Statement of EUROCAT and ICBDSR on ICD11

SUMMARY

EUROCAT (www.eurocat.ulster.ac.uk) and the ICBDSR (www.icbdsr.org) are the two international networks responsible for birth defect surveillance worldwide.

Congenital Anomaly surveillance is based on the ICD coding system. We identify in this statement which changes in moving from ICD10 to ICD11 can improve surveillance in future, and which aspects of ICD10 are important to maintain in any future coding system.

Main recommendations are:

a) that structural malformations and chromosomal anomalies continue to form a single chapter of ICD11, concerning congenital conditions diagnosed during pregnancy or in infancy.

b) that ICD11 coding should incorporate further clarity about the severity of the anomaly wherever possible, so that major and minor malformations can be distinguished, as well as unilateral and bilateral forms of anomaly.

c) that ICD11 retain the hierarchical nature of ICD10

d) that a number of specific coding changes be incorporated, including updating the coding for chromosomal anomalies, incorporating more specific coding for monogenic syndromes involving structural malformations, correcting incorrect use of the term “syndrome” and attending to other specific coding inaccuracies.

BACKGROUND

Purpose of the ICD coding system:

"The ICD has become the international standard diagnostic classification for all general epidemiological and many health management purposes. These include the analysis of the general health situation of population groups and monitoring of the incidence and prevalence of diseases and other health problems in relation to other variables such as the characteristics and circumstances of the individuals affected.

It is used to classify diseases and other health problems recorded on many types of health and vital records including death certificates and hospital records. In addition to enabling the storage and retrieval of diagnostic information for clinical and epidemiological purposes, these records also provide the basis for the compilation of national mortality and morbidity statistics by WHO Member States."

Since the thalidomide disaster in the 1960s, population-based registries of congenital anomaly have been established worldwide for the main purpose of surveillance to detect any increases in frequency of one or more types of anomaly which may be due to new or changing exposures, whether drugs or other environmental exposures. Spatial surveillance detecting geographical clustering or variation is also carried out. Registries have also in the last few decades been active in evaluating prenatal screening programmes, in terms of proportion of cases of congenital anomaly detected, and impact of terminations of pregnancy on livebirth prevalence rates. In addition, congenital anomaly registries are used to obtain data on effectiveness of prevention of congenital anomalies and for planning and measuring quality of care of affected individuals. The coding system for congenital anomalies needs to support the work of health care providers, public health workers and registries worldwide. The most basic need for ICD coding of malformations is to appreciate the extent of related mortality and morbidity, compare between countries and over time and to inform healthcare provision and policy decisions. The ICD code is often used in hospitals for finance

purposes, but it is not the main system for this (e.g. Snomed is increasingly used), and this is not the focus of this Statement.

CODING AND CLASSIFICATION NEEDS OF CONGENITAL ANOMALY REGISTERS

A high quality coding and classification system is crucial for surveillance of congenital anomalies

CODING SYSTEM

The purpose of coding congenital anomalies in EUROCAT and ICBDSR registries is to summarise unstandardised written text in such a way that the data can be analysed for surveillance and research purposes. Registries can encourage, but can rarely impose use of standard definitions and diagnostic tests and clinical follow-ups, and the coding system must allow for different levels of precision and accuracy of information provided by clinicians. Although coding should be done by experts, the reality across the world will be that the coding system should also facilitate accurate coding by less expert coders.

CLASSIFICATION SYSTEM

The purpose of a "classification" system for surveillance is to group together anomalies which share aetiologic, pathogenetic or clinical characteristics. There is a balance to be struck a) between "lumping" together heterogeneous sets of anomalies and "splitting" so finely that there are few cases in each group and b) between creating groups based on great precision and accuracy of diagnosis and coding and creating groups which take into account what can be realistically found in medical records for most cases.

A classification system does not theoretically need to be reflected in the coding system as it can regroup unrelated codes. However, practically a hierarchical coding system that reflects important aspects of classification is easier to implement, as it allows differing amounts of detail to be incorporated (e.g. anomaly of limb, limb reduction, absence of

hand) according to availability of detail in different countries and types of record, and it groups related anomalies together (in ICD10, an inconsistency is the separation of codes for neural tube defects). It also "educates" the coder as to the differential diagnosis of different malformations (e.g. under orofacial clefts, one immediately sees that there is a difference between cleft palate only and cleft lip with cleft palate.

