

Table S1 NCARDS inclusion and exclusion criteria for registerable congenital anomalies

Anomaly type*	Routine inclusion (per ICD-10 code blocks)	Inclusions subject to criteria	Exclusions	Differences with EUROCAT	Enhanced registration
Nervous system anomalies	Q00-Q07 Congenital malformations of the nervous system	MEASUREMENT / SEVERITY Q02 Microcephaly Q03.8 (part) Congenital ventriculomegaly [of lateral ventricle(s)] Q04.32 Reduction anomalies of cerebellum CLINICAL SIGNIFICANCE Q04.61 (part) Arachnoid cyst	EXCLUDE IN ISOLATION Q0780 Jaw-winking syndrome Q0782 Crocodile tears EXCLUDE always Q04.6 (part) Porencephaly	Q04.6 (part) Porencephaly - EUROCAT inclusion but NCARDS excludes Q04.61 Single congenital cerebral cyst - EUROCAT exclusion in isolation but NCARDS includes Arachnoid cyst	YES Q00* Anencephaly and similar malformations Q01* Encephalocele Q05* Spina bifida
Eye anomalies	Q10-Q15 Congenital malformations of eye	NA	EXCLUDE IN ISOLATION Q10.1 Congenital ectropion, Q10.2 Congenital entropion, Q10.3 Other congenital malformations of eyelid, Q10.5 Congenital stenosis or stricture of lacrimal duct Q13.2 (part) Anisocoria, congenital, Q13.5 Blue sclera Minor anomalies and dysmorphic features	NA	NO

Anomaly type*	Routine inclusion (per ICD-10 code blocks)	Inclusions subject to criteria	Exclusions	Differences with EUROCAT	Enhanced registration
Ear, face and neck anomalies	Q16 Congenital malformations of ear causing impairment of hearing	CLINICAL SIGNIFICANCE Q18.3 Webbing of neck	EXCLUDE IN ISOLATION Q17* Other congenital malformations of ear Q18.0-Q18.2 Branchial cleft malformations, Q18.4-Q18.9 Macrostomia, Microstomia, Macrocheilia, Microcheilia, Other congenital malformations of face and neck, Dysmorphic features NOS Minor anomalies and dysmorphic features	NA	NO

Anomaly type*	Routine inclusion (per ICD-10 code blocks)	Inclusions subject to criteria	Exclusions	Differences with EUROCAT	Enhanced registration
Heart and circulatory system anomalies	Q20-Q28 Congenital malformations of circulatory system	<p>PERSISTENCE</p> <p>Q21.10 Ostium secundum atrial septal defect (type II)</p> <p>CLINICAL SIGNIFICANCE</p> <p>Q24.8 Other specified congenital malformations of heart</p> <p>Q28.8 Other specified congenital malformations of circulatory system</p> <p>GESTATIONAL AGE AT DELIVERY & PERSISTENCE / SURGERY / SEVERITY</p> <p>Q25.0 Patent ductus arteriosus, Q25.6 Stenosis of pulmonary artery</p>	<p>EXCLUDE IN ISOLATION</p> <p>Q21.11 Patent foramen ovale</p> <p>Q24.6 Congenital heart block</p> <p>Q25.41 Persistent right aortic arch</p> <p>Q26.1 Persistent left superior vena cava,</p> <p>Q26.8 (part) Absence of superior vena cava, Interrupted inferior vena cava</p> <p>Q27.0 Congenital absence and hypoplasia of umbilical artery</p>	NA	<p>YES</p> <p>SERIOUS CARDIAC ANOMALIES</p> <p>Q20.0 Common arterial trunk</p> <p>Q20.1 Double outlet right ventricle</p> <p>Q20.3 Transposition of the great arteries</p> <p>Q20.4 Double inlet ventricle</p> <p>Q21.2* Atrioventricular septal defect</p> <p>Q21.3, Q21.82 Tetralogy of Fallot</p> <p>Q22.0 Pulmonary valve atresia</p> <p>Q22.4 Congenital tricuspid stenosis</p> <p>Q22.5 Ebstein's anomaly</p> <p>Q22.6 Hypoplastic right heart syndrome</p> <p>Q23.0 Congenital stenosis of aortic valve</p> <p>Q23.2, Q23.3 Congenital mitral stenosis and insufficiency</p> <p>Q23.4 Hypoplastic left heart</p> <p>Q25.1* Coarctation of aorta</p> <p>Q25.2 Aortic atresia, interrupted aortic arch</p> <p>Q26.2 Total anomalous pulmonary venous connection</p>

