3.5 Detailed Congenital Anomaly Coding Guidelines

Remember always to give as specified code as possible

Other specified congenital anomaly codes
(Q188, Q178 Q308, Q288, Q742, Q764, Q758, Q445, Q638 etc)
Use these codes only to code major anomalies not specifically mentioned in Q chapter. Describe anomaly in the text. Do not use these codes for minor anomalies listed in the list of minor anomalies for exclusion.
Coding Committee April 2018

Q00   Anencephaly and similar malformations

Q01   Encephalocele

Q02   Microcephaly

MICROCEPHALY
Report microcephaly if head circumference (occipito-frontal) is less than -3 SD for sex and GA. Add in written text the measurements and age at measurements. In case of maternal zika virus infection, use the code P358 for congenital viral infection in one of the malformation variables. Use local growth chart to confirm the diagnosis. Exclude secondary microcephaly (neonatal meningitis, birth asphyxia, extreme preterm birth)
Coding Committee June 2016

Q03   Congenital hydrocephalus

CONGENITAL HYDROCEPHALUS
Definition: Dilatation of ventricular system with impaired circulation and absorption of the cerebrospinal fluid. The dilatation should not be due to primary atrophy of the brain, with or without enlargement of the skull.
Please always specify the size of the ventricles.
Hydrocephalus cases can be coded using the following codes
Q030 Malformation of aqueduct of Sylvius
Q031 Atresia of foramina of Magendie and Luschka or Dandy-Walker anomaly
Approx 75% of cases with Dandy-Walker have hydrocephalus, but this code is the only way to report the Dandy-Walker anomaly
Q038 Congenital ventriculomegaly may not be due to fluid circulation abnormalities, but should be reported if the size of the ventricles is 15 mm or more. For less severe prenatally detected ventriculomegaly (10-14 mm) it is recommended to follow the case until further imaging and a final diagnosis has been found postnatally.
Q039 Unspecified congenital hydrocephalus
Coding Committee June 2011

Q0380 CLOVERLEAF SKULL: It is caused by the premature closure of several sutures and is apparent from birth. The ICD/BPA code is wrong. Use Q7503 in stead
Coding Committee June 2011

Q04 Other congenital malformations of brain

Q040/Q0400 CORPUS CALLOSUM
Malformation of/ agenesis of corpus callosum: do not use a hydrocephalus code for the dilatation of the ventricles associated with this anomaly.
Coding Committee June 2011

Q043 OTHER REDUCTION DEFORMITIES OF BRAIN
Aicardi syndrome, Joubert syndrome, Miller-Dieker syndrome and Walker-Warburg syndrome: Please code these genetic syndromes with the code Q878 and give the syndrome name in written text. Also give the code for the diagnosed cerebral anomaly in malf1 (Q043 for reduction deformity of brain, Q0433 for lissencephaly)
Coding Committee September 2018

Q0432 REDUCTION ANOMALIES OF CEREBELLUM
In livebirths this should be reported only if there is a significant reduction in the size of cerebellum. Mild reduction of the size of cerebellum is considered a minor anomaly.
Coding Committee April 2018

Q0435 HYDRANENCEPHALY
Congenital absence of cerebral hemispheres with preservation of midbrain and cerebellum. May result from widespread vascular occlusion, infections, prolonged severe hydrocephalus. Coding Committee June 2011

Q048 OTHER SPECIFIED CONGENITAL MALFORMATIONS OF BRAIN
Congenital ventriculomegaly should be reported as hydrocephaly (Q03 subchapter) if the size of the ventricles is 15 mm or more. For less severe prenatally detected ventriculomegaly (10-14 mm) it is recommended to follow the case until further imaging and a final diagnosis has been found postnatally. If reported to EUROCAT, give written text description without a Q-code. Asymmetric ventricles and ventriculomegaly secondary to neonatal cerebral hemorrhage or meningitis should not be reported to EUROCAT
Coding Committee April 2018

Q048 OTHER SPECIFIED CONGENITAL MALFORMATIONS OF BRAIN
Colpocephaly is a congenital brain abnormality in which the occipital horns - the posterior or rear portion of the lateral ventricles (cavities) of the brain - are larger than normal because white matter in the posterior cerebrum has failed to develop or thicken. The term colpocephaly should not be used for asymmetric or mildly dilated ventricles, as these are minor anomalies.
Coding Committee April 2018

Q05 Spina bifida

CODING OF SPINA BIFIDA
In ICD/BPA 10 coding of spina bifida should be based on one code only. The codes in Q05 describe both the site of the defect and if hydrocephalus is present or not. Code the highest position of the defect (ex: thoracic if both thoracic and lumbar). Add the 4.th digit to describe if the defect is open or closed. The BPA extension can be found under (http://www.eurocat-network.eu/content/EUROCAT-Q-Chapter-2008.pdf).
Coding Committee meeting 2006 and EUROCAT Communication July 2006

