monoclonal gammopathy (of undetermined significance) (D47.2), polyclonal hypergammaglobulinemia (D89.0), Waldenström macroglobulinemia (C88.0)	
E88.01	Alpha-1-antitrypsin deficiency AAT deficiency	273.4
E88.09	Other disorders of plasma-protein metabolism, not elsewhere classified Bisalbuminemia	273.8
E88.4	**Mitochondrial metabolism disorders** **Excludes 1:** disorders of pyruvate metabolism (E74.4), Kearns-Sayre syndrome (H49.81), Leber's disease (H47.22), Leigh's encephalopathy (G31.82), Mitochondrial myopathy, NEC (G71.3), Reye's syndrome (G93.7)	
E88.40	Mitochondrial metabolism disorder, unspecified	277.87
E88.41	MELAS syndrome Mitochondrial myopathy, encephalopathy, lactic acidosis and stroke-like episodes	277.87
E88.42	MERRF syndrome Myoclonic epilepsy associated with ragged-red fibers Code Also :myoclonic epilepsy (G40.3-)	277.87
E88.49	Other mitochondrial metabolism disorders	277.87
E88.8	**Other specified metabolic disorders**	
E88.89	Other specified metabolic disorders Launois-Bensaude adenolipomatosis	277.89

	Excludes 1: adult pulmonary Langerhans cell histiocytosis (J84.82)	
G11	**Hereditary ataxia** **Excludes 2**: cerebral palsy (G80.-), hereditary and idiopathic neuropathy (G60.-), metabolic disorders (E70-E88)	
G11.0	Congenital nonprogressive ataxia	334.2
G11.1	Early-onset cerebellar ataxia Early-onset cerebellar ataxia with essential tremors Early-onset cerebellar ataxia with myoclonus [Hunt's ataxia] Early-onset cerebellar ataxia with retained tendon reflexes Friedreich's ataxia (autosomal recessive) X-linked recessive spinocerebellar ataxia	334.0 334.3
G11.3	Cerebellar ataxia with defective DNA repair Ataxia telangeictasia [Louis-Bar] **Excludes 2:** Cockayne's syndrome (Q87.1), other disorders of purine and pyrimidine metabolism (E79.-), xeroderma pigmentosum (Q82.1)	334.8
G11.4	Hereditary stastic paraplegia	334.1
G11.8	Other hereditary ataxias	334.8
G11.9	Hereditary ataxia, unspecified Hereditary cerebellar ataxia, NOS Hereditary cerebellar degeneration Hereditary cerebellar disease Hereditary cerebellar syndrome	334.9
G12	**Spinal muscular atrophy and related syndromes**	
G12.0	Infantile spinal muscular atrophy, type I [Werdnig-Hoffman]	335.0
G12.1	Other inherited spinal muscular atrophy Adult form spinal muscular atrophy Childhood form, type II spinal muscular atrophy Distal spinal muscular atrophy Juvenile form, type III spinal muscular atrophy [Kugelberg-Welander] Progressive bulbar palsy of childhood [Fazio-Londe] Scapuloperoneal form spinal muscular atrophy	335.11
G12.9	Spinal muscular atrophy, unspecified	335.10
G23	Other degenerative diseases of basal ganglia **Excludes 2:** multi-system degeneration of the autonomic nervous system (G90.3)	
G23.0	Hallervorden-Spatz disease Pigmentary pallidal degeneration	333.91
G25	**Other extrapyramidal and movement disorders** **Excludes 2:** sleep related movement disorders (G47.6-)	
G25.3	Myoclonus	333.2
G25.8	**Other specified extrapyramidal and movement disorders**	
G25.82	Stiff-man syndrome	333.91
G31	**Other degenerative diseases of nervous system, not elsewhere classified** **Use Additional** code to identify: dementia with behavioral disturbance (F02.81), dementia without behavioral disturbance (F02.80) **Excludes 2**: Reye's syndrome (G93.7)	
