3.3 EUROCAT Subgroups of Congenital Anomalies

(Version 2022; implemented in DMS in May 2022, used for website prevalence tables from June 2022).

In EUROCAT, congenital anomalies are grouped together based on shared clinical characteristics and etiology. Only cases with major congenital anomalies, defined as structural changes that have significant medical, social or cosmetic consequences for the affected individual, are included in the subgroups. Cases with only minor anomalies are excluded (see Chapter 3.2).

The EUROCAT subgroups of congenital anomalies were updated in May 2022, based on advances in genetic diagnostic techniques, review of the literature, data extractions from the JRC-EUROCAT Central Database and expertise from the EUROCAT Coding and Classification Committee.

Guide 1.5 is used to code all cases of congenital anomaly and uses ICD10-BPA codes only. The ICD9-BPA codes and the minor anomalies pre-2005 are provided for retrospectively making subgroups pre-2005 when this coding system was used. New minor codes are added in green.

The new classification presented here is in use for the prevalence tables since June 2022 and applies to all individual cases present in the JRC-EUROCAT Central Database.

EUROCAT Subgroups	ICD10-BPA	ICD9-BPA	Comments	Excluded minor anomalies post-2005	Excluded minor anomalies pre-2005	Subgroup binary variable number (al)
All anomalies *$	Q-chapter, D215, D821, D1810^, P350, P351, P354, P358, P371	74, 75, 2377, 27910, 2281^, 7607, 7608, 76280, 7710, 7711, 77121	No ICD9-BPA code for sacral teratoma Q code of major malformation must be present for P350, P351, P354, P358, P371, Q86, Q7980, 7607, 7608, 76280, 7710, 7711, 77121	Exclude all minor anomalies as specified in Guide 1.5, section 3.2	Exclude all minor anomalies as specified in Guide 1.2 (ICD9 and ICD10)	al1
Nervous system anomalies $	Q00, Q01, Q02, Q03, Q04, Q05, Q06, Q07, Q8703	740, 741, 742, 759801		Q0461, Q0780, Q0782		al2
Neural Tube Defects	Q00, Q01, Q05	740, 741, 7420				al3
Anencephaly and similar	Q00	740				al4
Encephalocele and meningocele	Q01	7420	Exclude if associated with anencephaly subgroup			al5
Spina Bifida	Q05	741	Exclude if associated with anencephaly or encephalocele subgroups			al6
Hydrocephaly	Q03	7423	Exclude hydranencephaly 74232. Exclude if associated with NTD subgroup			al7
Severe microcephaly	Q02	7421	Exclude if associated with NTD subgroup			al8
Arhinencephaly / holoprosencephaly $	Q041, Q042, Q8703	74226, 759801	Exclude if associated with NTD subgroup			al9
Agenesis of corpus callosum $	Q0400	No code	Exclude if associated with NTD subgroup			al115
Eye anomalies	Q10-Q15	743		Q101-Q103, Q105, Q135	74365	al10
Anophthalmos / microphthalmos	Q110, Q111, Q112	7430, 7431				al11
Anophthalmos	Q110, Q111	7430				al12