THE CONGENITAL MALFORMATION CHAPTER

This chapter in ICD11 should concern conditions in which structural malformations arising during *in utero* development are the major clinical presentation, whatever their aetiology. These malformations are present during prenatal life and at birth, and are potentially diagnosable given appropriate technology within the first year of life, but may not be diagnosed in some health care systems until later, or never. Health and health care policy needs to group all these conditions together because:

a) initial responsibility for screening and diagnosis lies within two specific medical disciplines across the world: obstetrics and paediatrics, which coordinate their work in this area

b) most malformations potentially relate to environmental exposures acting during early pregnancy, many of these resulting in multiple types of congenital malformation in the same or different individuals and depending on timing of exposure (e.g rubella) and needing coherent health policies.

Chromosomal anomalies are included in the congenital malformation chapter and should remain there in ICD11. Many have structural malformations as their main presentation e.g. Trisomy 13 and 18. Most are diagnosed in early infancy due to structural malformations or dysmorphic features e.g Down Syndrome, although their main clinical problems (eg. mental retardation) may not yet be evident at the time of coding. Others e.g. Klinefelters Syndrome, may usually not have a structural malformation, but are increasingly commonly diagnosed due to prenatal and postnatal karyotyping and thus are best coded with other conditions dealt with by the same care and genetic counseling system.

In some countries the underlying chromosomal anomaly or Mendelian syndrome for a structural malformation may not be diagnosed, or diagnosed later, so it is important to keep the structural malformations together for comparative purposes. In this chapter in ICD11, we propose to include Mendelian syndromes commonly associated with structural malformations.

Brain anomalies are a difficult area. Brain development continues during late pregnancy and until two years of age, and therefore disorders of brain development are not entirely congenital. Brain malformations may underlie neurological and other developmental conditions eg. Cerebral palsy, autism, mental retardation. It is not possible to diagnose and code mental retardation and cerebral palsy in newborns. However, the structural brain malformation, if diagnosed, should be coded within the malformation chapter.

Many diseases in other chapters have a congenital or genetic origin, whether or not they are diagnosed in infancy. We propose retaining in the chapter only those conditions often presenting with a structural malformation, but giving additional guidance notes regarding other conditions (e.g. metabolic disorders, congenital deafness) which are present at birth and diagnosed in infancy and should be included in a wider assessment of "birth defects". The possibility of regrouping these other conditions in a single additional chapter should also be assessed.

SEVERITY

One of the most difficult problems for congenital anomaly surveillance is the distinction of "major" and "minor" anomalies. While either may indicate underlying teratogenic exposures, minor anomalies are inconsistently diagnosed (thus creating "noise" in the surveillance system) and have in themselves little functional or health consequence. EUROCAT has a list of such minor anomalies for exclusion (where they are not

associated with major anomalies). ICD11 should seek to make sure that all minor anomalies have specific codes that can be distinguished from major anomalies, and excluded where necessary depending on the purpose of analysis. Recording of minor anomalies can be useful for diagnostic purposes. Further we propose a subchapter with codes for dysmorphic features.

A related problem to that of “major” and “minor” is the severity of the congenital malformation, which is important for describing public health impact. Generally ICD10 does not incorporate severity grading, except where this is integral to the description of the malformation (e.g. absence of hand). We propose some additions for ICD11 to incorporate severity grading for malformations which vary in severity. Unilateral and bilateral should be distinguished as a measure of severity. Size of the dilatation of the renal pelvis in hydronephrosis or of the ventricular system in hydrocephalus are other examples.

MULTIPLE CODING

ICD11 should have guidance notes for those users who are only able to use one code to describe a case (e.g. for some death certificates).

However, the possibility of giving multiple codes would be the recommended norm. The ICD11 system should seek where possible to allow cases to be classified according to phenotype (eg. omphalocele of any origin, or congenital cataract of any origin) and aetiology (eg. exclude omphalocele of chromosomal origin, separate congenital rubella involving cataract).

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Part 2:

Specific problems related to coding of congenital malformations

ICD10 codes given as a reference

Including principles from ICD/BPA10 and Paediatric Cardiology codes (AEPC)

Q03: Hydrocephalus – Dandy-Walker

Not all cases with a Dandy Walker malformation have hydrocephalus (approx 75%). Using the ICD10 Dandy-Walker cases coded with a hydrocephalus code may have Dandy-Walker malformation only and not hydrocephalus. In general we find the present ICD10 Q03 chapter (hydrocephalus) problematic.

