DEFINITIONS AND CODING OF ANOMALY SUBGROUPS

Anomaly	Description Of Anomaly	ICD9 codes	ICD10 codes
Nervous system	**Nervous system anomalies include neural tube defects, hydrocephaly, microcephaly and other anomalies of the brain, spinal cord and nervous system**	**740, 741, 7420-7425, 7428, 7429**	**Q00, Q01, Q02, Q03, Q04, Q05, Q06, Q07**
Neural Tube Defects	*Neural tube defects include anencephaly, encephalocele, spina bifida and iniencephacy*	*7400-7420*	*Q00, Q01, Q05*
Anencephalus and similar	Absence or deficience of a major portion of the cranial vault, the covering skin and the brain tissue. (incompatible with life)	7400-7402	Q00
Encephalocele	Herniation of the brain and/or meninges through a defect in the skull (exclude if associated with anencephalus)	7420	Q01
Spina Bifida	Midline defect of the osseous spine ususally affecting the posterior arches resulting in a herniation or exposure of the spinal cord and/or meninges (exclude if associated with anencephalus, or encephalocele)	7410-7419	Q05
Hydrocephaly	Dilation of ventricular system, not due to primary atrophy of the brain, with or without enlargement of the skull (exclude hydranencephaly, or association with NTDs)	7423, excl 74232	Q03
Microcephaly	Reduction in the size of the brain with a skull circumference less than three standard deviations below the mean for gestation or age and reduced growth during first year of life (exclude association with NTDs)	7421	Q02
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		**7430-7436, 7438-7439 (excl. 74365)**	**Q10-Q15 (excl. Q105)**
Anophthalmos/micropthalmos		*7430, 7431*	*Q110 - Q112*
Anophthalmos	Unilateral or bilateral absence of the eye tissue.	7430	Q110, Q111
Micropthalmos	Small eye/eyes with smaller than normal axial length (exclude association with anophthalmos)	7431	Q112
Cataract	Alteration in the transparency of the crystalline lens	74332	Q120
Ear		**7440-7442 (excl. 74411, 74412)**	**Q16, Q17 (excl. Q179)**
Anotia/microtia		*74401, 74421*	*Q160, Q172*
Anotia	Absent pinna, with or without atresia of ear canal	74401	Q160
Microtia	Small or deformed pinna, with or without atresia of ear canal (exclude association with anotia)	74421	Q172
Congenital heart disease	**CHD includes malformations of heart, great vessels, and endocardial fibroelastosis (exclude PDA in preterm/ LBW babies (<2,500g or <37 weeks) - ICD9: 7470; ICD10: Q250)**	**7450-7459, 7460-7469, 7470-7474**	**Q20-Q26**
Anomalies of cardiac chambers and connections		*74500, 7451, 7453, 7457*	*Q20*
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 subvulvar septal defect.	74500	Q200
Transposition of great vessels (complete)	Total separation of circulation with the aorta arising from the right ventricle and the pulmonary artery from the left ventricle	74510	Q203

Anomaly	Description Of Anomaly	ICD9 codes	ICD10 codes
Single ventricle	Only one complete ventricle with an inlet valve and an outlet portion even though the outlet valve is atretic	7453	Q204
Malformations of cardiac septa		*74501, 7452, 7454-7456, 7458-7459*	*Q21*
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 acroos the VSD	7452	Q213
Malformations of valves		*7460-7467*	*Q22-Q23*
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
Aortic valve atresia/stenosis	Occlusion of aortic valve or stenosis of varying degree, often associated with bicuspid valves	7463	Q230
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
Malformations of the great arteries and veins	*exclude PDA in preterm/LBW babies (<2,500g or <37 weeks) - ICD9: 7470; ICD10: Q250*	*7470, 7471, 7472, 7473, 74742, 74743*	*Q25-Q26*
Coarctation of aorta	Constriction in the region of aorta where the ductus joins aorta	7471	Q251
Orofacial Clefts		**7490-7492**	**Q35-Q36**
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
Cleft palate	Fissure defect of the soft and/or hard palate(s) or submucous cleft without cleft lip (exclude association with cleft lip (7491-7492, Q36-Q37)	7490	Q35
Digestive system	**Includes tracheo-oesophagal fistula, oesophageal atresia and stenosis, atresia and stenosis of rectum and anal canal, atresia and stenosis of small intestine, Meckel's diverticulum, colon disorders, anomalies of intestinal fixation and anomalies of gallbladder, bile ducts and liver (exclude pyloric stenosis)**	**7503-7504, 7507-7519**	**Q39, Q402, Q403, Q408, Q409, Q41, Q42, Q43, Q44, Q45**
Tracheo-oesophagal fistula-Oesophageal atresia and stenosis	Occlusion or narrowing of the oesophagus with or without tracheo-oesophagael fistula	7503	Q390-Q394
Congenital absence, atresia and/or stenosis of the small intestine	Occlusion or narrowing of small intestine	7511	Q41
Congenital absence, atresia and/or stenosis of the duodenum	Occlusion or narrowing of duodenum	75110	Q410
Congenital absence, atresia and/or stenosis of other specified parts of small intestine	Occlusion or narrowing of jejumum or ileum	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, Q421, Q422, Q423