Anomaly type*	Routine inclusion (per ICD-10 code blocks)	Inclusions subject to criteria	Exclusions	Differences with EUROCAT	Enhanced registration
Respiratory system anomalies	Q30-Q34 Congenital malformations of respiratory system	CLINICAL SIGNIFICANCE Q30.8 Other congenital malformations of nose CAUSE Q33.6 Hypoplasia and dysplasia of lung	EXCLUDE IN ISOLATION Q31.5 Congenital laryngomalacia Q32.0 Congenital tracheomalacia Q32.2* Congenital bronchomalacia Q33.00 Congenital single lung cyst Q33.1* Accessory lobe of lung	NA	NO
Orofacial clefts	Q35-Q37 Cleft lip and cleft palate	MINOR FORMS Q35-Q37 Cleft lip and cleft palate	EXCLUDE IN ISOLATION Q35.7 Cleft uvula	NA	YES Q36*, Q37* Cleft lip with/without cleft palate
Abdominal wall	Q79.2 Exomphalos Q79.3 Gastroschisis Q79.5 Body Wall complex			NA	YES Q79.2 Exomphalos Q79.3 Gastroschisis

Anomaly type*	Routine inclusion (per ICD-10 code blocks)	Inclusions subject to criteria	Exclusions	Differences with EUROCAT	Enhanced registration
Gastrointestinal anomalies	Q38-Q45 Other congenital malformations of the digestive system; Q79.0* Congenital diaphragmatic hernia	<p>CLINICAL SIGNIFICANCE</p> <p>Q38.3 Other congenital malformations of tongue</p> <p>Q44.5 Other congenital malformations of bile ducts</p> <p>GESTATIONAL AGE AT DIAGNOSIS & SURGERY</p> <p>Q43.30 Malrotation of colon</p> <p>SURGERY</p> <p>Q43.5 Ectopic anus</p>	<p>EXCLUDE IN ISOLATION</p> <p>Q38.1 Ankyloglossia, Q38.2 Macroglossia, Q38.3 (part) Microglossia, Q38.4 (part) Congenital ranula, Q38.50 High arched palate</p> <p>Q40.0 Congenital hypertrophic pyloric stenosis, Q40.1 Congenital hiatus hernia, Q40.21 Dysmotility of stomach</p> <p>Q43.0* Meckel's diverticulum, Q43.20 Large intestinal dysmotility, Q43.81 Small intestinal dysmotility, Q43.82 Generalised intestinal dysmotility</p> <p>Q44.4 Choledochal cyst</p> <p>Q45.83 Congenital mesenteric cyst</p>	NA	<p>YES</p> <p>Q79.0* Congenital diaphragmatic hernia</p>

Anomaly type*	Routine inclusion (per ICD-10 code blocks)	Inclusions subject to criteria	Exclusions	Differences with EUROCAT	Enhanced registration
Reproductive system anomalies	Q50-Q56 Congenital malformations of genital organs	CLINICAL SIGNIFICANCE Q55.6 Other congenital malformations of penis	EXCLUDE IN ISOLATION Q50.1* Developmental ovarian cyst, Q50.2 Congenital torsion of ovary, Q50.5 Embryonic cyst of broad ligament, Q52.3 Imperforate hymen, Q52.4 (part) Congenital hypertrophy of hymen, Q52.5 Fusion of labia Q52.7 (part) Minor other congenital malformations of vulva Q54.4 Congenital chordee Q55.20 Retractable testis, Q55.21 Bifid scrotum EXCLUDE always Q53* Undescended testicle	Q52.7 (part) Congenital rectovulval fistula - EUROCAT exclusion in isolation but NCARDS includes	NO
Congenital anomalies of the kidney and urinary tract	Q60-Q64 Congenital malformations of the urinary system; Q794 Prune Belly	MEASUREMENT / SEVERITY Q62.0 Congenital hydronephrosis CLINICAL SIGNIFICANCE Q63.8 Other specified congenital malformations of kidney	EXCLUDE IN ISOLATION Q61.0 Congenital single renal cyst Q62.7* Congenital vesico-uretero-renal reflux Q63.3 Hyperplastic and giant kidney	NA	YES Q60.1 Bilateral renal agenesis