G31.81	Alpers disease Grey-matter degeneration	330.8
G31.82	Leigh's disease Subacute necrotizing encephalopathy	330.8
G52	**Disorders of other cranial nerves** **Excludes 2:** disorders of acoustic [8th] nerve (H93.3), disorders of optic [2nd] nerve (H46, H47.0), paralytic strabismus due to nerve palsy (H49.0-H49.2)	
G52.7	Disorders of multiple cranial nerves	352.6

	Polyneuritis cranialis Multiple cranial nerve palsies	
G60	**Hereditary and idiopathic neuropathy**	
G60.0	Hereditary motor and sensory neuropathy Charcot-Marie-Tooth disease Déjérine-Sottas disease Hereditary motor and sensory neuropathy, types I-IV Hypertrophic neuropathy of infancy Peroneal muscular atrophy (axonal type) (hypertrophic type) Roussy-Levy syndrome	356.0
G60.1	Refsum disease Infantile Refsum disease	356.3
G60.2	Neuropathy in association with hereditary ataxia	356.0
G71	**Primary disorders of muscles** **Excludes 2:** arthrogryposis multiplex congenita (Q74.3), metabolic disorders (E70-E88), myositis (M60.-)	
G71.0	Muscular dystrophy Autosomal recessive, childhood type, muscular dystrophy resembling Duchenne or Becker muscular dystrophy Benign [Becker] muscular dystrophy Benign scapuloperoneal muscular dystrophy with early contractures [Emery-Dreifuss] Congenital muscular dystrophy, NOS Congenital muscular dystrophy with specific morphological abnormalities of the muscle fiber Distal muscular dystrophy Facioscapulohumeral muscular dystrophy Limb-girdle muscular dystrophy Ocular muscular dystrophy Oculopharyngeal muscular dystrophy Scapuloperoneal muscular dystrophy Severe [Duchenne] muscular dystrophy	359.1
G71.1	**Myotonic disorders**	
G71.11	Myotonic muscular dystrophy Dystrophia myotonica [Steinert] Myotonia atrophica Myotonic dystrophy Proximal myotonic myopathy (PROMM) Steinert disease	359.21
G71.12	Myotonia congenita Acetazolamide responsive myotonia congenita Dominant myotonia congenita [Thomsen disease] Myotonia levior Recessive myotonia congenita [Becker disease]	359.22
G71.13	Myotonic chondrodystrophy Chondrodystrophic myotonia Congenital myotonic chondrodystrophy Schwartz-Jampel disease	359.23
G71.19	Other specified myotonic disorders Myotonia fluctuans Myotonia permanens Neuromyotonia [Isaacs] Isaac-Mertens syndrome Paramyotonia congenita (of von Eulenburg) Pseudomyotonia Symptomatic myotonia	359.29

G71.2	Congenital myopathies Central core disease Fiber-type disproportion Minicore disease Multicore disease Myotubular (centronuclear) myopathy Nemaline myopathy **Excludes 1**: arthrogryposis multiplex congenita (Q74.3)	359.0
G71.3	Mitochondrial myopathy, not elsewhere classified **Excludes 1**: Kearns-Sayre syndrome (H49.81), Leber's disease (H47.21), Leigh's encephalopathy (G31.82), mitochondrial metabolism disorders (E88.4.-), Reye's syndrome (G93.7)	359.89
G71.8	Other primary disorders of muscles	359.89
G71.9	Primary disorder of muscle, unspecified Hereditary myopathy, NOS	359.9
H49.8	**Other paralytic strabismus**	
H49.81	Kearns-Sayre syndrome Progressive external ophthalmoplegia with pigmentary retinopathy **Use Additional:** code for other manifestation, such as: heart block (I45.9)	
H49.811	Kearns-Sayre syndrome, right eye	277.87