CODING OF SPINA BIFIDA WITH ARNOLD CHIARI MALFORMATION.
In ICD/BPA9 there was a specified code for spina bifida with Arnold Chiari malformation. This code does not exist in ICD/BPA10. For coding spina bifida with Arnold Chiari malformation use the best possible code for spina bifida within Q05 (se coding tips) and add the code for Arnold Chiari: Q070
Coding Committee 2007

SPINA BIFIDA OCCULTA AND OTHER VARIATIONS
We include all spina bifida cases in EUROCAT - open or covered - in our prevalence.
We exclude spina bifida occulta if the only malformation is the vertebrae detected by x-ray and no neurological deficits.
If only tethered cord or lipomylmyelomeningocele is present we recommend you use the code Q068. This means that we record the case but outside the NTD subgroup.
We have followed the advice from Peter Harper: Practical genetic counselling.
Coding Committee August 2007

Q06   Other congenital malformations of spinal cord

Q068 TETHERED CORD.
Use the code Q068 "Other specified malformation of spinal cord" and specify tethered cord and spinal location in written text.
Coding Committee August 2007

Q068 LIPOMYELOMENINGOCELE
Use the code Q068 "Other specified malformation of spinal cord" and specify the malformation including location in text
Coding Committee August 2007

Q07   Other congenital malformations of nervous system

Q070 CODING OF SPINA BIFIDA WITH ARNOLD CHIARI MALFORMATION.
In ICD/BPA9 there was a specified code for spina bifida with Arnold Chiari malformation. This code does not exist in ICD/BPA10. For coding spina bifida with Arnold Chiari malformation use the best possible code for spina bifida within Q05 (se coding tips) and add the code for Arnold Chiari: Q070
Coding Committee 2007

Q10   Congenital malformations of eyelid, lacrimal apparatus and orbit

Q11   Anophthalmos, microphthalmos and macrophthalmos

Q12   Congenital lens malformations

Q13   Congenital malformations of anterior segment of eye

Q14   Congenital malformations of posterior segment of eye

Q15   Other congenital malformations of eye

Q16   Congenital malformations of ear causing hearing impairment

Q17   Other congenital malformations of ear

Q18   Other congenital malformations of face and neck

Q183 WEBBING OF NECK/PTERYGIUM
Conditions leading to webbed neck and requiring surgery include a collapsed cystic hygroma and multiple pterygium syndrome for example. These must be coded as major anomaly Q183. Mild webbed neck should be regarded as a minor anomaly unless requiring surgery and coded Q189.
Coding Committee April 2018

Q189 DYSMORPHIC FACE.
If a case with one or more major malformations also has a dysmorphic face but no syndrome diagnosis or karyotype anomaly, use the code Q189: "malformation of face and neck, unspecified" and give the written text: dysmorphic face. This code is on the list of minors for exclusion and therefore will not affect our prevalence data and subgroups. The advantage is that we will be able to see which cases in the total database may later prove to have a syndrome.
Coding Committee August 2007

Q189 MILD WEBBED NECK
Mild webbed neck should be regarded as a minor anomaly unless requiring surgery and coded Q189. Conditions leading to webbing and requiring surgery include a collapsed cystic hygroma and multiple pterygium syndrome for example. These must be coded as major anomaly Q183.
Coding Committee April 2018

Q189 BIFID TIP OF NOSE
This is a minor anomaly often associated with chromosomal abnormalities. It can be coded as part of a description of dysmorphic features using Q189. The code Q302 should not be used.
Coding Committee April 2018

Q189 CONGENITAL MALFORMATION OF FACE AND NECK, UNSPECIFIED
Use this code for dysmorphic features/dysmorphic face and not Q759
Coding Committee April 2018

Q20   Congenital malformations of cardiac chambers and connections

Q201 DOUBLE OUTLET RIGHT VENTRICLE
For this anomaly, the aorta is overriding the ventricular septum with at least 50% of the size. The pulmonary artery arises from the right ventricle
Coding Committee December 2016

Q202 DOUBLE OUTLET LEFT VENTRICLE
For this anomaly the aorta and the pulmonary artery arises exclusively or predominantly from the left ventricle
Coding Committee December 2016

Q203 TRANSPOSITION OF GREAT ARTERIES (TGA**)**
This code is for classical transposition of great arteries (complete transposition, d-transposition) with aorta arising from the right ventricle and the pulmonary artery arising from the left ventricle and with normal or almost normal size of both ventricles. Some infants have a VSD, associated with later diagnosis. The VSD should be coded separately. The patent ductus should not be coded. Malpositioned great arteries should be coded with Q208 unless more specified.
Coding Committee December 2016

