

Coding Of Anomalies

Anomaly	Description Of Anomaly	ICD9	ICD10	Comments
All Anomalies		74, 75, 27910, 2281, 76076, 76280, 7710, 7711, 77121	Q*, D215, D821, D1810, P350, P351, P371	Exclude all minor anomalies
Nervous system		740, 741, 742	Q00, Q01, Q02, Q03, Q04, Q05, Q06, Q07	
Neural Tube Defects	Neural tube defects include anencephalus, encephalocele, spina bifida and iniencephalus	740, 741, 7420	Q00, Q01, Q05	
Anencephalus and similar	Total or partial absence of brain tissue and the cranial vault. The face and eyes are present. (incompatible with life)	740	Q00	
Encephalocele	Cystic expansion of meninges and brain tissue outside the cranium. Covered by normal or atrophic skin.	7420	Q01	exclude if associated with anencephalus
Spina Bifida	Midline defect of the osseous spine usually affecting the posterior arches resulting in a herniation or exposure of the spinal cord and/or meninges	741	Q05	exclude if associated with anencephalus, or encephalocele
Hydrocephalus	Dilatation of ventricular system, not due to primary atrophy of the brain, with or without enlargement of the skull	7423, exclude 74232	Q03	exclude hydranencephaly (74232 icd 9 only), or association with NTDs
Microcephaly	A reduction in the size of the brain with a skull circumference less than three standard deviations below the mean for sex, age and ethnic origin. Definitions known to vary between clinicians and regions.	7421	Q02	exclude association with NTDs
Arhinencephaly/holoprosencephaly	Absence of the first cranial (olfactory) nerve tract. There is a spectrum of anomalies from a normal brain, except for the first cranial nerve tract, to a single ventricle (holoprosencephaly)	74226	Q041, Q042	
Eye		743	Q10-Q15	
Anophthalmos/microphthalmos	Anophthalmos: Unilateral or bilateral absence of the eye tissue. Clinical diagnosis; microphthalmos: Small eye/eyes with smaller than normal axial length. Clinical diagnosis	7430, 7431	Q110, Q111, Q112	
Anophthalmos	Unilateral or bilateral absence of the eye tissue. Clinical diagnosis	7430	Q110, Q111	
Congenital cataract	Alteration in the transparency of the crystalline lens	74332	Q120	
Congenital glaucoma	Large ocular globe as a result of increased ocular pressure in fetal life	74320	Q150	
Ear, face and neck		744	Q16, Q17, Q18	
Anotia	Absent pinna, with or without atresia of ear canal	74401	Q160	
Congenital heart defects		745, 746, 7470-7474	Q20-Q26	exclude PDA in preterm (<37 weeks) - ICD9: 7470; ICD10: Q250: Exclude peripheral pulmonary artery stenosis with GA < 37 weeks - Q256

Severe CHD §	Severe congenital heart defects have higher perinatal mortality and TOPFA rates. Most livebirths require surgery for survival. It includes: single ventricle, tricuspid atresia, Ebstein's anomaly, hypoplastic left heart, hypoplastic right heart, common a	74500, 74510, 7452-7453, 7456, 74600, 7461-7463, 7465-7467, 7471, 74720, 74742	Q200, Q201, Q203-Q204, Q212-Q213, Q220, Q224-Q226, Q230, Q232-Q234, Q251-Q252, Q262	
Common arterial truncus	Presence of a large single arterial vessel at the base of the heart (from which the aortic arch, pulmonary and coronary arteries originate), always accompanied by a large subvalvular septal defect.	74500	Q200	
Double outlet right ventricle §	Both aorta and the pulmonary artery connect to the right ventricle	no code	Q201	
Transposition of great vessels	Total separation of circulation with the aorta arising from the right ventricle and the pulmonary artery from the left ventricle	74510	Q203	
Single ventricle	Only one complete ventricle with an inlet valve and an outlet portion even though the outlet valve is atretic	7453	Q204	
Ventricular septal defect	Defect in the ventricular septum	7454	Q210	
Atrial septal defect	Defect in the atrial septum	7455	Q211	
Atrioventricular septal defect	Central defect of the cardiac septa and a common atrioventricular valve, includes primum ASD defects	7456	Q212	
Tetralogy of Fallot	VSD close to the aortic valves, infundibular and pulmonary valve stenosis and over-riding aorta across the VSD	7452	Q213	
Tricuspid atresia and stenosis	Obstruction of the tricuspid valve and hypoplasia of the right ventricle	7461	Q224	
Ebstein's anomaly	Tricuspid valve displaced with large right atrium and small right ventricle	7462	Q225	
Pulmonary valve stenosis	Obstruction or narrowing of the pulmonary valves which may impair blood flow through the valves	74601	Q221	
Pulmonary valve atresia	Lack of patency or failure of formation altogether of the pulmonary valve, resulting in obstruction of the blood flow from the right ventricle to the pulmonary artery	74600	Q220	
Aortic valve atresia/stenosis §	Occlusion of aortic valve or stenosis of varying degree, often associated with bicuspid valves	7463 (no code for atresia)	Q230	
Mitral valve anomalies	Atresia, stenosis or insufficiency of the mitral valve	7465, 7466	Q232, Q233	
Hypoplastic left heart	Hypoplasia of the left ventricle, outflow tract and ascending aorta resulting from an obstructive lesion of the left side of the heart	7467	Q234	
Hypoplastic right heart §	Hypoplasia of the right ventricle, always associated with other cardiac malformations	no code	Q226	
Coarctation of aorta	Constriction in the region of aorta where the ductus joins aorta	7471	Q251	
Aortic atresia/interrupted aortic arch	Atresia or interrupted connection of the aorta	74720	Q252	
Total anomalous pulm venous return	All four pulmonary veins drain to right atrium or one of the venous tributaries	74742	Q262	
PDA as only CHD in term infants (>=37 weeks)	Open duct in infancy or later and requiring invasive treatment	7470	Q250	Livebirths only
Respiratory		7480, 7484, 74850, 74852, 74858, 7486, 7488	Q300, Q32-Q34	Exclude Q336

