eTable 1: Congenital anomaly subgroups to use in the EUROlinkCAT Education study

Based on EUROCAT Subgroups of Congenital Anomalies (August 2016) with exclusions mentioned in doc 3.2 and doc 3.3 in Guide 1.4

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| --- | --- | --- | --- |
| EUROCAT Subgroups | ICD10-BPA | ICD9-BPA | Comments |
| All isolated structural anomalies | Q-chapter, D215, D821, D1810a, P350, P351, P371 | 74, 75, 27910, 2281a, 76076, 76280, 7710, 7711, 77121 | Excludes chromosomal, genetic and multiple anomalies  |
| Nervous System |  |  |  |
| Encephalocele | Q01 | 7420 | Exclude if associated with anencephalus subgroup |
| Spina Bifida | Q05 | 741 | Exclude if associated with anencephalus or encephalocele subgroups |
| Hydrocephalus | Q03 | 7423  | Exclude hydranencephaly 74232. Exclude association with anencephalus, encephalocele or spina bifida subgroups |
| Severe microcephaly | Q02 | 7421 | Exclude association with anencephalus, encephalocele or spina bifida subgroups |
| Arhinencephaly/holoprosencephaly | Q041, Q042 | 74226 |  |
| Eye |  |  |  |
| Anophthalmos / microphthalmos | Q110, Q111, Q112 | 7430, 7431 |  |
| Anophthalmos | Q110, Q111 | 7430 |  |
| Congenital cataract | Q120 | 74332 |  |
| Congenital glaucoma | Q150 | 74320 |  |
| Ear, Face and Neck |  |  |  |
| Anotia | Q160 | 74401 |  |
| Congenital Heart Defects (CHD) |  |  |  |
| All CHD  | Q20-Q26 | 745, 746, 7470-7474 | Exclude PDA with GA <37 weeks Exclude peripheral pulmonary artery stenosis with GA <37 weeks |
|  Severe CHD | Q200, Q201, Q203, Q204, Q212, Q213, Q220, Q224, Q225, Q226, Q230, Q232, Q233, Q234, Q251, Q252, Q262 | 74500, 74510, 7452, 7453, 7456, 7461, 7462, 74600, 7463, 7465, 7466, 7467, 7471, 74720, 74742 | ICD9-BPA has no code for hypoplastic right heart and double outlet right ventricle |
| Common arterial truncusb | Q200 | 74500 |  |
| Double outlet right ventricleb | Q201 | No code |  |
| Transposition of great vesselsb | Q203 | 74510 |  |
| Single ventricleb | Q204 | 7453 |  |
| Ventricular septal defect | Q210 | 7454 |  |
| Atrial septal defect | Q211 | 7455 |  |
| Atrioventricular septal defectb | Q212 | 7456 |  |
| Tetralogy of Fallotb | Q213 | 7452 |  |
| Triscuspid atresia and stenosisb | Q224 | 7461 |  |
| Ebstein’s anomalyb | Q225 | 7462 |  |
| Pulmonary valve stenosis | Q221 | 74601 |  |
| Pulmonary valve atresiab | Q220 | 74600 |  |
| Aortic valve atresia/stenosisb | Q230 | 7463 | ICD9-BPA has no code for atresia |
| Mitral valve anomaliesb | Q232, Q233 | 7465, 7466 |  |
| Hypoplastic left heartb | Q234 | 7467 |  |
| Hypoplastic right heartb | Q226 | No code |  |
| Coarctation of aortab | Q251 | 7471 |  |
| Aortic atresia/interrupted aortic archb | Q252 | 74720 |  |
| Total anomalous pulmonary venous returnb | Q262 | 74742 |  |
| PDA as only CHD in term infants (GA +37 weeks) | Q250 | 7470 |  |
| Respiratory |  |  |  |
|  Choanal atresia | Q300 | 7480 |  |
|  Cystic adenomatous malf of lung | Q3380 | No code |  |
| Oro-facial clefts |  |  |  |
|  Cleft lip with or without cleft palate | Q36, Q37 | 7491, 7492 |  |
|  Cleft palate | Q35 | 7490 |  |
| Digestive System |  |  |  |
| Oesophageal atresia with/ without trachea-oesophageal fistula | Q390-Q391 | 75030-75031 |  |
|  Duodenal atresia or stenosis | Q410 | 75110 |  |
| Atresia or stenosis of other parts of small intestine | Q411-Q418 | 75111-75112 |  |
|  Ano-rectal atresia and stenosis | Q420-Q423 | 75121-75124 |  |
| Hirschsprung’s disease | Q431 | 75130-75133 |  |
| Atresia of bile ducts | Q442 | 75165 |  |
| Diaphragmatic hernia | Q790 | 75661 |  |
| Abdominal wall defects |  |  |  |
|  Gastroschisis | Q793 | 75671 |  |
|  Omphalocele | Q792 | 75670 |  |
| Urinary |  |  |  |
|  Multicystic renal dysplasia | Q6140, Q6141 | 75316 |  |
|  Congential hydronephrosis | Q620 | 75320 |  |
| Genital |  |  |  |
|  Hypospadias | Q54 | 75260 |  |
| Indeterminate sex | Q56 | 7527 |  |
| Limb |  |  |  |
|  Limb reduction defects | Q71-Q73 | 7552-7554 |  |
|  Club foot – talipes equinovarus | Q660 | 75450 |  |
|  Hip dislocation and/or dysplasia | Q650-Q652, Q6580, Q6581 | 75430 |  |
|  Polydactyly | Q69 | 7550 |  |
|  Syndactyly | Q70 | 7551 |  |
| Other anomalies |  |  |  |
| Craniosynostosis | Q750 | 75600 |  |
| Situs inversus | Q893 | 7593 |  |
| **Chromosomal anomalies** |  |  |  |
|  Down syndrome  | Q90  | 7580  |  |
|  Turner syndrome  | Q96  | 75860, 75861, 75862, 75869  |  |
|  Klinefelter syndrome  | Q980-Q984  | 7587  |  |

Footnote: All Anomalies = ALL cases of congenital anomaly, excluding cases with only minor anomalies as defined in Section 3.2 in Guide 1.4 for cases born post-2005. Cases with more than one anomaly are only counted once in the “All Anomalies” subgroup.

EUROCAT ICD-9 codes are used with the British Paediatric Association (BPA) extension code: <https://eu-rd-platform.jrc.ec.europa.eu/sites/default/files/EUROCAT-ICD9-with-BPA-Extension.pdf>

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

b Included in Severe CHD subgroup

CHD, congenital heart defect; GA, gestational age; PDA, patent ductus arteriosus