Q03 and Q048: Hydrocephalus – ventriculomegaly

Hydrocephalus is defined as dilatation of ventricular system, not due to primary atrophy of the brain, with or without enlargement of the skull. The term ventriculomegaly is often used for prenatal diagnosis of ventricular enlargement. There is no specific ICD10 code for ventriculomegaly but most EUROCAT registries use the code Q048. For ICD11 it is important to have clear definitions of hydrocephalus and ventriculomegaly and specific codes for both.

Q04: Other malformations of the brain

MRI scans often give diagnosis of brain malformations that only can be coded as Q048 other specified brain malformations. ICD11 should be developed according to possible MRI scan diagnosis

Need to separate cerebellar vermis and hemisphere anomalies as this is aetiologically relevant.

Also a specific code for neuronal migration defect

Q00, Q01, Q05

It would be very valuable if all neural tube defects were coded together using the same prefix

Q05 and Q070: Spina bifida – Arnold Chiari

In ICD/BPA9 there was a specified code for spina bifida with Arnold Chiari malformation. This code does not exist in ICD10. We have seen that at local level spina bifida with Arnold Chiari is coded only with the code for Arnold Chiari Q070. In this way the spina bifida is not recorded by the code.

Q210: VSD – muscular and perimembraneous

In recent years many small muscular VSDs with early spontaneous closure are being diagnosed. A perimembraneous VSD is a much more important health problem as these cases more often needs surgery and more frequently have associated malformations, syndromes or karyotype anomalies. In ICD10 there is only code for both conditions: Q210. For ICD11 there should be a specific code for both types of VSDs

Q211: ASD – persistent foramen ovale

With the increasing use of echocardiography in neonatology it is evident that many newborns have a persistent foramen ovale for months and this without any clinical impact. It is important to have specific codes for both ASD secundum and foramen ovale. Otherwise health statistics on congenital heart disease will be inflated by a high number of persistent foramen ovale and these are not a cardiac malformation

Q22 and Q23: Cardiac valve atresias and stenosis should be separated

Pulmonary valve atresia and pulmonary valve stenosis can be coded separately in ICD. For the aortic valve, the mitral valve and the tricuspid valves atresia and stenosis have the same code. As there is large clinical difference between a cardiac valve atresia and a

minor cardiac valve stenosis, all four cardiac valves should have a specific code for atresia and a specific code for stenosis.

Q336: Lung hypoplasia/dysplasia

Congenital lung hypoplasia is often secondary to severe renal malformations with oligohydramnios. Further, long-term amniotic leakage also results in lung hypoplasia of the newborn. For aetiological purposes and for optimal mortality statistics it is important to be able to discriminate between primary and secondary lung hypoplasia. A code for secondary lung hypoplasia may be classified together with neonatal diseases.

Q400: Pyloric stenosis

This condition is aquired and not congenital. The code should therefore be classified as such. It is still important to have a specific code for infantile pyloric stenosis, but outside the chapter.

Q54: Hypospadia

Q540 codes for both balanic that is considered a minor form of hypospadia and for coronal for which the patients usually are referred for surgery. The coding in ICD11 should be changed so that balanic and coronal hypospadia have different codes.

Q620: Hydronephrosis.

This condition is often diagnosed prenatally and for many cases the dilatation resolves spontaneously after birth. Further the dilatation of the renal pelvis may be caused by either a stenosis or by ureteral reflux. EUROCAT has defined congenital hydronephrosis as: “Obstruction of the urinary flow from kidney to bladder. Only if renal pelvis is 10 mm or more after birth” For ICD11 coding of congenital hydronephrosis should be based on a definition of stenosis and with the possibility of coding severity based on size and uni/bilateral

Q627 Congenital vesicu-uretero-renal reflux

This condition is not a structural anomaly and may also be present for longer periods after urinary tract infections in infancy. The code does not belong in a chapter of structural malformations.

Q660 Club feet

Separate codes for unilateral club foot and bilateral club feet

Q70 Syndactyly

Codes for syndactyly differentiating between 2-3 and non-2-3 syndactyly of the foot.

