Clinical Definitions of Subgroups

EUROCAT Subgroup	Description	Surgery necessary for long term survival	Surgery usually recommended	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. Covere by normal or atrophic skin.	yes	yes	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	yes	yes	no
Hydrocephaly	Dilatation of ventricular system, not due to primary atrophy of the brain, with or without enlargement of the skull		yes	no
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	yes
Congenital glaucoma	Large ocular globe as a result of increased ocular pressure in fetal life		yes	yes
Ear				
Anotia	Absent pinna, with or without atresia of ear canal			no
Congenital heart disease				
Common arterial truncus	Presence of a large single arterial vessel at the base of the heart (from whi the aortic arch, pulmonary and coronary arteries originate), always accompanied by a large subvulvar septal defect.	yes	yes	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	yes	yes	no
Single ventricle	Only one complete ventricle with an inlet valve and an outlet portion even though the outlet valve is atretic	yes	yes	no
VSD	Defect in the ventricular septum	yes for large defects	yes if symptoms	yes
ASD	Defect in the atrial septum		yes for large defects	yes
AVSD	Central defect of the cardiac septa and a common atrioventricular valve, includes primum ASD defects	yes	yes	yes

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	Tetralogy of Fallot	VSD close to the aortic valves, infundibular and pulmonary valve stenosis and over-riding aorta across the VSD	yes	yes	yes
	Tricuspid atresia and stenosis	Obstruction of the tricuspid valve and hypoplasia of the right ventricle	yes	yes	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	yes	yes	no
	Aortic valve atresia/stenosis	Occlusion of aortic valve or stenosis of varying degree, often associated with bicuspid valves	yes for atresia	yes	yes for stenosis
	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	yes	yes	no
	Hypoplastic right heart	Hypoplasia of the right ventricle, always associated with other cardiac malformations	yes	yes	no
	Coarctation of aorta	Constriction in the region of aorta where the ductus joins aorta	yes	yes	yes
	Total anomalous pulmonary venous return	All four pulmonary veins drain to right atrium or one of the venous tributaries	yes	yes	no
Respiratory					
	Choanal atresia	Bony or membraneous choanae with no passage from nose to pharynx		yes	yes for unilateral
	Cystic adenomatous malf of lung	Cystic structures of the lung, usually unilateral	yes	yes	no
Orofacial clefts				yes	no
	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		yes	no
Digestive system					
	Oesophageal atresia with or without tracheo-oesophageal fis	Occlusion or narrowing of the oesophagus with or without tracheo-oesophagael fistula	yes	yes	no
	Duodenal atresia and stenosis	Occlusion or narrowing of duodenum	yes	yes	no
	Atresia and stenosis of other parts of small intestine	Occlusion or narrowing of other parts of small intestine	yes	yes	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	yes	yes	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	yes	yes
	Atresia of bile ducts	Congenital absence of the lumen of the extrahepatic bile ducts	yes	yes	yes
	Annular pancreas	pancreas surrounds the duodenum causing stenosis	yes	yes	yes

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	Diaphragmatic hernia	Defect in the diaphragm with protrusion of abdominal content into the thoracic cavity. Various degree of lung hypoplasia on the affected side	yes	yes	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	yes	yes	no
	Omphalocele	Herniation of abdominal content through the umbilical ring, the contents being covered by a membrane somethimes ruptured at the time of delivery	yes	yes	no
Urinary					
	Bilateral renal agenesis including Potter syndrome	Bilateral absence, agenesis, dysplasis or hypoplasia of kidneys including Potter's syndrome. Incompatible with life			no
	Renal dysplasia	Maldevelopment of kidney tissu			yes
	Congenital hydronephrosis	Obstruction of the urinary flow from kidney to bladder. Only if renal pelvis is 10 mm or more after birth		yes	yes
	Bladder extrophy	Defect in the closure of the bladder and lower abdominal wall	yes	yes	no
	Posterior urethral valve and/or prune belly	urethral obstruction with dilatation of bladder and hydronephrosis. In severe cases also distended abdomen	yes	yes	no
Genital					
	Hypospadias	The urethral meatus is abnormally located and is displaced proximally on the ventral surface of the penis		yes	yes
	Indeterminate sex	Includes true and pseudohermaphroditism male or female		yes	no
Limb					
	Limb reduction	Total or partial absence or severe hypoplasia of skeletal structure of the limbs			no
	Upper limb reduction	Total or partial absence or severe hypoplasia of skeletal structure of the upper limb(s)			no
	Lower limb reduction	Total or partial absence or severe hypoplasia of skeletal structure of the lower limb(s)			no
	Complete absence of a limb	Complete absence of a limb			no
	Club foot - talipes equinovarus	Foot anomaly with equinus of the heel, varus of the hindfoot and adductus of the forefoot		yes	no
	Hip dislocation and/or dysplasia	Location of the head of the femur outside its normal position			no
	Polydactyly	Extra digit or extra toe		yes	no
	Syndactyly	Partial or total webbing between 2 or more digits includes minor forms		yes	yes
	Arthrogryposis multiplex congenita	Multiple congenital contractures, some times caused by neurological disease		yes	no
Musculoskeletal					
	Thanatophoric dwarfism	Letal dwarfism with very short ribs and limbs			no
	Jeunes syndrome	Asphyxiating thoracic dysplasia with constricted thoracic cage and respiratory insufficiency, usually letal in infancy			no

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Achondroplasia	Short-limb dwarfism with characteristic face and lumbar lordosis			no
Craniosynostosis	Premature closure of cranial sutures		yes	yes
Congenital constriction bands/amniotic band	Bands in the amniotic fluid that causes constriction of part of the brain, body or limbs, including limb-body-wall complex			no
Other malformations				
Asplenia	Absence of the spleen			yes
Situs inversus	Inverse position of thoracic or abdominal organs or both			yes
Conjoined twins	Siamese twins	yes		no
Disorders of skin				no
Teratogenic syndromes				
Fetal alcohol syndrome	Fetal exposure to alcohol during pregnancy with following impact on fetal growth, facial appearence and development			yes
Valproate syndrome	Fetal exposure to valproat during pregnancy with impact on fetal growth, facial appearence and development. Often associated with spina bifida			yes
Warfarin syndrome	Fetal exposure to warfarin during pregnancy with impact on fetal growt and development			yes
Maternal infections resulting in malformations	Maternal viral infections during pregnancy			yes
Genetic syndromes and microdeletions				yes
Chromosomal				
Down's 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
Edward syndrome/trisomy 18	karyotype 47,xx +18 or 47,xy +18 and translocations/mosaicism			no
Turner's syndrome	karyotype 45,x or structural anomalies of X chromosome			yes
Klinefelter's syndrome	karyotype 47,xxy or additional x-chromosomes			yes
Cri-du-chat syndrome	Partial deletion of the short arm of chromosome 5			no
Wolff-Hirschorn syndrome	Partial deletion of the short arm of one chromosome 4			no