H49.812	Kearns-Sayre syndrome, left eye	277.87
H49.813	Kearns-Sayre syndrome, bilateral	277.87
H49.819	Kearns-Sayre syndrome, unspecified eye	277.87
K40	**Inguinal Hernia (report for female only)** **Includes:** bubonocele, direct inguinal hernia, double inguinal hernia, indirect inguinal hernia, inguinal hernia NOS, oblique inguinal hernia, scrotal hernia	
K40.00	Bilateral inguinal hernia, with obstruction, without gangrene, not specified as recurrent Bilateral inguinal hernia, with obstruction, without gangrene, NOS	550.12
K40.1	**Bilateral inguinal hernia, with gangrene (report for female only)**	
K40.10	Bilateral inguinal hernia, with gangrene, not specified as recurrent Bilateral inguinal hernia, with gangrene, NOS	550.02
K40.2	**Bilateral inguinal hernia, without obstruction or gangrene (female only)**	
K40.20	Bilateral inguinal hernia, without obstruction or gangrene, not specified as recurrent Bilateral inguinal hernia, NOS	550.92
K40.3	**Unilateral inguinal hernia, with obstruction, without gangrene (female only)** • Inguinal hernia (unilateral) causing obstruction without gangrene • Incarcerated inguinal hernia (unilateral) without gangrene • Irreducible inguinal hernia (unilateral) without gangrene • Strangulated inguinal hernia (unilateral) without gangrene Indicate right, left, or unspecified	
K40.30	Unilateral inguinal hernia, with obstruction, without gangrene, not specified as recurrent Inguinal hernia, with obstruction, NOS Unilateral inguinal hernia, with obstruction, without gangrene, NOS	550.1
K40.40	Unilateral inguinal hernia, with gangrene, not specified as recurrent Inguinal hernia with gangrene, NOS Unilateral inguinal hernia with gangrene, NOS	550
K40.9	**Unilateral inguinal hernia, without obstruction or gangrene (female only)** Indicate right, left, or unspecified	
K40.90	Unilateral inguinal hernia, without obstruction or gangrene, not specified as recurrent Inguinal hernia, NOS	550.9
K41	**Femoral hernia (female only)**	
K41.0	**Bilateral femoral hernia, with obstruction, without gangrene (female only)** • Femoral hernia (bilateral) causing obstruction, without gangrene	

	• Incarcerated femoral hernia (bilateral), without gangrene • Irreducible femoral hernia (bilateral), without gangrene • Strangulated femoral hernia (bilateral), without gangrene	
K41.00	Bilateral femoral hernia, with obstruction, without gangrene, not specified as recurrent Bilateral femoral hernia, with obstruction, without gangrene, NOS	552.02
K41.1	**Bilateral femoral hernia, with gangrene (female only)**	
K41.10	Bilateral femoral hernia, with gangrene, not specified as recurrent Bilateral femoral hernia, with gangrene, NOS	551.02
K41.2	**Bilateral femoral hernia, without obstruction or gangrene (female only)**	
K41.20	Bilateral femoral hernia, without obstruction or gangrene, not specified as recurrent Bilateral femoral hernia, NOS	553.02
K41.3	**Unilateral femoral hernia, with obstruction, without gangrene (female only)** • Femoral hernia (unilateral) causing obstruction, without gangrene • Incarcerated femoral hernia (unilateral), without gangrene • Irreducible femoral hernia (unilateral), without gangrene • Strangulated femoral hernia (unilateral), without gangrene Indicate right, left, or unspecified	
