3.6 EUROCAT Description of the Congenital Anomaly Subgroups

EUROCAT Subgroup	Description	Often diagnosed after one week of age
Nervous System		
Neural Tube Defects:	Neural tube defects inlcude anencephalus, encephalocele, spina bifida and iniencephalus	no
Anencephalus and similar	Total or partial absence of brain tissue and the cranial vault.The face and eyes are present. (incompatible with life)	no
Encephalocele	Cystic expansion of meninges and 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	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
Eye		
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
Cataract	Alteration in the transparency of the crystalline lens	yes
Congenital glaucoma	Large ocular globe as a result of increased ocular pressure in fetal life	yes
Ear		
Anotia	Absent pinna, with or without atresia of ear canal	no
Congenital heart defects (CHD)		
Severe CHD	13 subgroups of severe CHD as defined below	yes

EUROCAT Subgroup	Description	Often diagnosed after one week of age
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 subvulvar septal defect.	yes
Double outlet right ventricle	Both aorta and the pulmonary artery connect to the right ventricle	yes
Transposition of great vessels, complete	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
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	yes
Tetralogy of Fallot	VSD close to the aortic valves, infundibular and pulmonary valve stenosis and over-riding aorta across the VSD	yes
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 anomalies	Atresia, stenosis or insufficiency of the mitral valve	Yes for stenosis and insufficiency
Hypoplastic left heart	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	Hypoplasia of the right ventricle, always associated with other cardiac malformations	no
Coarctation of aorta	Constriction in the region of aorta where the ductus joins aorta	yes
Aortic atresia/interrupted aortic arch	Atresia or interrupted connection of the aorta	

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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	Open duct in infancy or later and requiring invasive treatment	yes
Respiratory		
Choanal atresia	Bony or membraneous choanae with no passage from nose to pharynx	Yes for unilateral
Cystic adenomatous malf of lung	Cystic structures of the lung, usually unilateral	No
Orofacial 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	
Cleft palate	Fissure defect of the soft and/or hard palate(s) or submucous cleft without cleft lip	No
Digestive system		
Oesophageal atresia with or without tracheo-oesophageal fistula	Occlusion or a long gap of the oesophagus with or without tracheo-oesophagael fistula	no
Duodenal atresia and stenosis	Occlusion or narrowing of duodenum	no
Atresia and stenosis of other parts of small intestine	Occlusion or narrowing of other parts of small intestine	no
Ano-rectal atresia and 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 cong 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
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
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

EUROCAT Subgroup	Description	Often diagnosed after one week of age
Urinary		
Bilateral renal agenesis including Potter syndrome	Bilateral absence, agenesis, dysplasia or hypoplasia of kidneys including Potter's syndrome. Incompatible with life	no
Multi cystic renal dysplasia	Multiple, non-communicating cysts of varying size in the kidney without functional kidney tissue	yes
Congenital hydronephrosis	Obstruction of the urinary flow from kidney to bladder. Only if renal pelvis is 10 mm or more after birth	yes
Bladder extrophy	Defect in the closure of the bladder and lower abdominal wall	no
Posterior urethral valve and/or prune belly	Urethral obstruction with dilatation of bladder and hydronephrosis. In severe cases also distended abdomen	no
Genital		
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		
Limb reduction	Total or partial absence or severe hypoplasia of skeletal structure of the limbs	no
Club foot - talipes equinovarus	Foot anomaly with equinus of the heel, varus of the hindfoot and adductus of the forefoot	no
Hip dislocation and/or dysplasia	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	yes
Other anomalies / syndromes		
Skeletal dysplasia	A large group of genetic diseases with developmental disorders of chondro-osseous tissue	Yes
Craniosynostosis	Premature closure of cranial sutures	Yes
Congenital constriction bands / Amniotic bands	Bands in the amniotic fluid that causes constriction of part of the brain, body or limbs, including limb-body-wall complex	No
Situs inversus	Inverse position of thoracic or abdominal organs or both	Yes
Conjoined twins	Siamese twins	No
Congenital skin disorders	A group of mainly genetic skin disorders in the newborn	No

EUROCAT Subgroup	Description	Often diagnosed after one week of age
VATER/VACTERL	Association with anomalies of Vertebra, anal atresia, cardiac, trachea-esophageal fistula, esophageal atresia, radial anomaly and limb defects	no
Vascular disruption anomalies (selected)	Anomalies likely to be due to vascular disruption	No
Laterality anomalies	Abnormal laterality mainly affecting heart and lungs	yes
Teratogenic syndromes with malformations	Congenital anomalies in pregnancies with known teratogenic exposure	Yes
Fetal alcohol syndrome	Fetal exposure to alcohol during pregnancy with following impact on fetal growth, facial appearance and development	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 malformation	Specific maternal viral infections during pregnancy resulting in congenital anomalies in the fetus or infant	Yes
Genetic syndromes and microdeletions	Clinically or genetically diagnosed syndromes with dysmorphic features or congenital anomalies with or without a microdeletion	Yes
Chromosomal		
Down syndrome	karyotype 47,XX +21 or 47,XY +21 and translocations/mosaicism	no
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
Klinefelter syndrome	karyotype 47,XXY or additional X-chromosomes	yes