## 3.5 Detailed Congenital Anomaly Coding Guidelines

This chapter includes the current advice for coding of specific anomalies. The chapter is updated by the EUROCAT Coding and Classification Committee whenever new coding issues appear.

**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 the 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 P354 for congenital viral infection in one of the malformation variables. Use the local growth chart to confirm the diagnosis. Exclude secondary microcephaly (neonatal meningitis, birth asphyxia, extreme preterm birth)

Coding Committee June 2016**,** adapted by the Coding Committee April 2022

### Q03 Congenital hydrocephaly

CONGENITAL HYDROCEPHALY

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.*
Hydrocephaly 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 hydrocephaly, 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 hydrocephaly

Coding Committee June 2011

DANDY-WALKER ANOMALY

The Dandy-Walker anomaly is included in the hydrocephaly subgroup under code Q031. However, not all cases of the Dandy-Walker anomaly have hydrocephaly. It is important to record whether hydrocephaly is present so that cases without hydrocephaly can be excluded in the hydrocephaly subgroup analysis.

Coding Committee May 2021

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 instead.

Coding Committee June 2011

### Q04 Other congenital malformations of brain

Q040/Q0400 CORPUS CALLOSUM

Malformation of/ agenesis of corpus callosum: do not use a hydrocephaly code for the dilatation of the ventricles associated with this anomaly.

Coding Committee June 2011

Q040 ANOMALIES OF CORPUS CALLOSUM

Partial agenesis of corpus callosum may be described very differently as hypoplastic, thin or hypogenesis of corpus callosum. Use this code for these anomalies. Some of these ultrasound and MRI findings may be minor anomalies. Use Q0400 for complete agenesis of corpus callosum which is a major anomaly.

Coding Committee June 2023

Q043 OTHER REDUCTION DEFORMITIES OF BRAIN

Aicardi syndrome, Joubert syndrome and Miller-Dieker 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, adapted by the Coding Committee April 2022

Q0432 REDUCTION ANOMALIES OF CEREBELLUM

In live births 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 hydrocephaly. Coding Committee June 2011

Q04* ANOMALIES OF SEPTUM PELLUCIDUM

“Cyst of septum pellucidum” and “Anomalies of septum pellucidum” are on the EUROCAT list of minors and should not be reported to EUROCAT with a major code. Add written text ”Agenesis of septum pellucidum” if this anomaly is part of septo-optic dysplasia coded Q044.

Coding Committee May 2021

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 a written text description without a Q-code. Asymmetric ventricles and ventriculomegaly secondary to neonatal cerebral haemorrhage 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 hydrocephaly is present or not. Code the highest position of the defect (ex: thoracic if both thoracic and lumbar). Add the 4th digit to describe if the defect is open or closed. The BPA extension can be found under https://eu-rd-platform.jrc.ec.europa.eu/sites/default/files/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 (see 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 the 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 (see coding tips) and add the code for Arnold Chiari: Q070.

Coding Committee 2007

### Q10 Congenital malformations of eyelid, lacrimal apparatus and orbit

Q100 BLEPHAROPHIMOSIS PTOSIS SYNDROME

For Blepharophimosis Ptosis syndrome please use the code Q878 for the syndrome name and add the syndrome name in written text. If the specific eye anomaly is present, use the code Q100.

Coding Committee October 2019

### Q11 Anophthalmos, microphthalmos and macrophthalmos

Q112 FRASER SYNDROME

For Fraser syndrome please use the code Q878 for the syndrome name and add Fraser syndrome in written text. If the specific eye anomaly is present, use the code Q112.

Coding Committee October 2019

Q112 Lenz Syndrome

For Lenz syndrome please use the code Q878 for the syndrome name and add Lenz syndrome in written text. If the specific eye anomaly is present, use the code Q112

Coding Committee October 2019

### 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

Q178 BRANCHIO-OTO-RENAL SYNDROME / MELNICK-FRASER SYNDROME

For Branchio-oto-renal / Melnick-Fraser syndrome please use the code Q878 for the syndrome name and add the syndrome name in written text. If the specific ear anomaly is present, use the code Q178.