EUROCAT Subgroups	ICD10-BPA	ICD9-BPA	Comments	Excluded minor anomalies post-2005	Excluded minor anomalies pre-2005	Subgroup binary variable number (al)
Congenital cataract	Q120	74332				al13
Congenital glaucoma	Q150	74320				al14
Ear, face and neck anomalies	Q16, Q17, Q18	744		Q170-Q175, Q179, Q180-Q182, Q184-Q187, Q1880, Q189	74411, 74412, 7443, 74491	al15
Anotia and atresia / stenosis / stricture of external auditory canal$	Q160, Q161	74400, 74401				al16
Congenital Heart Defects	Q20-Q26	745, 746, 7470-7474	Exclude PDA with GA <37 weeks Exclude peripheral pulmonary artery stenosis with GA < 37 weeks	Q2111,Q246, Q250 if GA <37 weeks, Q2541, Q256 if GA<37 weeks, Q261	Q250, 7470 if GA <37 weeks **	al17
Severe congenital heart defects~$	Q200, Q201, Q202, Q203, Q204, Q205, Q206, Q212, Q213, Q214, Q2182, Q220, Q224, Q225, Q226, *Q230, Q232,* Q234, Q242, *Q244, Q245*, Q251, Q252, *Q253*, Q262, Q263	7450, 7451, 7452, 7453, 7456, 7461, 7462, 74600, *7463*, *7465*, 7467, 74682, 7471, 74720, *74722*, 74742, 74743	No ICD9-BPA code for double outlet right ventricle, double outlet left ventricle, hypoplastic right heart, Ivemark atrial isomerism, subaortic valve stenosis and malformations of coronary arteries			al97
Common arterial truncus	Q200	74500				al18
Double outlet right ventricle	Q201	No code				al109
Double outlet left ventricle $	Q202	No code				al116
Complete transposition of great arteries (D-TGA)	Q203	74510				al19
Single ventricle	Q204	7453				al20
Corrected transposition of great arteries (L-TGA) $	Q205	74512				al117
VSD	Q210	7454				al21
ASD	Q211	7455	ICD10-BPA code includes common atrium	Q2111		al22
AVSD	Q212	7456	ICD9-BPA code includes common atrium			al23
Tetralogy and pentalogy of Fallot $	Q213, Q2182	7452				al24
Triscuspid atresia and stenosis	Q224	7461				al25
Ebstein's anomaly	Q225	7462				al26
Pulmonary valve stenosis	Q221	74601				al27
Pulmonary valve atresia	Q220	74600				al28
Aortic valve atresia/stenosis	Q230	7463	No ICD9-BPA code for aortic valve atresia			al29
Mitral valve atresia/ stenosis $	Q232	7465	No ICD9-BPA code for mitral valve atresia			al110
Hypoplastic left heart	Q234	7467				al30

EUROCAT Subgroups	ICD10-BPA	ICD9-BPA	Comments	Excluded minor anomalies post-2005	Excluded minor anomalies pre-2005	Subgroup binary variable number (al)
(HLH/HLHS)						
Hypoplastic right heart (HRH/HRHS)	Q226	No code				al31
Coarctation of aorta	Q251	7471				al32
Aortic atresia / interrupted aortic arch	Q252	74720				al111
Total anomalous pulmonary venous return	Q262	74742				al33
PDA as **only** CHD in term infants (GA +37 weeks)	Q250	7470	Live births only			al100
Respiratory anomalies $	Q300, Q32-Q34	7480, 74833-74835, 7484, 74850, 74852, 74858, 7486, 7488	Exclude lung hypoplasia Q336	Q320, Q322, Q3300, Q331	Q309, 74819	al34
Choanal stenosis or atresia	Q300	7480				al35
Congenital pulmonary airway malformations (CPAM)	Q3380	No code				al36
Oro-facial clefts $	Q35-Q37	7490, 7491, 7492	Exclude if associated with holoprosencephaly subgroup	Q357		al101
Cleft lip with or without cleft palate $	Q36, Q37	7491, 7492	Exclude if associated with holoprosencephaly subgroup			al102
Cleft palate $	Q35	7490	Exclude if associated with cleft lip subgroup. Exclude if associated with holoprosencephaly subgroup	Q357		al103
Gastro-intestinal anomalies $	Q38-Q45, Q790	750, 751, 75661		Q381, Q382, Q3850, Q400, Q401, Q4021, Q430, Q4320, Q4381, Q4382, Q444, Q4583	Q381, Q401, 7500, 7506	al40
Oesophageal atresia with or without tracheo-oesophageal fistula	Q390-Q391	75030-75031				al41
Duodenal atresia or stenosis	Q410	75110	Exclude if associated with annular pancreas subgroup			al42
Atresia or stenosis of other parts of small intestine $	Q411-Q418	75111-75112	Exclude if associated with gastroschisis or omphalocele subgroup			al43
Ano-rectal atresia or stenosis	Q420-Q423	75121-75124				al44
Hirschsprung's disease	Q431	75130-75133				al45
Atresia of bile ducts	Q442	75165				al46
Annular pancreas	Q451	75172				al47
Anomalies of intestinal fixation $	Q433	7514	Exclude if associated with gastroschisis or omphalocele subgroup			al119
Diaphragmatic hernia	Q790	75661				al48

