3.2 Minor Anomalies and other conditions for Exclusion

For EUROCAT use from 2005

Cases with only minor anomalies and unspecified anomalies for exclusion should not be transmitted to EUROCAT. Minor anomalies should be described in text, coded and transmitted to EUROCAT when they are in association with major anomalies. Where a case with one or more minor anomalies only is transmitted to EUROCAT, it will be excluded by computer if the minor anomalies have specific codes which allow recognition. Some minor anomalies do not however have specific codes and cases with such isolated anomalies must always be recognised and excluded at local level on the basis of the text description.

"Minor" anomalies are excluded, when isolated, because they have lesser medical, functional or cosmetic consequences (although they may be indicators of other problems) and experience shows that their definition and diagnosis and reporting varies considerably. At the present time, it is not useful to collect data at a European level on these anomalies. We also exclude anomalies which are not always truly congenital in origin, sometimes associated with preterm birth. In addition, we exclude poorly specified conditions and recommend that for any such cases more specific information be sought from medical records. Prenatal diagnosis of anomalies of uncertain severity should be confirmed after birth.

For allocation of cases to EUROCAT subgroups, only major malformations will be considered (codes for minor anomalies will be excluded). If a registry use major ICD10 codes to describe a minor anomaly or a syndrome feature, the prevalence will be higher than the true prevalence in the registry area and data will be less comparable to other regions. Use of major ICD10 codes for minor anomalies will also have a negative impact on the classification of cases by the multiple flowchart and the surveillance of multiple congenital anomalies.

Please note that the list is not exhaustive and not all dysmorphic features are mentioned. For some features commonly used by the registries, a code outside the Q-chapter is given.

ICD10 codes marked in red were added in the EUROCAT Guide 1.4 in 2018 and 2019. These codes were implemented in the new software JRC-EUROCAT DMS in 2020.

	Specified ICD10-BPA – if present
Head	
Aberrant scalp hair patterning	
Bony occipital spur	
Brachycephaly	
Compression facies	Q671
Depressions in skull, lacunar skull, temporal flattening	Q6740
Dolichocephaly	Q672
Dysmorphic face	Q189
Broad, prominent forehead	
Coarse facies	
Flattened face	
Frontal bossing / wide forehead	

Mid face hypoplasia	
Pointed facies	
Round head shape	
Sloping forehead	
Facial asymmetry	Q670
Flat occiput	
Macrocephalus	Q753
Metopic ridge, high metopic suture	
Other congenital deformities of skull, face and jaw (including all types of abnormally shaped skull without synostosis)	Q674
Plagiocephaly – head/skull asymmetry	Q673
Third fontanelle	
Skull, late closure	
Wormian bones	
Eyes	
Anisocoria	
Blue sclera	Q135
Congenital ectropion	Q101
Congenital entropion	Q102
Crocodile tears	Q0782
Dacryocystocele	H046
Downward slanting palpebral fissures	Q103
Dystopia canthorum	Q189
Epicanthic folds	Q189
Epicanthus inversus	Q189
Exophthalmos	H052
Hypertelorism	Q752
Hypotelorism	Q189
Other congenital malformations of eyelid	Q103
Oval shaped pupils	
Prominent/protruding eyes	H052
Short palpebral fissures	Q189
Stenosis or stricture of lacrimal duct	Q105
Synophrys	Q1880
Upward slanting palpebral fissures	Q103
Ears	
Absent tragus	
Accesorry auricle, preauricular appendage, tag or lobule	Q170
Asymmetric size	Q173
Auricular pit	
Bat ear, prominent, proturberant ear	Q175
Congenital absence of ear lobe	
Darwin's tubercle	
Double lobule	Q170
Lack of helical fold	Q173
Low set ears	Q174
Macrotia	Q171

