3.5 Detailed Congenital Anomaly Coding Guidelines

Remember always to give as specified code as possible

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 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

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

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

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

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

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

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

Q20 Congenital malformations of cardiac chambers and connections

Q204 SINGLE VENTRICLE, COMMON VENTRICLE, DOUBLE INLET LEFT VENTRICLE, COR TRILOCULARE BIATRIATUM
A single ventricle has absence or near total 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

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 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

TETRALOGY OF FALLOT
The ICD10-code for Tetralogy of Fallot is Q213. Do not use other additional cardiac codes for this malformation.
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

Q22 Congenital malformations of pulmonary and tricuspid valves

Q23 Congenital malformations of aortic and mitral valves

Q24 Other congenital malformations of heart

Q25 Congenital malformations of great arteries

Q26 Congenital malformations of great veins

Q27 Other congenital malformations of peripheral vascular system

Q28 Other congenital malformations of circulatory system

Q30 Congenital malformations of nose

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

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

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

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

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

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

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

Q61.40 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.

61.41 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.

Q61.8 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

Q61.9 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

HYDRONEPRHOSIS
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

Q63 Other congenital malformations of kidney

Q64 Other congenital malformations of urinary system

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

Q65 Congenital deformities of hip

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

Q72 Reduction defects of lower limb

Q73 Reduction defects of unspecified limb

Q74 Other congenital malformations of limb(s)

Q75 Other congenital malformations of skull and face bones

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

Q75.4 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

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

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 Q75.4 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

Q 8708 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

Q89 Other congenital malformations, not elsewhere classified

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:
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
Q24.9 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

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

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