Anomaly type*	Routine inclusion (per ICD-10 code blocks)	Inclusions subject to criteria	Exclusions	Differences with EUROCAT	Enhanced registration
Congenital deformities of the musculoskeletal system	Q65-Q68 Congenital musculoskeletal deformities	<p>PERSISTENCE Q65.80 & Q65.81 Dysplastic hip</p> <p>CAUSE Q68.8 (part) Arthrogryposis, not otherwise specified</p>	<p>EXCLUDE IN ISOLATION Q65.3-Q65.5 Congenital subluxation of hip, Q65.6* Unstable hip Q66.1-Q66.9 Congenital deformities of feet (except include talipes equinovarus) Q67* Congenital musculoskeletal deformities of head, face, spine and chest Q68.0 Congenital deformity of sternocleidomastoid muscle, Q68.10 Clinodactyly, Q68.21 Genu recurvatum, Q68.3-Q68.5 Congenital bowing of femur, tibia and fibula</p>	Q65.80 & Q65.81 Dysplastic hip - EUROCAT exclusion in isolation but NCARDRS includes	NO
Congenital malformations of the limbs	Q69-Q74 Congenital malformations of limb(s)	<p>CLINICAL SIGNIFICANCE Q70.2 Fused toes Q703 Webbed toes, Q70.9 Syndactyly, unspecified Q74.2 Other congenital malformations of lower limb(s), including pelvic girdle Q74.82 Congenital undergrowth of limb(s)</p>	<p>EXCLUDE IN ISOLATION Q74.0 (part) Congenital cubitus valgus, Q74.00 Accessory carpal bone</p>	NA	NO

Anomaly type*	Routine inclusion (per ICD-10 code blocks)	Inclusions subject to criteria	Exclusions	Differences with EUROCAT	Enhanced registration
Congenital malformations of other parts of the musculoskeletal system	Q75-Q79 Other congenital malformations of musculoskeletal system	<p>CAUSE</p> <p>Q75.02 Trigonocephaly</p> <p>CLINICAL SIGNIFICANCE</p> <p>Q75.8* Other specified congenital malformations of skull and face bones</p> <p>Q76.4 Other congenital malformations of spine, not associated with scoliosis</p> <p>GESTATIONAL AGE AT DIAGNOSIS</p> <p>Q79.5 (part) Congenital abdominal wall defect not otherwise specified</p> <p>CONSEQUENCE</p> <p>Q79.80 Congenital constriction bands</p>	<p>EXCLUDE IN ISOLATION</p> <p>Q75.00 (part) Brachycephaly, Q75.2 Hypertelorism, Q75.3*</p> <p>Macrocephaly</p> <p>Q76.0 Spina bifida occulta, Q76.43 Congenital lordosis, postural, Q76.5 Cervical rib, Q76.60 Congenital absence of rib, Q76.62 Accessory rib, Q76.71 Sternum bifidum</p> <p>Q79.5 (part) - Congenital divarication of recti</p>	NA	<p>YES</p> <p>Q77*/Q78* (part) Lethal and severe skeletal dysplasias e.g. Thanatophoric dysplasia, Short rib-polydactyly syndrome</p>

Anomaly type*	Routine inclusion (per ICD-10 code blocks)	Inclusions subject to criteria	Exclusions	Differences with EUROCAT	Enhanced registration
Integument anomalies	Q80-Q84 Congenital malformations of integument	NA	EXCLUDE IN ISOLATION Q82.5* Congenital non-neoplastic naevus, Q82.8 (part) Dermatoglyphic anomalies, Q82.80 Abnormal palmar creases, Q82.81 Accessory skin tags Q83.3 Accessory nipple Q84.2 (part) Persistent lanugo, Q84.5 (part) Enlarged or hypertrophic nails, Q84.6 Other congenital malformations of nails	NA	NO
Phakomatoses, not elsewhere classified	Q85 Phakomatoses, not elsewhere classified	NA	NA	NA	NO