Coding Committee October 2019

### 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

Q201 DOUBLE OUTLET RIGHT VENTRICLE (DORV)

Double outlet right ventricle (DORV) is the term used to describe a condition where both the pulmonary artery and aorta connect to the right ventricle. In all cases of DORV there is an associated ventricular septal defect (VSD).

There is some overlap between DORV and Fallot's tetralogy (Q21.3). In Fallot's there is overriding of the aorta, such that it is partially connected to both ventricles. However, if the over-ride exceeds 50%, the case should be coded as DORV. These cases are often referred to as DORV (Fallot's type). As with Fallot's the associated VSD **should not** be coded.

Coding Committee May 2021

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 a normal or almost normal size of both ventricles. Some infants have a VSD, associated with a 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

Q206 IVEMARK SYNDROME

For Ivemark syndrome please use the code Q878 for the syndrome name and add the syndrome name in written text. If the specific cardiac anomaly is present, use the code Q206

Coding Committee October 2019

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 ATRIAL SEPTAL DEFECT (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 after correction for gestational age in preterm births.

Coding Committee August 2007, adapted by the Coding Committee April 2022

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

Q223 Other congenital malformation of pulmonary valves

Use this code for the rare anomaly "absent pulmonary valves" and give written text description of the diagnosis. When the pulmonary valve is absent, there is regurgitation of blood in the right ventricle during diastole. This is different from pulmonary valve atresia (Q220), where the atretic valve blocks the blood flow. If the case also has tetralogy of Fallot (Q213), both Q223 and Q213 should be registered.

Coding Committee September 2025

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

Q233 CONGENITAL MITRAL VALVE INSUFFICIENCY

Congenital mitral valve insufficiency is a very rare anomaly. Echocardiography may describe mitral valve insufficiency secondary to other cardiac defects with dilatation of the left side of the heart such as a large PDA. Secondary mitral valve insufficiency and mild mitral valve insufficiency should not be reported to EUROCAT using this code.

Coding Committee May 2021

Q234 HYPOPLASTIC 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

Q248 OTHER SPECIFIED CHD and Q249 UNSPECIFIED CARDIAC ANOMALY

For terminations of pregnancy due to a major CHD without a specific cardiac code available, use the code Q248 and not the code Q249.

Coding Committee February 2024

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

Q251 COARCTATION OF AORTA

Coarctation of aorta can be classified as preductal (Q2510) and postductal (Q2511). Preductal coarctation of aorta gives critical and life-threatening symptoms when the ductus close within the first week after birth. The immediate treatment is prostaglandin infusion to open the duct before transfer to the surgical center. Postductal coarctation of aorta may be diagnosed later in infancy or childhood due to a cardiac murmur, missing femoral pulses and/ or hypertension. Please use the 4- digit code to describe which type of coarctation that is reported to EUROCAT.

Coding Committee November 2022

Q258 PRETERM CLOSURE OF THE DUCTUS

Use this code for the very rare situation where the ductus has closed before the baby is born.

Coding Committee April 2022

### Q26 Congenital malformations of great veins

Q265 Anomalous portal venous connection

Use this code for the rare anomaly “portosystemic or portocaval shunt”.

Coding Committee September 2025

### Q27 Other congenital malformations of peripheral vascular system

Q278 for MAPCA

The European Cardiology Society proposes 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 AND STENOSIS

This code should only be used for cases with choanal atresia and stenosis. Stenosis or atresia of nares and stenosis of piriform aperture should be coded with Q308 "Other congenital malformations of nose" or other codes in the Q30 subchapter. Minor anomalies of nose should be coded with Q189 with written text description.

Coding Committee June 2023

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

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

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

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 4th digit. See table under Coding documents (see Q-Chapter under Malformation Coding Guides).

Coding Committee August 2007

If unilateral cleft lip give the side of the defect in written text and state if the cleft lip is affecting both lip and gum/the alveolus.

Coding Committee November 2017

Q361 MEDIAN CLEFT LIP AND CYCLOPIA

Most cleft lips are uni- or bi-lateral, but very rarely a cleft can be in the true midline. These tend to be in association with other midline defects such as holoprosencephaly (Q042) or partial forms of that, including cyclopia (Q8703). If a median cleft lip is found it should be coded as Q361.