EUROCAT Subgroups	ICD10-BPA	ICD9-BPA	Comments	Excluded minor anomalies post-2005	Excluded minor anomalies pre-2005	Subgroup binary variable number (al)
Abdominal wall defects	Q792, Q793, Q795	75671, 75670, 75679				al49
Gastroschisis	Q793	75671				al50
Omphalocele	Q792	75670				al51
Congenital anomalies of kidney and urinary tract (CAKUT)	Q60-Q64, Q794	75261, 753, 75672		Q610, Q627, Q633		al52
Unilateral renal agenesis $	Q600	75301	ICD9-BPA code includes unilateral hypoplasia and dysplasia of kidney			al120
Bilateral renal agenesis including Potter sequence	Q601, Q606	75300	ICD9-BPA code includes bilateral hypoplasia and dysplasia of kidney			al53
Multicystic renal dysplasia	Q6140, Q6141	75316				al54
Congenital hydronephrosis including ureter obstruction $	Q620, Q621, Q623	75320, 75321, 75329	Exclude if associated with vesico-uretero-renal reflux (VUR, Q627) Exclude if associated with NTD subgroup			al55
Lobulated, fused and horseshoe kidney and ectopic kidney $	Q631, Q632	75332, 75333				al121
Bladder exstrophy and / or epispadias	Q640, Q641	75261, 7535				al56
Posterior urethral valves $	Q6420	75360				al57
Prune belly syndrome$	Q794	75672				al122
Genital anomalies $	Q50-Q52, Q54-Q56	7520-7524, 75260, 7527-7529		Q523, Q525, Q527, Q5520, Q5521, Q501, Q502, Q505, Q544	Q540, 75260#	al58
Hypospadias	Q54	75260		Q544	Q540, 75260#	al59
Indeterminate sex	Q56	7527				al60
Limb anomalies	Q65-Q74	7543-7548, 755		Q653-Q656, Q662-Q669, Q670-Q678, Q680, Q6810, Q6821, Q683-Q685, Q7400, Q658, Q659, Q661	75432, 75452, 75460, 75473, 75481, 75560	al61
Limb reduction Defects (LRD) $	Q71-Q73	7552-7554, 75551				al62
Transverse LRD $	Q710, Q712, Q7180, Q720, Q722, Q7280, Q730	75520, 75523, 75530, 75533, 75540				al123
Longitudinal preaxial LRD $	Q7131, Q714, Q7231, Q725	75526				al124
Longitudinal postaxial LRD $	Q715, Q726					al125
Longitudinal central LRD $	Q716, Q727	75551				al126
Intercalary LRD $	Q711, Q721, Q731	75521, 75522, 75531, 75532, 75541				al127
Club foot – talipes	Q660	75450	Exclude if associated			al66