Q204 SINGLE VENTRICLE, COMMON VENTRICLE, DOUBLE INLET LEFT VENTRICLE, COR TRILOCULARE BIATRIATUM
A single ventricle has absence of the ventricular septum. If there is a hypoplastic ventricle, the anomaly should be coded as hypoplastic left heart (Q234) or hypoplastic right heart (Q226)
Coding Committee November 2013

Q205 DISCORDANT ATRIOVENTRICULAR CONNECTION (Corrected transposition, levo-transposition, ventricular inversion**)**
This code is to be used when there is both atrioventricular discordance and ventriculo-arterial discordance. This means that the aorta and the pulmonary artery are transposed, with the aorta anterior and to the left of the pulmonary artery; the morphological left and right ventricles with their corresponding atrioventricular valves are also transposed
Coding Committee December 2016

ATRIAL ISOMERISM AND IVEMARK SYNDROME WITH ASPLENIA/POLYSPLENIA
Q206 is the code for atrial isomerism or Ivemark syndrome with or without asplenia/polysplenia. Add a code for the spleen anomalies if present: Q8900 asplenia or Q8908 polysplenia. Additional codes for situs inversus (Q893*) may also be added if present
Coding Committee June 2013

Q21   Congenital malformations of cardiac septa

Q211 ASD
For ASD use the 4-digit codes to distinguish between ASD secundum (Q2110) and persistent foramen ovale (Q2111). In registries where information is available for ASD secundum (Q2110) include only defects with flow across the defect still present 6 months after birth.
Coding Committee August 2007

Q213 TETRALOGY OF FALLOT
The ICD10-code for Tetralogy of Fallot is Q213. Do not use other additional cardiac codes for this malformation except if there is pulmonary valve atresia (Q220).
The cardiac malformation "VSD+pulmonary valve stenosis" is a different entity/disease than Tetralogy of Fallot as etiology, epidemiology and outcome are different.
EUROCAT Communication January 2005, edited by Coding Committee December 2016

Q22   Congenital malformations of pulmonary and tricuspid valves

Q226 HYPOPLASTIC RIGHT HEART
There is no clear definition of hypoplastic right heart, but in most cases it is a consequence of limited flow through the ventricle in fetal life due to tricuspid valve atresia or pulmonary (valve) atresia with

intact ventricular septum. The code Q226 may be used if the outcome is a univentricular heart. Add codes for the associated cardiac anomalies such as tricuspid atresia or pulmonary atresia.
Coding Committee December 2016

Q23   Congenital malformations of aortic and mitral valves

Q234 HYPOPLSTIC LEFT HEART
Hypoplastic left heart is a spectrum of cardiac defects characterized by severe underdevelopment of the left side of the heart. The definition includes atresia or marked hypoplasia of aortic orifice or valve with hypoplasia of ascending aorta and defective development of left ventricle (with or without mitral valve stenosis/atresia). The code Q234 includes all these anomalies on the left side of the heart. If there are anomalies on the right side of the heart, codes for these anomalies should be added. Do not code the patent ductus.
Coding Committee December 2016

Q24   Other congenital malformations of heart

Q248 OTHER SPECIFIED CHD
If a TOPFA is performed for a severe CHD without a final diagnosis, please use code Q248 for other specified CHD. Always give written text as specific as possible.
Coding Committee November 2017

Q249 UNSPECIFIED CARDIAC ANOMALY
Only for use if the cardiac diagnosis is certain and completely unknown. Do not use Q249 for a heart murmur. There must be a diagnosis of CHD before reporting to EUROCAT.
Do not use this code for cardiomyopathy – use I42 in the cardiac chapter.
Cardiac hypertrophy due to maternal diabetes is not a congenital heart defect and should not be reported with a CHD code, add in written text only.
Coding Committee November 2017

Q25   Congenital malformations of great arteries

Q250 PATENT DUCTUS (PDA)
Infants with patent ductus will be included as a major anomaly for term born babies only (GA ≥ 37 weeks). To be reported only if the PDA is still present 6 months after birth or if surgery/catheter closure is required. Many critically ill neonates have an open PDA for days or weeks with spontaneous closure. These babies should not be reported to EUROCAT. Do not code the PDA if part of a ductus dependent CHD such as transposition of great arteries (Q203), hypoplastic left heart (Q234) and coarctation of aorta (Q2510).
Coding Committee December 2016

Q26   Congenital malformations of great veins

Q27   Other congenital malformations of peripheral vascular system

Q278 for MAPCA
The European Cardiology Society propose to use the code Q278 for MAPCA (multiple aorto-pulmonary collateral arteries). This is not a perfect code, but the best to recommend. This anomaly

is not of the aorta, but of the arteries coming off the aorta. This anomaly is usually associated with Tetralogy of Fallot, but occasionally occurs as an isolated anomaly.
Coding Committee April 2017