K41.30	Unilateral femoral hernia, with obstruction, without gangrene, not specified as recurrent Femoral hernia, with obstruction NOS Unilateral femoral hernia, with obstruction NOS	552
K41.4	**Unilateral femoral hernia, with gangrene (female only)** Indicate right, left, or unspecified	
K41.40	Unilateral femoral hernia, with gangrene, not specified as recurrent Femoral hernia, with gangrene NOS Unilateral femoral hernia, with gangrene NOS	551
K41.9	**Unilateral femoral hernia, without obstruction or gangrene (female only)** Indicate right, left, or unspecified	
K41.90	Unilateral femoral hernia, without obstruction or gangrene, not specified as recurrent Femoral hernia, NOS Unilateral femoral hernia, NOS	553
L05	***(m) Pilonidal cyst and sinus***	
L05.9	***(m) Pilonidal cyst and sinus without abscess***	
L05.91	*(m) Pilonidal cyst without abscess* *(m) Parasacral dimple* *(m) Sacral dimple* *(m) Pilonidal dimple* *(m) Postanal dimple* *(m) Pilonidal cyst NOS*	685.1
L05.92	*(m) Pilonidal sinus without abscess* *(m) Coccygeal fistula* *(m) Coccygeal sinus without abscess* *(m) Pilonidal fistula*	685.1
L81	***(m) Other disorders of pigmentation*** **Excludes 1:** birthmark NOS (Q82.5), Peutz-Jeghers syndrome (Q85.8) **Excludes 2:** nevus - see Alphabetical Index	
L81.3	*(m) Café au lait spots*	709.09
L81.4	*(m) Other melanin hyperpigmentation*	709.09
L81.9	*(m) Disorder of pigmentation, unspecified*	709
M26	***(m) Dentofacial anomalies [including malocclusion]***	
M26.0	***(m) Major anomalies of jaw size*** **Excludes 1**: acromegaly (E22.0), Robin's syndrome (Q87.0)	
M26.01	*(m) Maxillary hyperplasia* *(m) Prominent maxilla*	524.01
M26.02	*(m) Maxillary hypoplasia*	524.03

M26.03	*(m) Mandibular hyperplasia*	524.02
M26.04	*(m) Mandibular hypoplasia*	524.04
M26.05	*(m) Macrogenia*	524.05
M26.06	*(m) Microgenia*	524.06
M26.09	Micrognathia Macrognathia *(m) Other specified anomalies of jaw (specify)*	524.09
P02	**Newborn (suspected to be) affected by complications of placenta, cord and membranes** **Includes:** conditions that have their origin in the fetal or perinatal period (before birth through the first 28 days after birth) even if morbidity occurs later Code First any current condition in newborn	
P02.8	Newborn (suspected to be) affected by other abnormalities of membranes Amniotic band syndrome Amniotic bands (constricting bands) Streeter's (Simonart's) bands P02.8 is only applicable to newborns of age 0 years.	762.8
P04	**Newborn (suspected to be) affected by noxious substances transmitted via placenta or breast milk Includes** nonteratogenic effects of substances transmitted via placenta **Excludes 2**: congenital malformations (Q00-Q99), neonatal jaundice from excessive hemolysis due to drugs or toxins transmitted from mother (P58.4), newborn in contact with and (suspected) exposures hazardous to health not transmitted via placenta or breast milk (Z77.-)	
P04.3	Newborn (suspected to be) affected by maternal use of alcohol Report any abnormal facial features even if minor **Excludes 1:** fetal alcohol syndrome (Q86.0)	760.1
P35	**Congenital viral diseases** **Includes:** infections acquired in utero or during birth Only applicable to newborns of age 0 years.	