Coding Committee October 2019, revised June 2023

### 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

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

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

Q380 VAN DER WOUDE'S SYNDROME

For Van der Woude's syndrome please use the code Q878 for the syndrome name and add the syndrome name in written text. If the specific lip anomaly is present, use the code Q380.

Coding Committee October 2019

### 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

Q431 HIRSCHSPRUNG'S DISEASE

In Hirschsprung's disease, ganglion cells are absent from the colon. Only cases with a diagnosis of Hirschsprung's disease based on a biopsy from the large intestine should be reported to EUROCAT.

Coding Committee February 2024

Q433 CONGENITAL MALFORMATIONS OF INTESTINAL FIXATION

Q4330 MALROTATION OF COLON

Intestinal rotation physiologically ends around 11 gestational weeks and can be coded as malrotation or major anomaly after 12 weeks of gestation. The period of intestinal fixation is a process that physiologically lasts

until shortly after birth and can be coded as a 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

Q447 OTHER CONGENITAL MALFORMATIONS OF LIVER

Major anomalies to be reported with this code include accessory liver, total or partial agenesis of liver and ectopic liver (left-sided). Hepatomegaly is an enlarged liver. It is a non-specific sign and is a symptom of an underlying disease. The correct code for hepatomegaly is R160. Liver calcifications, liver haemangioma and congenital liver cysts are all minor anomalies according to EUROCAT definitions. If these anomalies are reported to EUROCAT, codes outside the Q-chapter should be used. Q4471 is to be used only for Alagille syndrome.

Coding Committee October 2024

### Q45 Other congenital malformations of digestive system

### Q50 Congenital malformations of ovaries, fallopian tubes and broad ligaments

### Q51 Congenital malformations of uterus and cervix

Q518 MURCS SYNDROME

For MURCS syndrome: If the specific genital anomaly is present, use the code Q518. Do not use a code for a genetic anomaly.

MURCS association consists of a non-random association of **Mü**llerian duct aplasia (usually uterus and upper 2/3 of the vagina), **R**enal aplasia or ectopia, and **C**ervicothoracic **S**omite dysplasia (malformations of the radial ray, ribs, and cervicothoracic spine). Most of affected patients have congenital heart defects. It is also known as Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome type 2.

Coding Committee October 2019 and revised April 2022

Q518 KAUFMAN-McKUSICK SYNDROME

For Kaufman-McKusick syndrome please use the code Q878 for the syndrome name and add the syndrome name in written text. If the specific genital anomaly is present, use the code Q518.

Coding Committee October 2019

### Q52 Other congenital malformations of female genitalia

### Q53 Undescended testicle

### Q54 Hypospadias

HYPOSPADIAS

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 hypospadias (Q540 to Q543) instead of the unspecified code Q549. Please also give a written text description and fill in the surgery variable.

Note: Deficient or hooded foreskin by itself is not hypospadias.

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 the 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 a syndrome.

Coding Committee 2002

INDETERMINATE SEX

Indeterminate sex (Q564) is only to be used when the sex of the baby is not known or not determined by karyotype. 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 often over-used to describe genital anomalies (ambiguous genitalia) when the sex of the baby has already been assigned.

Coding Committee June 2012, edited October 2024

### Q60 Renal agenesis and other reduction defects of kidney

Q606 POTTER'S SEQUENCE

Potter's sequence refers to severe renal anomalies which lead to oligo/anhydramnios. Talipes equinovarus, distinct facial features (Potter facies) and lung hypoplasia can be present and should be coded. For EUROCAT it is important to also code the underlying kidney problem, e.g. bilateral renal agenesis, multicystic dysplastic kidneys or polycystic kidneys.

Coding Committee May 2021, edited October 2024

### 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

Q6141 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 HYDRONEPHROSIS

Only report hydronephrosis if renal pelvis is ≥ 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

Q630 ACCESSORY KIDNEY

Please note that accessory kidney and ectopic kidney is not the same. Accessory kidney means that there are more than 2 kidneys. This is very rare. An ectopic kidney is located below (pelvic), above (thoracic) or on the opposite side (crossed ectopia) of the kidney's normal position.

