3.6 EUROCAT Description of the Congenital Anomaly Subgroups

EUROCAT Subgroup *(see relevant ICD codes in Chapter 3.3)*	**Description**	**Often diagnosed after one month of age – if not diagnosed prenatally.**
Nervous System anomalies		
Neural Tube Defects:	Neural tube defects include anencephaly, encephalocele, spina bifida and iniencephaly.	no
Anencephaly and similar	Total or partial absence of brain tissue and the cranial vault including iniencephaly and craniorachischisis. The face and eyes are present. Incompatible with life.	no
Encephalocele and meningocele	Cystic expansion of meninges and/or brain tissue outside the cranium. Covered by normal or atrophic skin.	no
Spina Bifida	Midline defect of the osseous spine usually affecting the posterior arches resulting in a herniation or exposure of the spinal cord and/or meninges.	no
Hydrocephaly	Dilatation of ventricular system, not due to primary atrophy of the brain, with or without enlargement of the skull. Defined as minimum size of lateral ventricles at 15 mm.	no
Severe microcephaly	A reduction in the size of the brain with a skull circumference less than three standard deviations below the mean for sex, age and ethnic origin. Definitions known to vary between clinicians and regions.	yes
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).	yes
Agenesis of corpus callosum	Complete agenesis of the corpus callosum.	yes

EUROCAT Subgroup *(see relevant ICD codes in Chapter 3.3)*	**Description**	**Often diagnosed after one month of age – if not diagnosed prenatally.**
Eye anomalies		
Anophthalmos/ microphthalmos	-	
Anophthalmos	Unilateral or bilateral absence of the eye tissue. Clinical diagnosis.	no
Microphthalmos	Small eye/eyes with smaller than normal axial length. Clinical diagnosis.	yes
Congenital cataract	Clouding of the normally clear lens of the eye.	yes
Congenital glaucoma	Large ocular globe as a result of increased ocular pressure in fetal life.	yes
Ear, face and neck anomalies		
Anotia and atresia/stenosis /stricture of external auditory canal	Absent pinna and/or atresia or stenosis of ear canal.	no
Congenital heart defects (CHD)		
Severe CHD	18 subgroups of severe CHD as defined below.	no
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 subvalvar septal defect.	no
Double outlet right ventricle	Both aorta and the pulmonary artery connect to the right ventricle.	no
Double outlet left ventricle	Both aorta and the pulmonary artery connect to the left ventricle.	no

EUROCAT Subgroup *(see relevant ICD codes in Chapter 3.3)*	**Description**	**Often diagnosed after one month of age – if not diagnosed prenatally.**
Complete transposition of great arteries (D-TGA)	Total separation of circulation with the aorta arising from the right ventricle and the pulmonary artery from the left ventricle.	no
Single ventricle	Only one complete ventricle with an inlet valve and an outlet portion even though the outlet valve is atretic.	no
Corrected transposition of great arteries (L-TGA)	The ventricles are reversed with the left ventricle connected to right atrium and the pulmonary artery and the right ventricle connected to the left atrium and the aorta.	no
VSD	Defect in the ventricular septum.	yes
ASD	Defect in the atrial septum.	yes
AVSD	Central defect of the cardiac septa and a common atrioventricular valve, includes primum ASD defects.	no
Tetralogy and pentalogy of Fallot	VSD close to the aortic valves, infundibular and pulmonary valve stenosis and over-riding aorta across the VSD.	no
Tricuspid atresia and stenosis	Obstruction of the tricuspid valve and hypoplasia of the right ventricle.	no
Ebstein's anomaly	Tricuspid valve displaced with large right atrium and small right ventricle.	no
Pulmonary valve stenosis	Obstruction or narrowing of the pulmonary valves which may impair blood flow through the valves.	yes
Pulmonary valve atresia	Lack of patency or failure of formation altogether of the pulmonary valve, resulting in obstruction of the blood flow from the right ventricle to the pulmonary artery.	no
Aortic valve atresia/stenosis	Occlusion of aortic valve or stenosis of varying degree, often associated with bicuspid valves.	yes for stenosis
Mitral valve atresia/stenosis	Atresia or stenosis of the mitral valve.	yes for stenosis