Choanal atresia	Bony or membranous choanae with no passage from nose to pharynx	7480	Q300	
Cystic adenomatous malformation of lung §	Cystic structures of the lung, usually unilateral	no code	Q3380	
Oro-facial clefts		7490, 7491, 7492	Q35-Q37	exclude association with holoprosencephaly or anencephaly subgroups
Cleft lip with or without palate	Clefting of the upper lip with or without clefting of the maxillary alveolar process and hard and soft palate	7491, 7492	Q36-Q37	exclude association with holoprosencephaly or anencephaly subgroups
Cleft palate	Fissure defect of the soft and/or hard palate(s) or submucous cleft without cleft lip	7490	Q35	exclude association with cleft lip subgroup. Exclude association with holoprosencephaly or anencephaly subgroups
Digestive system		750, 751, 7566	Q38-Q45, Q790	
Oesophageal atresia with or without tracheo-oesophageal fistula	Occlusion or a long gap of the oesophagus with or without tracheo-oesophageal fistula	75030 - 75031	Q390-Q391	
Duodenal atresia or stenosis	Occlusion or narrowing of duodenum	75110	Q410	exclude if also annular pancreas subgroup
Atresia or stenosis of other parts of small intestine	Occlusion or narrowing of other parts of small intestine	75111-75112	Q411-Q418	
Ano-rectal atresia and stenosis	Imperforate anus or absence or narrowing of the communication canal between the rectum and anus with or without fistula to neighbouring organs	75121-75124	Q420-Q423	
Hirschsprung's disease	Absence of the parasympathetic ganglion nerve cells (aganglionosis) of the wall of the colon or rectum. May result in congenital megacolon	75130-75133	Q431	
Atresia of bile ducts	Congenital absence of the lumen of the extrahepatic bile ducts	75165	Q442	
Annular pancreas	Pancreas surrounds the duodenum causing stenosis	75172	Q451	
Diaphragmatic hernia	Defect in the diaphragm with protrusion of abdominal content into the thoracic cavity. Various degrees of lung hypoplasia on the affected side	75661	Q790	
Abdominal wall defects		75671, 75670, 75679	Q792, Q793, Q795	
Gastroschisis	Protrusion of abdominal contents through an abdominal wall defect lateral to an intact umbilical cord and not covered by a membrane	75671	Q793	
Omphalocele	Herniation of abdominal content through the umbilical ring, the contents being covered by a membrane sometimes ruptured at the time of delivery	75670	Q792	
Urinary		753, 75672, 75261	Q60-64, Q794	
Bilateral renal agenesis including Potter syndrome	Bilateral absence, agenesis, dysplasia or hypoplasia of kidneys including Potter's syndrome. Incompatible with life	75300	Q601, Q606	exclude unilateral Q600.
Multicystic renal dysplasia	Multiple, non-communicating cysts of varying size in the kidney without functional kidney tissue	75316	Q6140, Q6141	
Congenital hydronephrosis	Obstruction of the urinary flow from kidney to bladder	75320	Q620	
Bladder exstrophy and/or epispadia	Defect in the closure of the bladder and lower abdominal wall	75261, 7535	Q640, Q641	