Coding Committee November 2022

Q632 ECTOPIC KIDNEY

Please note that ectopic kidney and accessory kidney is not the same. See text for Q630.

Coding Committee November 2022

### 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

Q6420 POSTERIOR URETHRAL VALVES and Q6476 MEGACYSTIS

The diagnosis megacystis is usually a prenatal diagnosis and the primary anomaly may be difficult to diagnose by the prenatal ultrasound examination. For TOPFA please check the postmortem examination to find the primary cause and add the specific code, e.g. for posterior urethral valves, Prune-Belly, cloacal anomalies etc. Liveborn infants should be followed up until the primary anomaly is known.

Coding Committee April 2017 and February 2024

### 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

Q681 CAMPODACTYLY

Camptodactyly is a fixed flexion deformity of one or more fingers (or toes), which is permanent. Camptodactyly should only be reported to EUROCAT if the condition does not resolve spontaneously. Do not report this as an isolated finding from a fetal post mortem examination.

Coding Committee May 2021

### Q69 Polydactyly

### Q70 Syndactyly

### Q71 Reduction defects of upper limb

Q7130 CONGENITAL ABSENCE OF ALL FINGER(S) (remainder of hand intact).

This code should be used when one or more digits of the hand are absent. Please note that this refers only to digits 2 to 5, because absence of digit 1 should be coded with Q7131. Please always provide a detailed text description, because the Q7130 code is not specific for the different types of limb reduction defects. For example: absence of dig 2-5 is considered a transverse LRD, absence of dig 3 is considered a longitudinal central LRD and absence of dig 4-5 is considered a longitudinal postaxial LRD.

Coding Committee June 2023

Q7180 CONGENITAL ABSENCE OF ALL FINGERS (code is mentioned under Q7380 in the ICD10)

Use this code when all fingers are absent. This is a transversal reduction defect.

Coding Committee November 2022

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

Q7280 CONGENITAL ABSENCE OF ALL TOES (code is mentioned under Q7380 in the ICD10)

Use this code when all toes are absent. This is a transversal reduction defect.

Coding Committee November 2022

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

Q750 PFEIFFER SYNDROME

For Pfeiffer syndrome please use the code Q878 for the syndrome name and add the syndrome name in written text. If the specific skull anomaly is present, use the code Q750.

Coding Committee October 2019

Q7500 CORONAL CRANIOSYNOSTOSIS, BRACHYCEPHALY

Brachycephaly is on the list of minors for exclusion. Therefore, the code for a major craniosynostosis should not be used for this minor anomaly although brachycephaly is mentioned under Q7500 in the Q-chapter of the ICD10/BPA. Please use the code Q674 for brachycephaly without craniosynostosis.

Coding Committee February 2024

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 hydrocephaly chapter, which is wrong. Use Q7503 for this anomaly**.**

Coding Committee June 2011

Q754 MANDIBULOFACIAL DYSOSTOSIS – FRANCESCHETTI AND TREACHER-COLLINS

WHO recommends the code Q754 and ICD/BPA10 recommend the code Q870A. Both codes will be given in the syndrome guide. EUROCAT recommends 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

Q792 OMPHALOCELE

Prenatal ultrasound scans in the first trimester may show a physiological herniation of the midgut that may look like an omphalocele but will disappear in the second trimester. These cases should not be reported to EUROCAT.

Coding Committee June 2023

Q795 LIMB BODY WALL COMPLEX OR BODY STALK ANOMALY

Q795 “Other congenital malformations of the abdominal wall” is the recommended code to use in malf 1 and always add ‘limb body wall complex’ in the text description (please use this exact wording). Code all major anomalies which include encephalocele and craniofacial defects, internal organ defects, limb defects (mainly LRD), clubfoot and others.