EUROCAT Subgroup *(see relevant ICD codes in Chapter 3.3)*	**Description**	**Often diagnosed after one month of age – if not diagnosed prenatally.**
Hypoplastic left heart (HLH/HLHS)	Hypoplasia of the left ventricle, outflow tract and ascending aorta resulting from an obstructive lesion of the left side of the heart.	no
Hypoplastic right heart (HRH/HRHS)	Hypoplasia of the right ventricle, always associated with other cardiac malformations.	no
Coarctation of aorta	Constriction of the aorta, usually where the ductus joins aorta.	yes
Aortic atresia/interrupted aortic arch	Atresia or interrupted connection of the aorta.	no
Total anomalous pulmonary venous return	All four pulmonary veins drain to right atrium or one of the venous tributaries.	no
PDA as only CHD in term infants (GA +37 weeks)	Open duct in infancy or later and requiring invasive treatment.	yes
Respiratory anomalies		
Choanal stenosis or atresia	Bony or membranous atresia or stenosis of the choanae with no or limited passage from nose to pharynx.	yes for unilateral
Congenital pulmonary airway malformations (CPAM)	Developmental malformation of the lower respiratory tract with cystic structures of the lung, usually unilateral.	no
Oro-facial clefts		
Cleft lip with and without cleft palate	Clefting of the upper lip with or without clefting of the maxillary alveolar process and hard and soft palate.	no

EUROCAT Subgroup *(see relevant ICD codes in Chapter 3.3)*	**Description**	**Often diagnosed after one month of age – if not diagnosed prenatally.**
Cleft palate	Fissure defect of the soft and/or hard palate(s) or submucous cleft without cleft lip.	no
Gastro-intestinal anomalies		
Oesophageal atresia with or without tracheo-oesophageal fistula	Occlusion or a long gap of the oesophagus with or without trachea-oesophageal fistula.	no
Duodenal atresia or stenosis	Occlusion or narrowing of duodenum.	no
Atresia or stenosis of other parts of small intestine	Occlusion or narrowing of other parts of small intestine.	no
Ano-rectal atresia or stenosis	Imperforate anus or absence or narrowing of the communication canal between the rectum and anus with or without fistula to neighbouring organs.	no
Hirschsprung's disease	Absence of the parasympatic ganglion nerve cells (aganglionosis) of the wall of the colon or rectum. May result in congenital megacolon.	yes
Atresia of bile ducts	Congenital absence of the lumen of the extrahepatic bile ducts.	yes
Annular pancreas	Pancreas surrounds the duodenum causing stenosis.	yes
Anomalies of intestinal fixation	Abnormal rotation and fixation of the intestine, may be due to Ladd's bands.	yes
Diaphragmatic hernia	Defect in the diaphragm with protrusion of abdominal content into the thoracic cavity. Various degree of lung hypoplasia on the affected side.	no

EUROCAT Subgroup *(see relevant ICD codes in Chapter 3.3)*	**Description**	**Often diagnosed after one month of age – if not diagnosed prenatally.**
Abdominal wall defects		
Gastroschisis	Protrusion of abdominal contents through an abdominal wall defect lateral to an intact umbilical cord and not covered by a membrane.	no
Omphalocele	Herniation of abdominal content through the umbilical ring, the contents being covered by a membrane sometimes ruptured at the time of delivery.	no
Congenital anomalies of kidney and urinary tract (CAKUT)		
Unilateral renal agenesis	Unilateral absence of kidney.	yes
Bilateral renal agenesis including Potter sequence	Bilateral absence, agenesis, dysplasia or hypoplasia of kidneys including Potter's syndrome. Incompatible with life.	no
Multicystic renal dysplasia	Multiple, non-communicating cysts of varying size in the kidney without functional kidney tissue.	yes
Congenital hydronephrosis including ureter obstruction	Obstruction of the urinary flow from kidney to bladder due to atresia or stenosis of the ureter or blockage of the ureter by a ureterocele. Hydronephrosis is only included if the renal pelvis is 10 mm or more after birth.	yes
Lobulated, fused and horseshoe kidney and ectopic kidney	Abnormal morphology, fusion and/or position of the kidney.	yes
Bladder exstrophy and/or epispadias	Defect in the closure of the bladder and lower abdominal wall and/or dorsal position of the urethra.	no

EUROCAT Subgroup *(see relevant ICD codes in Chapter 3.3)*	**Description**	**Often diagnosed after one month of age – if not diagnosed prenatally.**
Posterior urethral valve	Urethral obstruction with dilatation of bladder and hydronephrosis. In severe cases also distended abdomen	no
Prune belly syndrome	Deficient abdominal musculature, cryptorchidism and urinary tract abnormalities.	no
Genital anomalies		
Hypospadias	The urethral meatus is abnormally located and is displaced proximally on the ventral surface of the penis.	yes
Indeterminate sex	Includes true and pseudohermaphroditism male or female.	no
Limb anomalies		
Limb reduction defects (LRD):	Total or partial absence or severe hypoplasia of skeletal structure of the limbs	no
Transverse LRD	Absence of distal structure of the limb with proximal structures more or less normal, including amelia.	no
Longitudinal preaxial LRD	Absence or severe hypoplasia of preaxial structures of the limb (thumb, first metacarpal, radius, hallux, first metatarsal, tibia). In addition, the second digit of the hand or foot may be involved.	no
Longitudinal postaxial LRD	Absence or severe hypoplasia of postaxial structures of the limb (little finger, fifth metacarpal, ulna, fifth toe, fifth metatarsal, fibula). In addition, the fourth digit of the hand or foot may be involved.	no
Longitudinal central LRD	Absence or severe hypoplasia of central digits with or without absence or severe hypoplasia of central metacarpal/metatarsal bones, including typical and atypical types of split hand-split foot and monodactyly	no
Intercalary LRD	Absence or severe hypoplasia of a proximal-intercalary part of a limb when the distal structures (the digits), whether normal or malformed, are present. For example, absence of humerus and/or radius and ulna with a (near) normal hand or absence of femur and/or tibia and fibula with a (near) normal foot	no

