Subgroups for EUROlinkCAT studies

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 | Subgroup binary variable number (al) |
| All anomalies \* | Q-chapter, D215, D821, D1810^, P350, P351, P371 | 74, 75, 27910, 2281^, 76076, 76280, 7710, 7711, 77121 |  | al1 |
| Structural anomalies |  |  |  |  |
|  Spina Bifida | Q05 | 741 | Exclude if associated with anencephalus or encephalocele subgroups | al6 |
|  Hydrocephalus | Q03 | 7423  | Exclude hydranencephaly 74232. Exclude association with NTD subgroup | al7 |
|  Severe microcephaly | Q02 | 7421 | Exclude association with NTD subgroup | al8 |
|  Congenital cataract | Q120 | 74332 |  | al13 |
| Congenital Heart Defects | Q20-Q26 | 745, 746, 7470-7474 | Exclude PDA with GA <37 weeks Exclude peripheral pulmonary artery stenosis with GA < 37 weeks | al17 |
|  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 HRH and double outlet right ventricle | al97 |
|  Transposition of great vessels | Q203 | 74510 |  | al19 |
|  VSD | Q210 | 7454 |  | al21 |
|  ASD | Q211 | 7455 |  | al22 |
|  AVSD | Q212 | 7456 |  | al23 |
|  Tetralogy of Fallot | Q213 | 7452 |  | al24 |
|  Pulmonary valve stenosis | Q221 | 74601 |  | al27 |
|  Aortic valve atresia/stenosis | Q230 | 7463 | ICD9-BPA has no code for atresia | al29 |
|  Mitral valve anomalies | Q232, Q233 | 7465, 7466 |  | al110 |
|  Hypoplastic left heart | Q234 | 7467 |  | al30 |
|  Coarctation of aorta | Q251 | 7471 |  | al32 |
|  PDA as only CHD in term infants (GA +37 weeks) | Q250 | 7470 | Livebirths only | al100 |
|  Cystic adenomatous malf of lung | Q3380 | No code |  | al36 |
|  Cleft lip with or without cleft  palate | Q36, Q37 | 7491, 7492 |  | al102 |
|  Cleft palate | Q35 | 7490 |  | al103 |
|  Oesophageal atresia with/ without trachea-oesophageal fistula | Q390-Q391 | 75030-75031 |  | al41 |
|  Duodenal atresia or stenosis | Q410 | 75110 |  | al42 |
|  Atresia or stenosis of other parts of small intestine | Q411-Q418 | 75111-75112 |  | al43 |
|  Ano-rectal atresia and stenosis | Q420-Q423 | 75121-75124 |  | al44 |
|  Diaphragmatic hernia | Q790 | 75661 |  | al48 |
|  Gastroschisis | Q793 | 75671 |  | al50 |
|  Omphalocele | Q792 | 75670 |  | al51 |
|  Multicystic renal dysplasia | Q6140, Q6141 | 75316 |  | al54 |
|  Cong hydronephrosis | Q620 | 75320 |  | al55 |
|  Hypospadias | Q54 | 75260 |  | al59 |
|  Limb reduction defects | Q71-Q73 | 7552-7554 |  | al62 |
|  Club foot – talipes equinovarus | Q660 | 75450 | For morbidity | al66 |
|  Hip dislocation and/or dyspasia | Q650-Q652, Q6580, Q6581 | 75430 | For morbidity | al67 |
|  Polydactyly | Q69 | 7550 | For morbidity | al68 |
|  Syndactyly | Q70 | 7551 | For morbidity | al69 |
|  Craniosynostosis | Q750 | 75600 |  | al75 |
|  |  |  |  |  |
| **Chromosomal anomalies** |  |  |  |  |
|  Down syndrome  | Q90  | 7580  | With or without al17 and al40 | Al89 |
|  |  |  |  |  |
| All subgroups below analysed as rare |  |  |  |  |
|  |  |  |  |  |
| **Chromosomal anomalies** |  |  |  |  |
|  Trisomy 13 | Q914-Q917 | 7581 | For mortality | Al90 |
|  Trisomy 18 | Q910-Q913 | 7582 | For mortality | Al91 |
|  Turner syndrome  | Q96  | 75860, 75861, 75862, 75869  |  | Al92 |
|  Klinefelter syndrome  | Q980-Q984  | 7587  |  | Al93 |
|  |  |  |  |  |
| Rare structural anomalies with a EUROCAT subgroup |  |  |  |  |
|  Encephalocele | Q01 | 7420 | Exclude if ass with anencephalus subgroup | al5 |