Q28   Other congenital malformations of circulatory system

Q30   Congenital malformations of nose

Q300 CHOANAL ATRESIA /CHARGE SYNDROME
For CHARGE syndrome please use the code Q878 for the syndrome name and add CHARGE syndrome in written text. If choanal atresia is present, code with Q300

Q302 FISSURED, NOTCHED OR CLEFT NOSE
Bifid tip of nose is a minor anomaly and the code Q189 should be used together with written text. Fissured, notched and cleft nose are major anomalies.
Coding Committee April 2018

Q31   Congenital malformations of larynx

Q32   Congenital malformations of trachea and bronchus

Q33   Congenital malformations of lung

LUNG HYPOPLASIA
Lung hypoplasia associated with diaphragmatic hernia or bilateral renal agenesis is a consequence of the first malformation and it will be counted/considered as a single malformation. Lung hypoplasia after preterm rupture of the membranes is not a malformation and should therefore not be reported to EUROCAT as a case.
EUROCAT Communication November 2003

Q336 HYPOPLASIA AND DYSPLASIA OF LUNGS
Bronchopulmonary dysplasia is an acquired condition due to preterm birth and the correct code is P270 or P271. These cases should not be reported to EUROCAT and Q-codes should not be used for this disease.
Coding Committee April 2018

Q3380 CCAM - Congenital cystadenomatoid malformation of the lung
If a CCAM is detected antenatally, please code for this anomaly postnatally (and hence send the case to EUROCAT) whether or not the CCAM is confirmed by X-ray after birth. The clinical status of the baby, and whether the CCAM has been confirmed, should be added by text. This will allow us to accurately document the prevalence of this anomaly.
Coding Committee June 2013

Q34   Other congenital malformations of respiratory system

Q35   Cleft palate

CLEFT PALATE
Use only one code within chapter Q35-37. Find the code which describes the malformation in the best way. Cleft lip with cleft palate has a single code
EUROCAT Communication November 2003

CLEFT PALATE
The coding committee has decided to recommend the use of the WHO codes instead of the BPA codes for cleft palate. See table under Coding documents (see Q-Chapter under Malformation Coding Guides)
Coding Committee August 2007

Q35 CLEFT PALATE
There is no specific code for submucous cleft palate. We recommend to use the code Q353 cleft soft palate and give written text description about details (complete, incomplete)
Coding Committee November 2017

Q36   Cleft lip

CLEFT LIP
Use only one code within chapter Q35-37. Find the code which describes the malformation in the best way. Cleft lip with cleft palate has a single code
EUROCAT Communication November 2003

CLEFT LIP
The coding committee has decided to recommend the use of the WHO codes instead of the BPA codes for cleft lip. For Q369 we still recommend to use the BPA 4.th digit. See table under Coding documents (see Q-Chapter under Malformation Coding Guides)
Coding Committee August 2007

CLEFT LIP
If unilateral cleft lip give the side of defect in written text and state if the cleft lip is affecting both lip and gum/the alveolus.
Coding Committee November 2017

Q37   Cleft palate with cleft lip

CLEFT LIP AND PALATE
Use only one code within chapter Q35-37. Find the code which describes the malformation in the best way. Cleft lip with cleft palate has a single code
EUROCAT Communication November 2003

CLEFT LIP AND PALATE
The coding committee has decided to recommend the use of the WHO codes instead of the BPA codes for cleft lip and palate. See table under Coding documents (see Q-Chapter under Malformation Coding Guides)
Coding Committee August 2007

CLEFT LIP AND PALATE
Find the most appropriate code in Q37 for your case. If unilateral cleft lip give the side of defect in written text and state if the cleft lip is affecting both lip and gum/the alveolus. Also describe the position of the cleft palate in written text.
Coding Committee November 2017

Q38   Other congenital malformations of tongue, mouth and pharynx

Q39   Congenital malformations of oesophagus

Q40   Other congenital malformations of upper alimentary tract

Q41   Congenital absence, atresia and stenosis of small intestine

Q42   Congenital absence, atresia and stenosis of large intestine

Q43   Other congenital malformations of intestine

Q433 CONGENITAL MALFORMATIONS OF INTESTINAL FIXATION
Q4330 MALROTATION OF COLON
Intestinal <u>rotation</u> physiologically ends around 11 gestational weeks, and can be coded as malrotation or major anomaly after 12 weeks of gestation. The period of intestinal <u>fixation</u> is a process that physiologically lasts until shortly after birth and can be coded as major anomaly if present after this time-period and needs surgery.
Coding Committee April 2018

Q435 ECTOPIC ANUS, MISPLACED ANUS
An anterior anus is one that is positioned closer than normal to the vagina or scrotum. It should only be reported to EUROCAT if surgery was required to re-position it.
Coding Committee April 2018