Q71-73 Limb reduction defects

Transverse limb anomalies need a code

Q774: Achondroplasia / hypocondroplasia

These two skeletal dysplasias are clinically very different and both should have their own specific code

Q79: Abdominal wall defects

Abdominal wall defects are clinically well-defined groups of malformations. For gastroschisis there is a clear increasing trend in many regions and many studies are being done for aetiological purposes. They are an important group for surveillance.
At the moment they are classified under musculo-skeletal anomalies not elsewhere classified in ICD10. EUROCAT has proposed to WHO to classify abdominal wall defects under gastro-intestinal malformations (for ICD10 as chapter Q46.

Q86: Malformation syndromes due to known exogenous causes, not elsewhere classified

Fetal alcohol syndrome and two drugs (warfarin and hydantoin) have their own codes. Other important drug syndromes are missing (anti-epileptic drugs, thalidomide) Malformations as part of maternal infections and other teratogenic syndromes are important for aetiology surveillance.

Q87: Syndromes

In general this section should include syndromes that are frequently diagnosed in infancy. It is important with separate codes for those of known genetic origin and for those with unknown (but presumed single) etiology.

The distinction requested here may be more to separate those syndromes of single gene aetiology since only a handful now do not have a known aetiology (eg Kabuki). The logic here being that the non-mendelian ones could be environmentally sensitive and so worth monitoring etc. Also, the non-inherited (new dominant) syndromes need monitoring.

The syndrome part of ICD10 now includes sequences that per definition are isolated malformations and not syndromes.

The name “syndrome” should only be given to those diagnoses that fulfill the syndrome criteria. Sequences (Pierre- Robin, Moebius, sirenomelia) and structural anomalies (hypoplastic left heart syndrome) should not be given the name syndrome in ICD11

Q897: multiple congenital malformations

Using this code cases reported cannot be classified further. This code is only really useful where only one code is allowed i.e. not useful for registries, but perhaps for vital records.

Q90-99: Chromosomal anomalies.

With the advances in technology for detection of chromosomal anomalies and microdeletions a clear distinction between syndrome and chromosomal syndrome is important. We think that chromosomal syndromes are those which affect more than one gene.

We propose that microdeletions are coded by their clinical syndrome name and then a specific code for a diagnosed microdeletion is added [i.e. a single or few codes covering all microdeletions under a certain size. Not a code for each type of microdeletion].

Conditions without a present ICD10 code.

Cystic hygroma

This condition is an important prenatal marker of congenital malformations. Terminations of pregnancy are performed after prenatal diagnosis of cystic hygroma without further diagnostics. Therefore it is important to have a specific code for this within the malformation chapter.

Congenital sacral teratoma needs a specified code

Hydrocephalus secondary to neonatal conditions:

In ICD10 hydrocephalus can be coded as congenital (Q03) or acquired (G91). This gives a problem for coding of hydrocephalus after neonatal diseases (preterm birth, infections) as these conditions are insults to the developing brain. Therefore some may code as acquired hydrocephalus and others as congenital hydrocephalus. Clear definitions should be given and the optimal situation was a code in the neonatal chapter for acquired hydrocephalus in infancy

Part 3: Proposed coding system for ICD11

The ICD-10 system of letters designating specific chapters has been very successful and a great improvement over the ICD-9 system of numbers only. To conserve this element for ICD-11 would be ideal as long as the system is clearly distinguishable from those used before.

A proposed method would be to keep the ICD-10 chapter letters as in:

But use a second letter for each subheading before numbers for specific anomalies coded continuing the 'tree' system of additional numbers for additional detail. This would allow up to 26 subheadings rather than the previous 9.

Thus, for congenital anomalies the subheadings could be:

QA CNS
QB NTD
QC Eyes
QD Ears
QE CHD
QF Respiratory system
QG Cleft lip and cleft palate
QH Digestive system
QJ Abdominal wall defects
QK Genital organs
QL Urinary tract
QM Neuromuscular
QN Skull, face, spine and bony thorax,
QP Limb defects
QQ Skeletal dysplasias

QR Skin, breast and integument

QS Mendelian (single gene) Syndromes

QT New dominant and non-mendelian syndromes

QV Syndrome features

QW Miscellaneous congenital anomalies, not elsewhere classified

QX Chromosome abnormalities