|  Arhinencephaly / holoprosencephaly | Q041, Q042 | 74226 |  | al9 |
|  Anophthalmos / microphthalmos | Q110, Q111, Q112 | 7430, 7431 |  | al11 |
|  Anophthalmos | Q110, Q111 | 7430 |  | al12 |
|  Congenital glaucoma | Q150 | 74320 |  | al14 |
|  Anotia | Q160 | 74401 |  | al16 |
|  Common arterial truncus | Q200 | 74500 |  | al18 |
|  Double outlet right ventricle | Q201 | No code |  | al109 |
|  Single ventricle | Q204 | 7453 |  | al20 |
|  Triscuspid atresia and stenosis | Q224 | 7461 |  | al25 |
|  Ebstein’s anomaly | Q225 | 7462 |  | al26 |
|  Pulmonary valve atresia | Q220 | 74600 |  | al28 |
|  Hypoplastic right heart | Q226 | No code |  | al31 |
|  Aortic atresia / interrupte aortic arch | Q252 | 74720 |  | al111 |
|  Total anom pulm venous return | Q262 | 74742 |  | al33 |
|  Choanal atresia | Q300 | 7480 |  | al35 |
|  Hirschsprung’s disease | Q431 | 75130-75133 |  | al45 |
|  Atresia of bile ducts | Q442 | 75165 |  | al46 |
|  Annular pancreas | Q451 | 75172 |  | al47 |
|  Indeterminate sex | Q56 | 7527 |  | al60 |
|  Situs inversus | Q893 | 7593 |  | al79 |
|  VATER/VACTERL | Q8726 | 759895 |  | al112 |
|  |  |  |  |  |
| New subgroups for EUROlinkCAT |  |  |  |  |
|  |  |  |  |  |
| Structural anomalies |  |  |  |  |
| Anomalies of corpus callosum | Q040 | 74221 |  | aud1 |
| Anomalies of intestinal fixation | Q433 | 7514 |  | aud3 |
| Unilateral renal agenesis | Q600 | No code |  | aud4 |
| Accessory kidney /Double or triple kidney and pelvis | Q630 | 75330, 75331 |  | aud5 |
| Bladder exstrophy  | Q641 | 7535 |  | aud6 |
| Epispadia | Q640 | 75261 |  | aud7 |
| Posterior urethral valves | Q6420 | 75360 |  | aud8 |
| Prune Belly | Q794 | 75672 |  | aud9 |
| Arthrogryposis multiplex congenita | Q743 | 75580 |  | aud10 |
|  |  |  |  |  |
| Genetic syndromes |  |  |  |  |
| Di George syndrome  | D821 | 27910 |  | aud14 |
| Goldenhar syndrome | Q8704 | 75606 |  | aud15 |
| Cornelia de Lange syndrome | Q8712 | 759821 |  | aud16 |
| Noonan syndrome | Q8714 | 759896 |  | aud17 |
| Prader-Willi | Q8715 | 759872 |  | aud18 |
| Beckwith Wiedeman syndrome | Q8730 | 759874 |  | aud20 |
| Williams syndrome | Q8784 | No code |  | aud21 |
| Angelman syndrome | Q8785 | No code |  | aud22 |
|  |  |  |  |  |
| Chromosomal anomalies |  |  |  |  |
| Wolff-Hirschorn syndrome | Q933 | 75832 |  | aud23 |
| Cri-du chat syndrome | Q934 | 75831 |  | aud24 |
| Karyotype XXX | Q970 | 75885 |  | aud25 |
|  |  |  |  |  |
| Sequences |  |  |  |  |
| Pierre-Robin sequence | Q8708 | 75603 |  | aud27 |

**Footnote:** aud13 subgroup **-** Meckel-Gruber syndrome (Q6190) - has been removed from the list as it is not going to be analysed due to a very small number of live births.

Standardisation committee meeting September 2018:

Decision to remove subgroups for fetal alcohol syndrome, valproate syndrome and maternal infections due to small numbers and too heterogeneous subgroup for maternal infection.

12.11.2018: More subgroups have been deleted due to small numbers in the dataset (inclusion criteria is at least 5 livebirths in 3 registries). Excluded are:

Megalencephaly

Ectodermal dysplasia

Alagille syndrome

Holt-Oram

Caudal regression sequence

**Revision date**: 14 January 2020. For WP4, code 75331, “Double or triple kidney and pelvis,” was added to the aud5 subgroup.