Q44   Congenital malformations of gallbladder, bile ducts and liver

Q45   Other congenital malformations of digestive system

Q50   Congenital malformations of ovaries, fallopian tubes and broad ligaments

Q51   Congenital malformations of uterus and cervix

Q52   Other congenital malformations of female genitalia

Q53   Undescended testicle

Q54   Hypospadias

HYPOSPADIA
Definition: The urethral meatus is abnormally located and is displaced proximally on the ventral surface of the penis – in mild cases on the glans itself and in more severe cases at some points along the ventral surface of the penile shaft.
It is strongly recommended to use a specified code for hypospadia (Q540 to Q543) instead of the unspecified code Q549. Please also give written text description and fill in the surgery variable.
Note: Deficient or hooded foreskin by itself is not hypospadia.
Coding Committee August 2007

Q55   Other congenital malformations of male genital organs

Q556 OTHER CONGENITAL MALFORMATIONS OF PENIS
This code is for major anomalies of penis only. See list of minor anomalies and other conditions for exclusion.
Coding Committee April 2018

Q56   Indeterminate sex and pseudohermaphroditism

Indeterminate sex to be coded under malformations, not as syndrome
Coding Committee 2002

INDETERMINATE SEX
Problem: Indeterminate sex (Q564) is often over used to describe genital anomalies (ambiguous genitalia) when the sex of the baby has already been assigned.
If known to be male with ambiguous genitalia use a code to describe the genital anomaly where possible or Q559 if further details are unknown or without a specified code
If known to be female with ambiguous genitalia use a code to describe the genital anomaly where possible or Q529 if further details are unknown or without a specified code Indeterminate sex (Q564) is only to be used when the sex of the baby is not known or not determined by karyotype
Coding Committee June 2012

Q60   Renal agenesis and other reduction defects of kidney

Q61   Cystic kidney disease

Q6140 Multicystic dysplastic kidney, unilateral
This is distinct from polycystic kidneys. MCDK is usually unilateral and involves cysts of varying sizes separated by dysplastic parenchyma. The shape of the kidney is irregular and the normal renal architecture is lost. Multicystic dysplastic kidneys often shrink and disappear but if they are seen first as MCDK they should be coded as this and not as renal agenesis.
Coding Committee June 2011

6141 Multicystic dysplastic kidney, bilateral
Approximately 20% of MCDK are bilateral. This is usually a lethal condition that is primarily detected prenatally. The features are as above.

Coding Committee June 2011

Q618   Other cystic kidney disease
Included here should be cystic kidneys associated with a systemic condition such as Tuberous sclerosis, MODY 5 (Maternal diabetes and renal cysts), Bardet-Biedl etc
Coding Committee June 2011

Q619   Cystic kidney disease, unspecified
Included here should be: Kidneys that have cysts but normal parenchyma in between and prenatally kidneys that appear particularly bright (and often larger) than normal that are not polycystic or classic multicystic dysplasia
Coding Committee June 2011

Q62   Congenital obstructive defects of renal pelvis and congenital malformations of ureter

Q620 HYDRONEPRHOSIS
Only report hydronephrosis if renal pelvis is $\geq$ 10 mm after birth
Coding Committee 2003

Q620 HYDRONEPHROSIS
Defined as an obstruction of the urinary flow from kidney to bladder. Report only major cases defined as a renal pelvis at or above 10 mm after birth. Specify in written text if the hydronephrosis is unilateral or bilateral and give the maximum size of the renal pelvis measured postnatally. Hydronephrosis caused by vesico-ureteral reflux should not be reported to EUROCAT.
Coding Committee December 2007

Q621-Q626 and Q628 CONGENITAL OBSTRUCTIVE DEFECTS OF RENAL PELVIS AND CONGENITAL MALFORMATIONS OF URETER
If these anomalies are diagnosed and associated with hydronephrosis with a diameter of 10 mm or more add the code for hydronephrosis Q620 and give measurement in written text – see coding tip for hydronephrosis.
Coding Committee December 2016

Q63   Other congenital malformations of kidney

Q64   Other congenital malformations of urinary system

Q644 MALFORMATION OF URACHUS
Report cases with urachus anomalies to EUROCAT if surgery is required and if the diagnosis is made before one year of age
Coding Committee April 2017

OEIS COMPLEX
Q6410 Cloacal exstrophy. This code will include cases with OEIS complex as the literature state that these conditions are within the same spectrum. For OEIS complex, give the code Q6410 in malformation 1 and add codes for all major malformations of the case.
Coding Committee May 2010

