3.4 Multiple Congenital Anomaly Algorithm

Multiple anomaly flow-chart for monitoring of multiple anomalies
At the moment, this should be for the Central Database only.

Definition of a multiple congenital anomaly case (MCA):
Two or more unrelated major structural malformations that cannot be explained by an underlying syndrome or sequence

This means that the process of the flowchart is to find cases with two or more codes within the Q chapter, unless the case is transferred to other groups according to the steps described below.

Name for groups:
C: chromosomal
B: genetic syndrome, skeletal dysplasia and monogenic disorder
N: NTD isolated
A: isolated cardiac
R: isolated renal
I: isolated other
O: non-syndrome outside malformation chapter
M: potential multiple anomalies
T: teratogenic syndrome

Minor, unspecified and invalid codes.
The following codes are ignored in the flowchart, but appear in individual case output:

Guide 1.4 list of minors post 2005 to be used for all years
Balanced chromosomal rearrangements (7584 or Q95) as the only code
Multiple Malformation code (7597 or Q897)
Unspecified malf code (7599 or Q899)
No valid ICD code

Group X contains cases with only the above-listed codes.

Outside Q-chapter codes (except the few codes accepted in "all anomalies")
These codes are ignored by the flowchart process but appear in the individual case output

Accepted non Q codes: D215, D821, D1810, P350, P351, P371

ICD/BPA9: 27910, 2281, 76076, 76280, 7710, 7711, 77121

The flow-chart

For 3 and 4 digit codes mentioned here, the coding also includes the codes with more digits
Only Q-codes are valid for the process after step 2
This is a hierarchical procedure

Step 1

Exclude all cases with a chromosomal code
ICD/BPA9: all 759 cases except 7584
Q90-Q93 excluding Q936
Q96-Q99, • Transfer to group C

Step 2

Exclude all cases with genetic syndrome codes, skeletal dysplasia and congenital skin disorder codes
ICD/BPA9: 7598, 27910, 7571, 7573, 75581 (Larsen syndrome), 75601 (Crouzon), 75604 (Mandibulofacial dysostosis)
No ICD9 subgroup for skeletal dysplasia
Excluding 759801,759895, 759844
Q87 Excluding Q8703, Q8704, Q8706, Q8708, Q8724, Q8726,
Q936, D821,
Q77, Q7800, Q782-788, Q7402
Q80-Q82
Q4471 Alagille syndrome, Q6190 Meckel-Gruber, Q7484 Larsen syndrome
Q751 Crouzon /craniofacial dysostosis, Q754 Mandibulofacial dysostosis (Treacher Collin)
Q7581Frontonasal dysplasia • Transfer to group B

Step 3

Exclude all cases with a code for teratogenic syndrome code
No ICD9 subgroup for teratogenic syndromes, only the codes for infection 7710, 7711, 77121 and fetal alcohol syndrome 76076
Q86, P350, P351, P371 • Transfer to group T

Step 4

Exclude all cases with a heterogenous syndrome code
ICD9: 756110, 75680, 759611, 3568 (not exactly the same as ICD10)
Q761, Q7982, Q8581, Q8706 • Transfer to group M

Step 5

Exclude all cases with only NTD codes
ICD/BPA9: 740-741, 7420
Q00-Q01, Q05 • Transfer to group N

Step 6

Exclude all cases with codes only in cardiac chapter
ICD/BPA9: 745-746, 7470-7474
Q20-Q26 • Transfer to group A

Step 7
Exclude all cases with codes only in renal chapter
ICD/BPA9: 753, 756.72, 752.61
Q60 – Q64, Q794 • Transfer to group R

Step 8
Exclude all cases with **only one code** within Q chapter
Include known local coding variations/errors
(for our own purpose, transfer also cases with only cleft codes – chapter Q35-Q37)
ICD/BPA9: 740-759 (transfer to groups like ICD10 – see codes for group N, A, and R earlier)
If Q00-Q01, Q05 • Transfer to group N
If Q20-Q26 • Transfer to group A
If Q60-Q64, Q794 • Transfer to group R
If only one other Q-code or D1810 or D215/icd9 2281 • Transfer to group I

Step 9
Exclude all cases with codes only in eye chapter
ICD/BPA9: 743
Q10-Q15 • Transfer to group I

Step 10
Exclude all cases with codes only with limb reduction defects
ICD/BPA9: 7552-7554
Q71-Q73 • Transfer to group I

Step 11
Exclude all cases with codes only for hypospadias
ICD/BPA: 75260
Q54 • Transfer to group I

Step 12
Exclude all cases with codes only for polydactyly
ICD/BPA9: 7550
Q69 • Transfer to group I