Microtia/small ears	Q172
Narrow external auditory meatus	
Posterior angulation	Q173
Primitive shape	Q173
Pointed ear, Vulcan ear, simple ear	Q173
Unspecified and minor malformation of ear	Q179
Nose	
Anteverted nares	Q189
Bifid tip of nose	Q189
Broad nasal root, anomaly of nasal root	Q189
Depressed nasal bridge	Q189
Deviation of nasal septum	Q6741
Dysmorphic nose	Q189
Flat nose	Q189
Flattened nasal bridge	Q189
Notched alas	
Pinched nose	Q189
Prominent nasal bridge	Q189
Saddle nose	Q189
Small/hypoplastic nares	Q189
Small pointed nose	Q189
Underdeveloped nasal bones	Q189
Upturned nose	Q189
Wide nasal root	Q189
Oral regions	
Aberrant frenula	
Absent /hypoplasia depressor anguli oris (asymmetric crying face)	
Alveolar crest	
Anomalies of philtrum, elongated philtrum	Q189
Bifid uvula / cleft uvula	Q357
Borderline small mandible/ minor micrognathia	
Disturbances in tooth eruption	
Enamel hypoplasia	
Glossoptosis	
High arched palate	Q3850
Macrocheilia	Q186
Macroglossia / hemi-hypertrophy of tongue	Q382
Macrostomia	Q184
Malformed teeth	
Microcheilia	Q187
Microglossia	
Microstomia	Q185
Mid-oral tongue position	
Neonatal teeth	
Prominent jaw	Q189
Ranula	
Retrognathia/ receding chin	Q674

Short philtrum	Q189
Thin lips	Q189
Tongue tie or cyst of tongue	Q381
Neck	
Broad neck	Q189
Congenital malformation of face and neck, unspecified	Q189
Congenital thymic hypoplasia	
Mild webbed neck	
Other branchial cleft malformations	Q182
Preauricular sinus or cyst	Q181
Short neck	Q189
Sinus, fistula or cyst of branchial cleft	Q180
Thymus involution	
Thyreoglossal cyst	
Torticollis	Q680
Hands	
Accessorry carpal bones	Q7400
Arachnodactyly	
Clinodactyly (5th finger)	Q6810
Duplication of thumbnail	
Enlarged or hypertrophic nails	Q845
Other congenital malformations of nails	Q846
Overlapping fingers	
Short fingers (4. 5. th finger)	
Single/abnormal palmar crease	Q8280
Small fingers	
Subluxation of phalangeal bones	
Unusual dermatoglyphics	
Feet -Limb	
Bulbous toes	
Clicking hip, subluxation or unstable hip	Q653-Q656
Hip dysplasia and other specified/unspecified hip anomalies	Q658, Q659
Clubfoot of postural origin - other cong deformities of feet	Q668
Congenital deformity of feet, unspecified	Q669
Congenital pes planus	Q665
Enlarged or hypertrophic nails	Q845
Gap between toes (1st-2nd)	
Hallux varus – other congenital varus deformities of feet	Q663
Metatarsus varus – other congenital valgus deformities of feet	Q666
Metatarsus varus or metatarsus adductus	Q662
Overlapping toes	
Pes cavus	Q667
Prominent calcaneus	
Recessed toes (4th, 5th)	
Rocker bottom feet	Q6680
Short great toe	
Syndactyly (2nd-3rd toes)	

Talipes or pes calcaneovalgus	Q664
Talipes calcaneovarus	Q661
Skin	
Accessory nipples	Q833
Accessory skin tags	Q8281
Angioma	
Cafe-au-Iait spot	
Depigmented spot	
Epibulbar dermoid	
Hemangioma if no treatment is required	
Heterochromia of hair	
Hypoplasia of toe nails	Q846
Lymphangioma if no treatment is required	
Mongoloid spot (whites)	Q8252
Neavus flammeus	Q8250
Persistent lanugo	
Pigmented naevus – congenital non-neoplastic naevus	Q825
Strawberry naevus	Q8251
Unusual placement of nipples/ wide spaced nipples	
Skeletal	
Abortive 12th rib	
Absence of rib/hypoplastic rib	Q7660
Accessory rib	Q7662
Bipartite vertebrae	
Bifid ribs	
Cervical rib	Q765
Congenital bowing of femur	Q683
Congenital bowing of fibula and tibia	Q684
Congenital bowing of long bones of leg, unspecified	Q685
Congenital bowing of upper limb	
Congenital deformity of spine	Q675
Congenital lordosis, postural	Q7643
Coronal clefts of vertebrae, incomplete	
Cubitus valgus	
Depressed sternum	
Duplication of ribs	
Fused rib, single	
Genu recurvatum	Q6821
Genua valgum	
Genua varum	
No ossification of os coccyx	
Ovoid configuration of vertebrae	
Prominent sternum	
Sacral dimple	L059
Shieldlike chest, other congenital deformities of chest	Q678
Spina bifida occulta	Q760
Sternum bifidum	Q7671