Coding Committee May 2010, adapted by the Coding Committee April 2022

Q796 EHLERS-DANLOS SYNDROME

EUROCAT accepts cases of Ehlers-Danlos syndrome as this diagnosis has a Q code, even though it is not associated with congenital anomalies. However, this is a very variable condition and cases with Ehlers-Danlos type 3 (also known as benign hypermobility) should **not** be included as this is not a clear diagnosis and no genetic mutations have ever been associated with it. EDS types 1, 2 and 4 are well recognised and can be included, especially if the specific mutation is known, EDS type 9 is now known as occipital horn syndrome. Most of the others are exceptionally rare, but could be included if it is a certain diagnosis.

Coding Committee May 2021

Q798 POPLITEAL WEB SYNDROME

For Popliteal web syndrome please use the code Q878 for the syndrome name and add the syndrome name in written text. If the specific musculoskeletal anomaly is present, use the code Q798

Coding Committee October 2019

Q7980 CONGENITAL CONSTRICTION BANDS /AMNIOTIC BANDS

Amniotic band sequence is a group of congenital anomalies of unknown aetiology, that might occur when bands of amnion peel away from the sac and attach or wrap around parts of the fetus, disrupting normal development. Clinical manifestation is very variable. The bands usually develop late in first trimester or early in the second trimester. For reporting to EUROCAT, use the code for amniotic bands in malf1 to describe the aetiology of the anomalies and add Q codes and written text description for all the major anomalies caused by the amniotic bands. Do not report cases with amniotic bands without major anomalies.

Coding Committee October 2019

### 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 a 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 recommends the code Q754 and ICD/BPA10 recommend the code Q870A. Both codes will be given in the syndrome guide. EUROCAT recommends 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

Q8703 CYCLOPIA

Cyclopia is a rare form of lethal holoprosencephaly (HPE) due to incomplete cleavage of the prosencephalon during embryogenesis, leading to failure of the orbits of the eye to divide into two cavities. It may also occur with a median cleft lip and has been reported as a finding in several genetic syndromes. If a syndrome diagnosis has been made, it is important to include that as well.

Coding Committee October 2019

Q8703 CYCLOPIA AND MEDIAL CLEFT

Most cleft lips are uni- or bi-lateral, but very rarely a cleft can be in the true midline. These tend to be in association with other midline defects such as holoprosencephaly (Q042) or partial forms of that, including cyclopia (Q8703). If a medial cleft lip is found it should be coded as Q361.

Coding Committee October 2019, revised June 2023

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

Q870E PENA-SHOKEIR SYNDROME (fetal akinesia)

The foetal akinesia/hypokinesia sequence (or Pena-Shokeir syndrome type I) is characterized by multiple joint contractures, facial anomalies and pulmonary hypoplasia. Whatever the cause, the common feature of this sequence is decreased foetal activity. Use ode Q870E and update the case when the final diagnosis is established.

Coding Committee November 2022

Q871 Congenital malformation syndromes predominantly associated with short stature

Q871 is the recommended code for campomelic dysplasia

Coding Committee September 2025

Q8724 SIRENOMELIA

Sirenomelia is a rare developmental defect characterized by fusion of the lower limbs and associated with some degree of lower extremity reduction. Patients also present with severe anomalies of the lower spine (e.g. sacral agenesis), as well as the urogenital and lower gastrointestinal tract (e.g. renal agenesis, absent bladder, rectal/anal atresia, and absent internal genitalia). Most cases are stillborn, or die during or shortly after birth. For reporting to EUROCAT give the code Q8724 for sirenomelia in malf1 and add Q codes and written text description for all the major anomalies included.

Coding Committee October 2019

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 and Miller-Dieker 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, adapted by the Coding Committee April 2022

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

Q878 OTHER SPECIFIED MALFORMATION SYNDROME

For Branchio-oto-renal syndrome /Melnick-Fraser syndrome (Q178), Ivemark (Q206), Van der Woude (Q380), Fraser syndrome (Q112), Lenz syndrome (Q112), (Q178), Kaufman-McKusick syndrome (Q518), Pfeiffer syndrome (Q750), Popliteal web syndrome (Q798), Q100 blepharophimosis ptosis syndrome, please use the code Q878 for the syndrome name and give syndrome name in written text. Code all specific anomalies included in the syndrome and use there the codes mentioned above.