Q6420 POSTERIOR URETHRAL VALVES

Remember to use the 4-digit ICD/BPA10 code for this anomaly. If this anomaly is diagnosed and associated with hydronephrosis with a diameter of 10 mm or more add the code for hydronephrosis Q620 and give measurement in written text – see coding tip for hydronephrosis.
Coding Committee December 2016

Q6476 MEGACYSTIS
The diagnosis megacystis is usually a prenatal diagnosis. For livebirths please try to find the cause and add the code, eg for posterior urethral valves.
Coding Committee April 2017

Q65   Congenital deformities of hip

Q6580 and Q6581 HIP DYSPLASIA
Report to EUROCAT only if the dysplasia is still present at ultrasound examination 6 weeks after birth.
Coding Committee December 2016

Q66   Congenital deformities of feet

CODING OF CLUBFOOT
Congenital clubfoot (Q660) is a major malformation for inclusion in the EUROCAT database. Another name for congenital clubfoot is talipes equinovarus and this name is used in the ICD10 written text. Clubfoot of postural origin is on the EUROCAT list of minor anomalies for exclusion (Q668). Any isolated case with this code is currently EXCLUDED from the EUROCAT database, although the code includes unspecified clubfoot. If you have a case of congenital clubfoot, you must make sure that you use the correct codes above, or your case will be excluded from the subgroup.
EUROCAT Communication December 2002

Q660 CLUBFOOT/TALIPES EQUINOVARUS
Clubfoot cases requiring surgery or Ponsetti treatment should be reported to EUROCAT as a major congenital anomaly using the code Q660. If the foot anomaly is of postural origin and not receiving treatment as mentioned, use the code Q668 and the anomaly will be classified as a minor anomaly
Coding Committee November 2013

Q67   Congenital musculoskeletal deformities of head, face, spine and chest

Q674 MICROGNATHIA /OTHER CONGENITAL DEFORMITIES OF SKULL, FACE AND JAW
This code SHOULD be used for MILD micrognathia – see coding tip for Pierre-Robin (Q8708). The code Q674 is classified as a minor anomaly
Coding Committee November 2013

Q68   Other congenital musculoskeletal deformities

Q69   Polydactyly

Q70   Syndactyly

Q71   Reduction defects of upper limb

Q7482 CONGENITAL UNDERGROWTH OF LIMBS .
Use this code for reporting short limbs without a specified diagnosis. Note that short limbs diagnosed prenatally must be followed up after birth for a final diagnosis. Do not use codes for limb reduction defects (Q718 and Q728) or codes for skeletal dysplasia unless specifically diagnosed.
Coding Committee November 2017

Q72   Reduction defects of lower limb

Q7482 CONGENITAL UNDERGROWTH OF LIMBS
Use this code for reporting short limbs without a specified diagnosis. Note that short limbs diagnosed prenatally must be followed up after birth for a final diagnosis. Do not use codes for limb reduction defects (Q718 and Q728) or codes for skeletal dysplasia unless specifically diagnosed.
Coding Committee November 2017

Q73   Reduction defects of unspecified limb

Q74   Other congenital malformations of limb(s)

Q7482 congenital undergrowth of limbs.
Use this code for reporting short limbs without a specified diagnosis. Note that short limbs diagnosed prenatally must be followed up after birth for a final diagnosis. Do not use codes for limb reduction defects (Q718 and Q728) or codes for skeletal dysplasia unless specifically diagnosed.
Coding Committee November 2017

Q75   Other congenital malformations of skull and face bones

Q7502 TRIGONENCEPHALY
This is often used to describe a somewhat triangular head shape but, for EUROCAT cases, should only be used where this is due to premature fusion of the metopic suture requiring treatment.
Coding Committee April 2018

Q7503 CLOVERLEAF SKULL
ICD/BPA 10 recommends a code in the hydrocephalus chapter, which is wrong. Use Q7503 for this anomaly**.**
Coding Committee June 2011

Q754 Mandibulofacial dysostosis – Franceschetti and Treacher-Collins
WHO recommend the code Q754 and ICD/BPA10 recommend the code Q870A. Both codes will be given in the syndrome guide. EUROCAT recommend from now to use the code Q754, to give written text description and to use the OMIM code 154500 for definite Treacher- Collins syndrome. Use OMIM code only where family history and biological markers confirm the syndrome
Coding Committee August 2007

Q759 CONGENITAL MALFORMATION OF SKULL AND FACE BONE, UNSPECIFIED
Do not use this code for dysmorphic features affecting face. Always use code Q189.
Coding Committee April 2018

Q76   Congenital malformations of spine and bony thorax

Q77   Osteochondrodysplasia with defects of growth of tubular bones and spine

SKELETAL DYSPLASIA
 If a final diagnosis of a lethal or severe skeletal dysplasia is not possible, as in TOP or neonatal
deaths without post mortem examination, use the code Q788. For late diagnosed unspecified
skeletal dysplasias use Q789
Coding Committee August 2007