Anomaly	Description Of Anomaly	ICD9 codes	ICD10 codes
Internal urogenital system-ovaries uterus and renal system		**7520-7523, 7529, 7530, 7531, 7532, 7533, 7534-7539**	**Q50, Q510-Q514, Q517-Q519, Q60, Q61, Q62, Q63, Q641-Q649**
Bilateral renal agenesis	Bilateral absence, agensis, dysplasis or hypoplasia of kidneys including Potter's syndrome. Incompatible with life (exclude unilateral. exclude unspecified: Q602, Q605)	75300	Q601, Q604, Q606
Cystic kidney disease	Presence of single or multiple cyst(s) enlarging kidney tissue	7531	Q61
Congenital hydronephrosis	Obstruction of the urinary flow from kidney to bladder	75320	Q620
Bladder exstrophy	Defect in the closure of the bladder and lower abdominal wall	7535	Q641
External genital system	**Includes hypospadias, epispadias, indeterminate sex, and other anomalies such as absence of testis, aplasia or hypoplasia of scrotum, penis (exclude 7525, Q53)**	**7524, 7525, 7526, 7527, 7528**	**Q515, Q516, Q52, Q53, Q54, Q55, Q56, Q640**
Hypospadias	Agenesis of the distal urethra and opening of the urethra on the ventral side of the penis behind the coronary sulcus (exclude chordee: Q544; exclude glandular hypospadias Q540)	75260	Q541-Q543, Q548, Q549
Indeterminate sex	Includes true and pseudohermaphroditism male or female	7527	Q56
Limb	**Limb anomalies include limb reduction, polydactyly, syndactyly, congenital dislocation of the hip and other limb anomalies (exclude 75432, 75452, 75460, 75473, 75560, Q662, Q664, Q668)**	**7543-7544, 7545-7547, 7550-7551, 7552-7554, 7555-7556, 7558-7559**	**Q650-Q656, Q66, Q682-Q685, Q69, Q70, Q71, Q72, Q73, Q74**
Limb reduction	*Total or partial absence or severe hypoplasia of skeletal structure of the limbs*	*7552-7554*	*Q71-Q73*
Upper limb reduction		7552	Q71
Complete absence of upper limb		75520	Q710
Absence of upper arm and forearm with hand present		75521	Q711
Absence of both forearm and hand		75523	Q712
Absence of hand and fingers		75524	Q713
Longitudinal reduction defect/shortening of arm		75525-75527	Q714-Q718
Lower limb reduction		7553	Q72
Complete absence of lower limb		75530	Q720
Absence of thigh and lower leg with foot present		75531	Q721
Absence of both lower leg and foot		75533	Q722
Absence of foot and toe		75534	Q723
Longitudinal reduction defect/shortening of leg		75535, 75536	Q724-Q728
Polydactyly	Extra digit or extra toe	7550	Q69
Syndactyly	Partial or total webbing between 2 or more digits includes minor forms	7551	Q70

Anomaly	Description Of Anomaly	ICD9 codes	ICD10 codes
Musculoskeletal and connective tissue	**A heterogeneous group of anomalies including anomalies of diaphragm, abdominal wall, tongue, branchial cleft, auricular sinus, nose, face, skull, neck, thorax, bones, muscles and cartilages (exclude 74491, 74819, 75481,75610, 75636, Q189, Q309, Q676, Q760, Q767)**	**7444-7445, 7448-7449, 7480-7481, 7501-7502, 7540-7542, 7548, 7560-7568, 7569, 5240, 5249**	**Q18, Q30, Q380, Q382, Q389, Q67, Q680, Q688, Q75-Q79, Q8704, Q8705, Q8708,Q870A, K070, K079**
Choanal atresia	Bony or membraneous choanae with no passage from nose to pharynx	7480	Q300
Craniosynostosis	Premature closure of cranial sutures	75600	Q750
Pierre Robin Syndrome	Micrognatia, glossoptosis and often cleft palate	75603	Q8708
Mandibulofacial dystosis (Treacher-Collins and Franceschetti)	Malar and mandibular hypoplasia, malformation of ear, often cleft palate	75604	Q754, Q870A
Goldenhar's Syndrome	Facial, auriculo and vertebral malformations,usually unilateral	75606	Q8704
Chondrodystrophies and osteodystrophies	Heterogeneous group of dwarfism and other skeletal syndromes	7564, 7565	Q77, Q78
Diaphragmatic hernia	Defect in the diaphragm with protrusion of abdominal content into the thoracic cavity. Various degree of lung hypoplasia on the affected side	75661	Q790
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
Gastroschisis	Protrusion of abdominal contents through an abdominal wall defect lateral to an intact umbilical cord and not covered by a membrane	75671	Q793
Prune Belly Syndrome	Syndrome with deficient abdominal muscle and urinary obstruction/distension. May be secondary to urethral obstruction.	75672	Q794
Chromosomal	**(exclude balanced translocations: 7584, Q95)**	**7580-7583, 7585-7589**	**Q90-Q94, Q96-Q99**
Down Syndrome (trisomy 21)	karyotype 47xx +21 or 47xy +21	7580	Q90
Patau syndrome (trisomy 13)	karyotype 47xx +13 or 47xy +13	7581	Q914-Q917
Edward syndrome (trisomy 18)	karyotype 47xx +18 or 47xy +18	7582	Q910-Q913
Other trisomies and partial trisomies of autosomes		7585	Q92
Monosomies and deletions from the autosomes		7583	Q93
Turner's syndrome	karyotype 45x	75860, 75861, 75862, 75869	Q96
Klinefelters syndrome	karyotype 47xxy	7587	Q980-Q984
Anomalies outside normal range	**Relevant anomalies not coded within the range 740 to 759 of ICD 9 (International Classification of Disease, 9th edition, WHO Geneva 1992) or the Q chapter of ICD 10 (10th edition, WHO Geneva 1992)**		

Definitions and codes follow EUROCAT guidelines: http://www.eurocat.ulst.ac.uk