Posterior urethral valve and/or prune belly	Urethral obstruction with dilatation of bladder and hydronephrosis. In severe cases also distended abdomen	75360, 75672	Q6420, Q794	
Genital		7520-7524, 75260, 75262, 7527-7529	Q50-Q52, Q54-Q56	
Hypospadias	The urethral meatus is abnormally located and is displaced proximally on the ventral surface of the penis	75260	Q54	
Indeterminate sex	Includes true and pseudohermaphroditism male or female	7527	Q56	
Limb		7543-7548, 755	Q65-Q74	
Limb reduction defects	Total or partial absence or severe hypoplasia of skeletal structure of the limbs	7552-7554	Q71-Q73	
Club foot - talipes equinovarus	Foot anomaly with equinus of the heel, varus of the hindfoot and adductus of the forefoot	75450	Q660	
Hip dislocation and/or dysplasia	Location of the head of the femur outside its normal position	75430	Q650-Q652, Q6580, Q6581	
Polydactyly	Extra digit or extra toe	7550	Q69	
Syndactyly	Partial or total webbing between 2 or more digits includes minor forms	7551	Q70	
Other anomalies / syndromes				
Skeletal dysplasias §	A large group of genetic diseases with developmental disorders of chondro-osseous tissue	no code	Q7402, Q77, Q7800, Q782-Q788	
Craniosynostosis	Premature closure of cranial sutures	75600	Q750	
Congenital constriction bands/amniotic band	Bands in the amniotic fluid that causes constriction of part of the brain, body or limbs, including limb-body-wall complex	76280	Q7980	
Situs inversus	Inverse position of thoracic or abdominal organs or both	7593	Q893	
Conjoined twins	Siamese twins	7594	Q894	
Congenital skin disorders	A group of mainly genetic skin disorders in the newborn	7571, 7573	Q80-Q82	
VATER/VACTERL	Association with anomalies of Vertebra, anal atresia, cardiac, trachea-esophageal fistula, esophageal atresia, radial anomaly and limb defects	759895	Q8726	
Vascular disruption anomalies §	Anomalies likely to be due to vascular disruption	no code	Q0435, Q411-Q412, Q418, Q710, Q712-Q713, Q720, Q722-Q723, Q730, Q793, Q795, Q7980, Q7982, Q8706	
Lateral anomalies §	Abnormal laterality mainly affecting heart and lungs	no code	Q206, Q240, Q3381, Q890, Q893	
Teratogenic syndromes with malformations §	Congenital anomalies in pregnancies with known teratogenic exposure	no code	Q86, P350, P351, P371	
Fetal alcohol syndrome	Fetal exposure to alcohol during pregnancy with following impact on fetal growth, facial appearance and development	76076	Q860	
Valproate syndrome §	Fetal exposure to valproate during pregnancy with impact on fetal growth, facial appearance and development. Often associated with spina bifida	no code	Q8680	
Maternal infections resulting in malformations	Specific maternal viral infections during pregnancy resulting in congenital anomalies in the fetus or infant	7710, 7711, 77121	P350, P351, P371	

Genetic syndromes + microdeletions	Clinically or genetically diagnosed syndromes with dysmorphic features or congenital anomalies with or without a microdeletion	755810, 75601, 75604, 7598, 27910 exclude 759801, 759895, 759844	Q4471, Q6190, Q7484, Q751, Q754, Q7581, Q87, Q936, D821 exclude Q8703, Q8704, Q8706, Q8708, Q8716, Q8724, Q8726	exclude associations and sequences
Chromosomal		7580-7583, 7585-7589	Q90-Q93, Q96-Q99 exclude Q936	microdeletions excluded Q936
Down Syndrome	karyotype 47,xx +21 or 47,xy +21 and translocations/mosaicism	7580	Q90	
Patau syndrome/trisomy 13	karyotype 47,xx +13 or 47,xy +13 and translocations/mosaicism	7581	Q914-Q917	
Edward syndrome/trisomy 18	karyotype 47,xx +18 or 47,xy +18 and translocations/mosaicism	7582	Q910-Q913	
Turner syndrome	karyotype 45,x or structural anomalies of X chromosome	75860, 75861, 75862, 75869	Q96	
Klinefelter syndrome	karyotype 47,xy or additional x-chromosomes	7587	Q980-Q984	
Anomalies outside normal range				