Coding Committee October 2019

Q878 OTHER SPECIFIED MALFORMATION SYNDROME

If a congenital anomaly is clearly hereditary, use both the specific congenital anomaly code and Q878 (other specified syndrome). The MCA algorithm will then recognize the case as having a genetic origin. Examples of hereditary anomalies: postaxial polydactyly in at least two first-degree relatives, cleft palate in three consecutive generations, holoprosencephaly due to a ZIC2 mutation or a bicuspid aortic valve due to a NOTCH1 mutation.

Coding Committee April 2022

### 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

Q897 PENTALOGY OF CANTRELL / THORACO-ABDOMINAL SYNDROME

Pentalogy of Cantrell is characterized by the presence of anomalies of the diaphragm, abdominal wall, pericardium, heart and lower sternum. Pentalogy of Cantrell should be coded with Q897 in malf1 and the name should be added in written test. All separate anomalies should be coded and described in text as well. As the etiology is unknown, these cases are included in the EUROCAT surveillance.

Coding Committee October 2019

Q8980 CAUDAL DYSPLASIA SEQUENCE / CAUDAL REGRESSION SEQUENCE

Caudal regression sequence is a rare congenital anomaly with abnormal fetal development of the lower part of the spine. Affected areas can include the lower back and limbs, the genitourinary tract, and the gastrointestinal tract. Sacral agenesis (Q7641) is part of the same spectrum. For reporting to EUROCAT give the code Q8980 for caudal regression sequence in malf1 and add Q codes and written text description for all the major anomalies included in the sequence.

Coding Committee October 2019

### Q90-Q99 Chromosomal anomalies

Q90-98 CHROMOSOMAL ANOMALIES

Use the specific 3-digit code for the diagnosed karyotype and avoid using unspecified codes such as Q909, Q913, Q917, Q969 and Q989.

Coding Committee June 2023

**Array results**: Report only clearly pathogenic variants. Only report cases with de novo copy number variants unless the parent in familial cases also has clinical manifestations of the condition (dysmorphic features or congenital anomalies).

Coding Committee June 2015, revised October 2019

### 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, microduplications or partial trisomy.

Coding Committee June 2011, revised October 2019

Q925 Duplications with other complex rearrangements

Use this code for all unbalanced translocations.

Coding Committee September 2025

### 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

CODING OF MICRODELETIONS: Due to technological changes, it is increasingly difficult to discriminate between "genetic" and "chromosomal" cases, especially in relation to microdeletions. We now recommend the code Q935 to be used for coding for deletions and microdeletions without a specified code.

Coding Committee October 2019

### Q95 Balanced rearrangements and structural markers, not elsewhere classified

Q952 or Q953 BALANCED REARRANGEMENTS IN ABNORMAL INDIVIDUAL

If a case has a balanced translocation disrupting one gene which leads to a congenital anomaly, use both the specific congenital anomaly code and Q878 (other specified syndrome). The MCA algorithm will then recognize the case as having a genetic origin.

Coding Committee April 2022

### 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

CODING OF UNIPARENTAL DISOMY (UPD) AND METHYLATION DEFECTS

At present, there is no code for this. Will be coded as part of the syndrome. Write the result in the genetic test variable.

Coding Committee October 2019

**Outside Q-chapter:**

D180 is the correct code for haemangiomas

D181 is the correct code for lymphangiomas

L059 is the correct code for sacral dimple.

Coding Committee October 2024

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

P354 CONGENITAL ZIKA VIRUS

The code P354 (previously P358) to be used for all cases exposed to zika virus infection in pregnancy. Code P354 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 A925 – new specific code for "zika virus disease". These codes to be used only for zika virus.

Coding Committee December 2016, adapted by the Coding Committee April 2022

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

P023 TRAP sequence

Add this code if the anomalies for the twin reported to EUROCAT are explained by TRAP sequence.

Coding Committee November 2022

G710 WALKER-WARBURG SYNDROME

Walker Warburg = congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies = G710, not Q878, do not include as EUROCAT case.

Coding Committee April 2022

## 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