Q78   Other osteochondrodysplasias

SKELETAL DYSPLASIA
 If a final diagnosis of a lethal or severe skeletal dysplasia is not possible, as in TOP or neonatal
deaths without post mortem examination, use the code Q788. For late diagnosed unspecified
skeletal dysplasias use Q789
Coding Committee August 2007

Q79   Congenital malformations of the musculoskeletal system, not elsewhere classified

LIMB-BODY-WALL COMPLEX
Q795 "Other congenital malformations of the abdominal wall" is the recommended code to us e in
malf 1 and always give written text. Code all major anomalies which include encephalocele and
craniofacial defects, internal organ defects, limb defects (mainly LRD), clubfoot.
Coding Committee May 2010

Q80   Congenital ichthyosis

Q81   Epidermolysis bullosa

Q82   Other congenital malformations of skin

Q8281 ACCESSORY SKIN TAGS
This is a minor anomaly.
Coding Committee April 2018

Q83   Congenital malformations of breast

Q84   Other congenital malformations of integument

Q85   Phakomatoses, not elsewhere classified

Q86   Congenital malformation syndromes due to known exogenous causes, not elsewhere classified

SUBGROUP: Teratogenic syndromes with congenital anomalies
Definition: syndrome caused by an environmental teratogen
Include as a EUROCAT case if at least one major anomaly present and you are sure about the aetiology (drug exposure, maternal infection etc)
Put the appropriate code in the syndrome field and codes for the associated congenital anomalies in the congenital anomaly fields
Specified codes for teratogenic syndromes are listed in the EUROCAT syndrome Guide and in the ICD/BPA10 Q-chapter
Always give text description of the syndrome and the associated anomalies (including minor anomalies and dysmorphic features without using a code for a major anomaly)
Coding Committee June 2012

Q860 FETAL ALCOHOL SYNDROME (dysmorphic)
Cases reported to EUROCAT as fetal alcohol syndrome must as minimum have dysmorphic features and/or major anomalies. Alcohol consumption must be confirmed locally. Add codes for all major anomalies
Coding Committee May 2010

Q87   Other specified congenital malformation syndromes affecting multiple systems

Q870A and Q754 Mandibulofacial dysostosis – Franceschetti and Treacher-Collins
WHO recommend the code Q754 and ICD/BPA10 recommend the code Q870A. Both codes will be given in the syndrome guide. EUROCAT recommend from now to use the code Q754, to give written text description and to use the OMIM code 154500 for definite Treacher- Collins syndrome.
Coding Committee August 2007

Q8708 PIERRE ROBIN
Pierre Robin is a sequence derived from micrognathia (hypoplastic mandible) leading to displacement of the tongue and obstructing the closure of the palate. It may be part of a genetic syndrome, but otherwise considered an isolated malformation. Correct coding will include Q8708 and written text in malf 1, a code for micrognathia (K070) in malf 2 and a cleft palate code in malf 3
Coding Committee February 2013

Q878 OTHER SPECIFIED SYNDROME
This code must always be accompanied with a written text with the syndrome name.
 EUROCAT Communication November 2004

Q878 OTHER SPECIFIED SYNDROMES
Aicardi syndrome, Joubert syndrome, Miller-Dieker syndrome and Walker-Warburg syndrome:
Please code these genetic syndromes with the code Q878 and give the syndrome name in written text. Also give the code for the diagnosed cerebral anomaly in malf1 (Q043 for reduction deformity of brain, Q0433 for lissencephaly)
Coding Committee September 2018

Q878 OTHER SPECIFIED SYNDROME
For CHARGE syndrome please use the code Q878 for the syndrome name and add CHARGE syndrome in written text. If choanal atresia is present, code with Q300
Coding Committee September 2018

Q89   Other congenital malformations, not elsewhere classified

Q897 MULTIPLE CONGENITAL MALFORMATIONS, NOT ELSEWHERE CLASSIFIED
This code should **always** be accompanied with codes (and text description) of all the separate major congenital anomalies that are present. The case will not be included in the surveillance of multiples unless specified codes are given.
Coding Committee April 2018

Q90-Q99 Chromosomal anomalies

Array results: Report only clearly pathogenic variants and if uncertain, include only copy number variants (CNVs) (duplications or deletions) larger than 1 MB. Only report cases with de novo CNVs unless the parent in familial cases also has clinical manifestations of the condition (dysmorphic features or congenital anomalies).
Coding Committee June 2015

Q90   Down syndrome

Q91   Edwards syndrome and Patau syndrome

Q92   Other trisomies and partial trisomies of the autosomes, not elsewhere classified

Q923 to be used for partial chromosomal duplication or partial trisomy.
Coding Committee June 2011