EUROCAT Subgroup *(see relevant ICD codes in Chapter 3.3)*	**Description**	**Often diagnosed after one month of age – if not diagnosed prenatally.**
Club foot - talipes equinovarus	Foot anomaly with inward and downward rotation of the foot necessitating treatment (cast or surgery),	no
Hip dislocation	Location of the head of the femur outside its normal position.	no
Polydactyly	Extra digit or extra toe.	no
Syndactyly	Partial or total webbing between 2 or more digits includes minor forms, excluding 2-3 cutaneous syndactyly of the toes.	yes if mild
Other anomalies / syndromes		
Craniosynostosis	Premature closure of cranial sutures.	yes
Congenital constriction bands / amniotic band sequence resulting in major malformations	Clinical diagnosis of anomalies that are likely to be caused by constriction of body parts by amniotic bands. However, bands are not always identified. The clinical spectrum ranges from mild (constriction rings of a digit) to severe (limb reduction defects, neural tube defects, oro-facial clefts, abdominal wall defects etc.).	no
Situs inversus	Inverse position of thoracic or abdominal organs or both	yes
Conjoined twins	Siamese twins	no
VATER/VACTERL association	Association with anomalies of Vertebra, anal atresia, cardiac, trachea-oesophageal fistula, oesophageal atresia, renal anomaly and limb defects	no
Pierre Robin sequence	Micrognathia, glossoptosis and airway obstruction. A posterior median palatal cleft may be present as well.	no
Caudal regression sequence	Aplasia or hypoplasia of the sacrum and lumbar spine, with variable involvement of the lower limbs, gastrointestinal and urogenital tracts and nervous system.	no

EUROCAT Subgroup ***(see relevant ICD codes in Chapter 3.3)***	**Description**	**Often diagnosed after one month of age – if not diagnosed prenatally.**
Sirenomelia	The fusion of lower limbs often associated with severe spinal, urogenital and gastrointestinal anomalies and the presence of a single umbilical artery	no
Septo-optic dysplasia	A midline developmental defect in the prosencephalon with anomalies in the septum pellucidum, optic nerves and hypothalamus.	yes
Vascular disruption anomalies	Anomalies likely to be due to vascular disruption	no
Laterality anomalies	Abnormal laterality mainly affecting heart and lungs	yes
Teratogenic syndromes resulting in major anomalies	Congenital anomalies in pregnancies with known exposure to a teratogenic agent.	yes
Valproate syndrome	Fetal exposure to valproate during pregnancy with impact on fetal growth, facial appearance and development. Often associated with spina bifida	yes
Maternal infections resulting in major malformation	Specific maternal viral infections during pregnancy resulting in congenital anomalies in the fetus or infant	yes
Genetic disorders (genetic syndromes, hereditary skin disorders, skeletal dysplasias and chromosomal anomalies)	All disorders with a known or presumed genetic cause (e.g. mutation, deletion/duplication, large chromosomal aberration, methylation disorder, uniparental disomy, etc.).	
Skeletal dysplasias	A large group of genetic diseases with developmental disorders of chondro-osseous tissue	yes
Down syndrome / trisomy 21	karyotype 47,XX +21 or 47,XY +21 and translocations/mosaicism	no

EUROCAT Subgroup ***(see relevant ICD codes in Chapter 3.3)***	**Description**	**Often diagnosed after one month of age – if not diagnosed prenatally.**
Patau syndrome / trisomy 13	karyotype 47,XX +13 or 47,XY +13 and translocations/mosaicism	no
Edwards syndrome / trisomy 18	karyotype 47,XX +18 or 47,XY +18 and translocations/mosaicism	no
Turner syndrome	karyotype 45,X or structural anomalies of X chromosome	yes
Triploidy and polyploidy	A karyotype of 69,XXX, 69,XXY or 69, XYY constitutes triploidy, which can also be mosaic. Tetraploidy includes fetuses / miscarriages with 92 chromosomes.	no

Last updated: May 2022