Depressed sternum / pectus excavatum	Q676
Prominent sternum / pectus carinatum	Q677
Brain	
Anomalies of septum pellucidum	
Arachnoid cysts	
Asymmetric ventricles, normal size	
Banana shaped cerebellum	
Cerebellar hypoplasia, mild	
Cerebral atrophy	
Choroid plexus cysts	
Cyst of septum pellucidum	
Enlarged cisterna magna, isolated	
Jaw-winking syndrome, Marcus Gunn's syndrome	Q0780
Periventricular leukomalacia	
Single congenital cerebral cyst	Q0461
Thin or hypoplastic corpus callosum	
Ventriculomegaly < 15 mm	
Cardiovascular	
Absence or hypoplasia of umbilical artery, single umbilical artery	Q270
Absence of vena cava superior	
Functional or unspecified cardiac murmur	R011
Cardiomegaly	I517
Cardiomyopathy	I429
Deviation of the heart axis	
Patent ductus arteriosus if GA < 37 weeks	Q250 if GA <37 weeks
Patent or persistent foramen ovale	Q2111
Peripheral pulmonary artery stenosis	Q256 if GA < 37 weeks
Persistent left superior vena cava	Q261
Persistent right aortic arch	Q2541
Persistent right umbilical vein	
Congenital heart block	Q246
Pulmonary	
Accessory lobe of lung	Q331
Azygos lobe of lung	Q3310
Bronchomalacia	Q322
Congenital laryngeal stridor	Q314
Single cyst of the lung	Q3300
Hyperplasia of thymus	
Laryngomalacia	Q3140
Pleural effusion	
Pulmonary hypoplasia, secondary	
Relaxation of diaphragm	
Thymus involution	
Tracheomalacia	Q320
Vocal cord palsy	
Gastro-intestinal	
Abdominal cyst not needing surgery	

Accessory spleen	
Anterior anus without surgery	
Choledochal cyst	Q444
Congenital adrenal hypoplasia	Q8911
Congenital cholestasis	
Congenital mesenteric cyst	Q4583
Cyst of spleen	
Diastasis recti	
Dilatation of intestine	
Functional gastro-intestinal disorders	Q4021, Q4320, Q4381, Q4382
Hepatomegaly	R160
Hiatus hernia	Q401
Inguinal hernia	K409
Liver cyst	
Meckel's diverticulum	Q430
Plica of anus	
Pyloric stenosis	Q400
Splenomegaly	R161
Transient choledochal cyst	
Umbilical hernia	
Renal	
Enlarged/thickened bladder	
Hydronephrosis with a pelvis dilatation less than 10 mm	
Hyperplastic and giant kidney	Q633
Single renal cyst	Q610
Vesico-ureteral-renal reflux	Q627
External genitals	
Bifid scrotum	Q5521
Buried penis	
Congenital chordee	Q544
Congenital adrenogenital disorders	E250
Congenital malformation of vulva	Q527
Congenital torsion of ovary	Q502
Curvature of penis	
Cysts of vulva	
Deficient or hooded foreskin/prepuce	N47
Developmental ovarian cyst(s)	Q501, Q5010, Q5011
Embryonic cyst of broad ligament	Q505
Enlarged clitoris	
Foreskin tethered to the scrotum	N47
Fusion of labia	Q525
Hydrocele of testis	P835
Hymen imperforate	Q523
Hypertrophy of hymen	
Hypoplasia of penis/micropenis	
Phimosis	N47
Prominent labia minora	

Retractile testis	Q5520
Seminal vesicle cyst	
Testicular torsion	N44
Transient ovarian cyst	
Undescended testicle	Q53
Unspecified ectopic testis	Q530
Vaginal skin tag	
Other	
Congenital malformation, unspecified	Q899
Chromosomal	
Balanced chromosomal rearrangements	Q95
Balanced translocations or inversions in normal individuals	
Balanced autosomal rearrangement in abnormal individual	Q952
Individuals with marker heterochromatin	
Individuals with autosomal fragile site	

"Non-congenital" anomalies

Pyloric stenosis – there is controversy about the congenital nature of the majority of cases.
Patent ductus arteriosus in babies <37 weeks
Hydrocephaly where a result of preterm birth rather than congenital: all cases among preterm births should be thoroughly checked before registration.

Poorly specified anomalies

Functional or unspecified cardiac murmur
Laryngomalacia and tracheomalacia
Functional gastro-intestinal disorders
Undescended testicle. Registries may choose to record this locally if they can follow-up all babies to ascertain whether the testis descends normally.
Unspecified ectopic testis
Vesico-ureteral reflux. Registries should record and transmit to EUROCAT the underlying anomaly, if present.
Clicking hip
Clubfoot where there is no further specification of whether malformation or postural origin