Q93   Monosomies and deletions from the autosomes, not elsewhere classified

Q935 to be used for partial chromosomal deletions or partial monosomies including those detected by array
Coding Committee June 2011

CODING OF MICRODELETIONS: We recommend coding of both the syndrome and the microdeletion. This means that the syndrome should be coded in the syndrome field using both the ICD10/BPA code and give the syndrome name in the text field. In malformation 1 give the code for microdeletion (Q936) and give the name of the microdeletion in written text. Please note that microdeletions are considered syndromes and not chromosomal anomalies. Coding example: Case with Prader-Willi syndrome and 15q11-13 del: Code Q8715 in syndrome field and write "Prader-Willi" in text field. In malformation 1 field use code Q936 and write "15q11-13 del" in text field.
Coding committee meeting 2005

Q95   Balanced rearrangements and structural markers, not elsewhere classified

Q96   Turner syndrome

Q97   Other sex chromosome abnormalities, female phenotype, not elsewhere classified

Q98   Other sex chromosome abnormalities, male phenotype, not elsewhere classified

Q982 KLINEFELTER MALE WITH KARYOTYPE 46XX
This condition does not exist and the code should not be used
Coding Committee May 2010

Q984 KLINEFELTER, UNSPECIFIED
Alternative codes will usually be possible and better
Coding Committee May 2010

Q99   Other chromosome abnormalities, not elsewhere classified

Outside Q-chapter:

D180 is the correct code for haemangiomas
D181 is the correct code for lymphangiomas
Coding Committee September 2018

P351 CONGENITAL CMV INFECTION
Infants with congenital CMV infections should be reported to EUROCAT if there are associated major
congenital anomalies (microcephaly according to EUROCAT definition, other structural anomalies).
Code P351 in the syndrome variable and major malformations in the malformation variables
Coding Committee December 2016

P358 CONGENITAL ZIKA VIRUS
This code to be used for all cases exposed to zika virus infection in pregnancy. Code P358 in the
syndrome variable and major malformations (microcephaly, other cerebral anomalies and all other
major and minor anomalies) in the malformation variables. Specify in text for all codes.
Since this is a new infection, report all cases irrespective of diagnosed congenital anomalies. Code
the maternal illness during pregnancy as A928 – "other specified mosquito-borne viral fevers". These
codes to be used only for zika virus.
Coding Committee December 2016

P371 CONGENITAL TOXOPLASMOSIS
Infants with congenital toxoplasmosis should be reported to EUROCAT if there are associated major
congenital anomalies (hydrocephaly, other structural anomalies). Code P371 in the syndrome
variable and major malformations in the malformation variables
Coding Committee December 2016

K070 MICROGNATHIA
This code is the recommended code for SEVERE micrognathia. See coding tip for Pierre-Robin
(Q8708)
Coding Committee November 2013

Please remember that the correct code for **cystic hygroma is D1810** and for **sacral teratoma D215**
Central registry January 2008

TRAP SEQUENCE

Twin **R**eversed **A**rterial **P**erfusion is a rare complication of monochorionic twin pregnancies, involving an acardiac parasitic twin and an otherwise normal "pump" twin. The acardiac twin fails to develop a head, arms and a heart.

Cases of TRAP sequence should have as **a minimum** the following essential codes and essential text:

P023   TRAP sequence

Q248   Acardia (this is better than Q89.8 as it at least specifies heart)

Q00.00 Anencephaly

Other common malformations in TRAP sequence (eg. absence of upper limbs, rudimentary alimentary tract) should also be coded, but the 3 codes above with text are suggested as a minimum.

Coding Committee February 2013 and November 2017

CODING OF PRE-PREGNANCY DIABETES

For surveillance and research on etiology it is important that we can find all cases in the EUROCAT database with pre-pregnancy diabetes. Further type-1 diabetes in increasing in prevalence among children and young people. Pre-pregnancy diabetes is coded very heterogeneous among registries. Not all registries code maternal disease before pregnancy or drug use. At the coding committee meeting in Graz in 2006 we recommended to code illness before pregnancy with codes within E10-E14, drugs with ATC codes for insulin and to code P701 "infant of diabetic mother" in the malformation variable (not the syndrome variable), even if the case is a TOPFA

Coding Committee June 2006

PRETERM COMPLICATIONS

Most complications to preterm birth, including bronchopulmonary dysplasia and persistent fetal circulation, are reported with a code in the P-chapter and will not be relevant for EUROCAT. Terminations, spontaneous abortions after GA 20 weeks and preterm birth with lung hypoplasia due to early rupture of membranes are not EUROCAT cases. Limb contractures and retrognathia due to early rupture of membranes are secondary diagnoses and not EUROCAT cases.

Coding Committee April 2017

Version OCTOBER 2018