P35.0	Congenital Rubella Syndrome Rubella, congenital Congenital rubella pneumonitis	771.0
P35.1	Congenital cytomegalovirus infection (CMV) Cytomegalovirus infection, congenital	771.1
P35.2	Congenital herpesviral [herpes simplex] infection Herpes simplex Encephalitis herpes Meningoencephalitis herpes	771.2
P35.3	Congenital viral hepatitis	771.2
P35.8	Congenital infection, other specified Congenital varicella [chickenpox] includes congenital varicella	771.2
P35.9	Congenital viral disease, unspecified	771.2
P37	**Other congenital infectious and parasitic diseases** **Excludes 2**: congenital syphilis (A50.), infectious neonatal diarrhea (A00-A09), necrotizing enterocolitis in newborn (P77.-), noninfectious neonatal diarrhea (P78.3), ophthalmia neonatorum due to gonococcus (A54.31), tetanus neonatorum (A33)	
P37.0	Congenital tuberculosis P37.0 is only applicable to newborns of age 0 years.	771.2
P37.1	Congenital toxoplasmosis Hydrocephalus due to congenital toxoplasmosis P37.1 is only applicable to newborns of age 0 years.	771.2
P95	Stillbirth • Deadborn fetus NOS • Fetal death of unspecified cause • Stillbirth NOS **Excludes 1:** maternal care for intrauterine death (O36.4), missed abortion (O02.1), outcome of delivery, stillbirth (Z37.1, Z37.3, Z37.4, Z37.7)	**779.6**

Stillbirth Codes (O36.4, Z37)

ICD-10	Notes & Description	ICD-9
O36.4	**Maternal care for intrauterine death** • **Maternal care for intrauterine fetal death NOS** • **Maternal care for intrauterine fetal death after completion of 20 weeks of gestation** • **Maternal care for late fetal death** • **Maternal care for missed delivery** **Only applicable to female maternity patients aged 12 - 55 years inclusive.** **Should only be used on the maternal record - not on the newborn record.** **Use a code under Z3A to document the exact week during the pregnancy.** **Excludes 1:** missed abortion (O02.1), stillbirth (P95)	
O36.4XX0	Maternal care for intrauterine death, not applicable or unspecified	656.40 656.41 656.43
O36.4XX1	Maternal care for intrauterine death, fetus 1	656.41 656.43
O36.4XX2	Maternal care for intrauterine death, fetus 2	656.41 656.43
O36.4XX3	Maternal care for intrauterine death, fetus 3	656.41 656.43
O36.4XX4	Maternal care for intrauterine death, fetus 4	656.41 656.43
O36.4XX5	Maternal care for intrauterine death, fetus 5	656.41 656.43
O36.4XX9	Maternal care for intrauterine death, other fetus	656.41 656.43
Z37.1	Outcome of delivery, single stillborn • Z37.1 is only applicable to female maternity patients aged 12 - 55 years inclusive. • Z37.1 is considered unacceptable as a principal diagnosis as it describes a circumstance which influences an individual's health status but not a current illness or injury, or the diagnosis may not be a specific manifestation but may be due to an underlying cause.	V27.1
Z37.3	Outcome of delivery, twins, one liveborn and one stillborn • Z37.3 is only applicable to female maternity patients aged 12 - 55 years inclusive. • Z37.3 is considered unacceptable as a principal diagnosis as it describes a circumstance which influences an individual's health status but not a current illness or injury, or the diagnosis may not be a specific manifestation but may be due to an underlying cause.	V27.3
Z37.4	Outcome of delivery, twins, both stillborn • Z37.4 is only applicable to female maternity patients aged 12 - 55 years inclusive. • Z37.4 is considered unacceptable as a principal diagnosis as it describes a circumstance which influences an individual's health status but not a current illness or injury, or the diagnosis may not be a specific manifestation but may be due to an underlying cause.	V27.4
Z37.6	Outcome of delivery, other multiple birth, some liveborn Only applicable to female maternity patients aged 12 - 55 years inclusive. Considered unacceptable as a principal diagnosis as it describes a circumstance which influences an individual's health status but not a current illness or injury, or the diagnosis may not be a specific manifestation but may be due to an underlying cause.	V27.6
Z37.60	Multiple births, unspecified, some liveborn	
Z37.61	Triplets, some liveborn	
Z37.62	Quadruplets, some liveborn	
Z37.63	Quintuplets, some liveborn	
Z37.64	Sextuplets, some liveborn	
Z37.69	Other multiple births, some liveborn	
Z37.7	Outcome of delivery, other multiple birth, all stillborn Only applicable to female maternity patients aged 12 - 55 years inclusive.	V27.7