EUROCAT Subgroups	ICD10-BPA	ICD9-BPA	Comments	Excluded minor anomalies post-2005	Excluded minor anomalies pre-2005	Subgroup binary variable number (al)
equinovarus $			with NTD or bilateral renal agenesis/Potter subgroup			
Hip dislocation $	Q650-Q652	75430	Exclude if associated with NTD subgroup			al67
Polydactyly	Q69	7550				al68
Syndactyly	Q70	7551				al69
Other anomalies / syndromes						
Craniosynostosis	Q750	75600				al75
Congenital constriction bands / amniotic band sequence resulting in major malformations $	Q7980	76280	Q code of major malformation must be present			al76
Situs inversus	Q893	7593				al79
Conjoined twins	Q894	7594				al80
VATER/VACTERL association	Q8726	759895				al112
Pierre-Robin sequence $	Q8708	75603				al118
Caudal regression sequence $	Q8980	No code				al128
Sirenomelia $	Q8724	No code				al129
Septo-optic dysplasia $	Q044	74284				al130
Vascular disruption anomalies $	Q0435, Q411, Q412, Q418, Q710, Q712, Q7180, Q720, Q722, Q7280, Q730, Q793, Q7980, Q7982, Q8706	74232, 75111-75119, 75520, 75522-75524, 75530, 75532-75534, 75540, 75671, 75679, 75680, 76280	No ICD9-BPA code for Moebius, Q code of major malformation must be present for Q7980 and 76280.			al113
Laterality anomalies $	Q206, Q240, Q3381, Q890, Q893	7590, 7593	No ICD9-BPA code for isomerism of atria and broncho-pulmonary isomerism			al114
Teratogenic syndromes resulting in major malformations $	Q86, P350, P351, P354, P358, P371	7607, 7608, 7710, 7711, 77121	Q code of major malformation must be present			al82
Valproate syndrome $	Q8680	No code	Q code of major malformation must be present			al84
Maternal infections resulting in major malformations $	P350, P351, P354, P358, P371	7710, 7711, 77121	Q code of major malformation must be present			al86
Genetic disorders $ (genetic syndromes, hereditary skin disorders, skeletal dysplasias and chromosomal anomalies)	D821, Q4471, Q611, Q612, Q613, Q6190, Q7402, Q7484, Q751, Q754, Q7581, Q77, Q780-Q788, Q796, Q800-Q824, Q8282, Q8283, Q850,	2377, 27910, 75311, 75312, 75313, 75581, 75601, 75604, 7564, 75650-75658, 7571, 75730,	Exclude Associations and sequences: Q8703, Q8704, Q8706, Q8708, Q8724, Q8726, 759801, 759844, 759895			al105

EUROCAT Subgroups	ICD10-BPA	ICD9-BPA	Comments	Excluded minor anomalies post-2005	Excluded minor anomalies pre-2005	Subgroup binary variable number (al)
	Q851, Q8581, Q87, Q8934, Q90-Q93, Q96-Q99	75732-75736, 7580-7583, 7585-7589, 75934, 7595, 75961, 7598				
Skeletal dysplasias $	Q7402, Q77, Q780-Q788,	7564, 75650-75658				al104
Down syndrome / trisomy 21	Q90	7580				al89
Patau syndrome / trisomy 13	Q914-Q917	7581				al90
Edwards syndrome / trisomy 18	Q910-Q913	7582				al91
Turner syndrome $	Q96	75860, 75861, 75869				al92
Triploidy and polyploidy $	Q927	75858				al131

* All Anomalies = all cases with a major structural congenital anomaly, excluding cases with only minor anomalies as defined in Section 3.2 in Guide 1.5 for cases born post-2005. Cases with more than one anomaly are only counted once in the "All Anomalies" subgroup.

$ new subgroup created in 2022 or existing subgroup which changed in 2022

^ ICD10 code D1810 (ICD 9 code 2281) is the code for cystic hygroma

** The additional PDA exclusion (<2500 grams) listed in Guide 1.2 is not applied

~ The severe CHD subgroup was based on the paper by Dolk H, Loane M, Garne E and EUROCAT working group. (Congenital heart defects in Europe: prevalence and perinatal mortality, 2000 to 2005. Circulation. 2011; 123: 841-849). The following CHDs are included: single ventricle, hypoplastic right heart, hypoplastic left heart, tricuspid valve atresia, Ebstein anomaly, common arterial truncus, double outlet right ventricle, double outlet left ventricle, complete and corrected transposition of the great arteries, atrioventricular septal defect, tetra- and pentalogy of Fallot, pulmonary valve atresia, tricuspid valve stenosis, aortic valve atresia/stenosis, mitral valve atresia/stenosis, coarctation of aorta, aortic atresia/interrupted aortic arch, total and partial anomalous pulmonary venous return, Ivemark atrial isomerism, aortopulmonary window, cor triatriatum, subaortic valve stenosis, supravalvular aortic stenosis and malformations of the coronary arteries.

Severe CHD with codes shown in *italics* should only be included in specific studies when diagnosed in the first year of life.

The ICD9 code for hypospadias did not differentiate between the different types of hypospadias therefore minor cases of hypospadias (glandular l) were excluded at local registry level until 1 January 2005. Thereafter all types of hypospadias were included.

Version 31.05.2022