Step 13
Exclude all cases with codes only for reduction defects of brain
ICD/BPA9: 7422
Q04 • Transfer to group I

Step 14
Exclude all cases with codes only for hip anomalies
ICD/BPA9: 75430
Q65 • Transfer to group I

Step 15
Exclude all cases with codes only for syndactyly
ICD/BPA9: 7551
Q70 • Transfer to group I

Step 16
Exclude all cases with codes only for syndactyly + polydactyly
ICD/BPA9: 7550 and 7551
Q69 and Q70 • Transfer to group I

Step 17
Exclude all cases with codes only for small intestinal atresia
ICD/BPA9: 75111, 75112, 75119
Q41 • Transfer to group I

Step 18
Exclude all cases with codes only for facial clefts
ICD/BPA9 7490, 7491, 7492
Q35, Q36, Q37 • Transfer to group I

Step 19
Exclude all cases with the code for balanced chromosomal rearrangements (Q95 or 7584) and only one other Q-code
ICD/BPA9: 7584 (transfer to groups like ICD10 – see codes for group N, A, and R earlier)
Q95
If (Q00-Q01, Q05) and Q95 • Transfer to group N
If Q20-Q26 and Q95 • Transfer to group A
If (Q60-Q64, Q794) and Q95 • Transfer to group R
If only one other Q-code and Q95 • Transfer to group I

Step 20
Exclude all cases with only outside Q chapter codes (without Q-codes)
ICD/BPA9: outside 740-759
2281 and 76280 accepted as outside malformation chapter codes
Not beginning with Q
D1810 and D215 accepted as outside Q-code • Transfer to group O

Step 21
Exclude all known sequences or combinations of anomalies without other anomaly codes
(NB: Anyone of these codes may be used more than once – disregard duplicate codes)
Spina bifida – talipes – hydrocephalus:
ICD/BPA9 741 coded with 7545 and/or 7423
Q05 coded with Q66 and/or Q03 • Transfer to group N

Bilateral renal aplasia/dysplasia – lung hypoplasia - talipes:
ICD/BPA9 75300 coded with 74851 and/or 7545
Q601/Q606 coded with Q336 and/or Q66 • Transfer to group R

Omphalocele/gastroschisis – malrotation of gut – small intestinal atresia
ICD/BPA9 75670/75671 coded with 7514 and/or 7511
Q792/Q793 coded with Q433 and/or Q41 • Transfer to group I

Anal atresia - rectovaginal fistula
ICD/BPA9 7512 coded with 75242
Q42 coded with Q522 • Transfer to group I

Diaphragmatic hernia – lung hypoplasia
ICD/BPA9 75661 coded with 74851
Q790 coded with Q336 • Transfer to group I

Anencephalus - adrenal hypoplasia
ICD/BPA9 740 coded with 75911
Q00 coded with Q891 • Transfer to group N

Unspecified hydrocephalus - reduction defect of brain
ICD/BPA9 74239 coded with 7422
Q039 coded with Q04 • Transfer to group I

Unspecified hydrocephalus – Arnold-Chiari
no ICD/BPA9 code for Arnold-Chiari
Q039 coded with Q070 • Transfer to group I

NTD – Arnold Chiari
no ICD/BPA9 code for Arnold-Chiari
Q01 or Q05 coded with Q070 • Transfer to group N

Amniotic band sequence
ICD/BPA9: 76280
All cases with the code Q7980 • Transfer to group I

Caudal dysplasia sequence
no ICD/BPA9 code for caudal dysplasia sequence
All cases with the code Q8980 • Transfer to group I

Sirenomelia sequence
ICD/BPA9: All cases coded with 759844
All cases coded with Q8724 • Transfer to group I

Cyclops sequence
ICD/BPA9: All cases coded with 759801
All cases coded with Q8703 • Transfer to group I

Pierre Robin sequence
ICD/BPA9: All cases coded with 756030 as only code or with 7490, 7491, 7492
All cases coded with Q8708 as only code or with Q35-Q37 • Transfer to group I

Holoprosencephaly – median cleft lip
ICD/BPA9: All cases coded with 74226 and 74912
All cases coded with Q042 and Q361 • Transfer to group I

Step 22

The remaining cases are group M: potential multiple anomalies. Manual evaluation of all remaining cases before final inclusion into multiple anomaly group - or inclusion in one of the other groups.

Notes:

Then need to output group M cases as individual case lists with text description of anomalies as well as codes plus variables: ID no, registry, year of birth, type of birth, twin, GA, BW, karyotype (including written text), postmortem examination, when discovered

For the website review of potential multiple cases, a subgroup for “poorly specified cases” has to be added (could go to group X)

Ester Garne

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