Anomaly type*	Routine inclusion (per ICD-10 code blocks)	Inclusions subject to criteria	Exclusions	Differences with EUROCAT	Enhanced registration
Teratogenic syndromes	Q86* Congenital malformation syndromes due to known exogenous causes, not elsewhere classified	MATERNAL HISTORY & CONSEQUENCE Q86.0 Fetal alcohol syndrome KNOWN AETIOLOGY & CONSEQUENCE Q86.1-Q86.8* Fetal hydantoin syndrome, Fetal warfarin syndrome, Other congenital malformation syndromes due to known exogenous causes	NA	NA	NO
Other congenital malformation syndromes	Q87* Other specified congenital malformation syndromes (multiple systems)	NA	EXCLUDE IN ISOLATION Q87.4 (part) Arachnodactyly not otherwise specified	NA	NO
Other anomalies	Q89* Other congenital malformations, not elsewhere classified	NA	EXCLUDE IN ISOLATION Q89.9 Congenital malformation, unspecified	NA	NO

Anomaly type*	Routine inclusion (per ICD-10 code blocks)	Inclusions subject to criteria	Exclusions	Differences with EUROCAT	Enhanced registration
Chromosomal	Q90-Q99 Chromosomal	IF REGISTRABLE ANOMALY Q95* Balanced rearrangements and structural markers, not elsewhere classified	EXCLUDE always Q95.0 Balanced translocation and insertion in normal individual, Q95.1 Chromosome inversion in normal individual, Q95.4 Individuals with marker heterochromatin, Q95.5 Individuals with autosomal fragile site	NA	YES Q90* Down's syndrome Q91* Edwards' syndrome and Patau's syndrome
Congenital neoplasms	D15.1 Cardiac rhabdomyoma D18.10 Cystic hygroma (congenital) D21.5 Sacrococcygeal teratoma, Sacral teratoma D21.9 Rhabdomyoma of other organs [i.e. not heart] D48.7 Teratoma, not elsewhere classified	CLINICAL SIGNIFICANCE D18.0 Haemangioma, any site, D18.1 Lymphangioma, any site	NA	EUROCAT exclusion but NCARDRS includes D15.1 Cardiac rhabdomyoma, D18.0 Haemangioma, any site, D18.1 Lymphangioma, any site, D21.9 Rhabdomyoma of other organs [i.e. not heart], D48.7 Teratoma, not elsewhere classified	NO
Di George syndrome	D82.1 Di George syndrome	NA	NA	NA	NO

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Waardenburg syndrome	E70.30 Waardenburg syndrome	NA	NA	NA	NO
Cystic fibrosis	E84* Cystic fibrosis	NA	NA	EUROCAT exclusion but NCARDS includes	NO
Spinal muscular atrophy	G12* Spinal muscular atrophy and related syndromes	NA	NA	EUROCAT exclusion but NCARDS includes	NO
Congenital chylothorax	NA	NA	EXCLUDE IN ISOLATION I89.8 Chylothorax (lymphatic); J94.0 Chylothorax (chylous)	NA	NO
Paralysis of vocal cords and larynx	NA	NA	EXCLUDE IN ISOLATION J38.0 Paralysis of vocal cords and larynx	NA	NO
Micrognathia		SEVERITY K07.0 Micrognathia	NA	NA	NO
Placental transfusion syndromes	P02.3 Fetus and newborn affected by placental transfusion syndromes	INCLUDE IN ISOLATION P02.3 (part) Twin reversed arterial perfusion sequence IF REGISTRABLE ANOMALY P02.3 (part) Twin-to-twin transfusion syndrome	NA	NA	NO

Anomaly type*	Routine inclusion (per ICD-10 code blocks)	Inclusions subject to criteria	Exclusions	Differences with EUROCAT	Enhanced registration
Congenital infections	P35.8 Congenital zika virus infection	IF REGISTRABLE ANOMALY P35.0 Congenital rubella syndrome, P35.1 Congenital cytomegalovirus infection, P37.1 Congenital toxoplasmosis	NA	NA	NO
Hydrops fetalis	P83.2 Hydrops fetalis not due to haemolytic disease	NA	EXCLUDE always P56* Hydrops fetalis due to haemolytic disease	EUROCAT exclusion but NCARDS includes	NO
Congenital hypotonia	NA	NA	EXCLUDE always P94.2 Congenital hypotonia	NA	NO

* Anomaly type is organised according to ICD-10/BPA system¹ with some amendments to align with EUROCAT subgroup coding², but allowing greater granularity.

¹World Health Organization. ICD-10: International Statistical Classification of Diseases and Related Health Problems. Geneva: World Health Organization; 2010.

²https://eu-rd-platform.jrc.ec.europa.eu/system/files/public/eurocat/Guide_1.5_Chapter_3.3.pdf