3.4 Multiple Congenital Anomaly Algorithm for monitoring of multiple anomalies

Definition of a multiple congenital anomaly case (MCA): Two or more unrelated major congenital anomalies in different organ systems that cannot be explained by an underlying syndrome or sequence.

The aim of the multiple congenital anomaly algorithm is to find cases with two or more codes within the ICD10 Q chapter, unless the case is transferred to other groups according to the steps described below.

Currently the MCA algorithm is intended to be used only at the Central Registry level (implemented in DMS and used after the data submission of March 2023).

Name for groups:
G: genetic disorders (genetic syndromes, hereditary skin disorders, skeletal dysplasias and chromosomal anomalies)
N: isolated NTD
A: isolated cardiac
R: isolated renal
I: isolated other
O: non-syndrome outside malformation chapter (teratogenic syndromes without a major congenital anomaly are included here)
M: potential multiple congenital anomalies
T: teratogenic syndromes resulting in major malformations
X: minor, unspecified and invalid codes

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

Guide 1.5 list of minors post 2005 to be used for all years (version dated 31.05.2022)
Balanced chromosomal rearrangements (7584 or Q95) as the only code
Multiple Malformation code (7597 or Q897)
Unspecified malformation code (7599 or Q899)
No valid ICD code

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

The following codes are accepted codes for major congenital anomalies:
Accepted major ICD9/BPA codes: all codes within 740-759, which have not been specified as minor by EUROCAT and 2377, 27910, 2281.

Accepted major ICD10/BPA codes: all Q codes, which have not been specified as minor by EUROCAT and D215, D821 and D1810.

Table: Multiple congenital anomaly algorithm

Steps*	Rule	ICD9-BPA codes**	ICD10-BPA codes**	Transfer to:
Step 0	Exclude all cases with only non-accepted ICD9/BPA or ICD10/BPA codes	outside 740-759, except 2377, 27910 and 2281 which are accepted as outside malformation chapter codes	outside Q chapter, except D215, D821 and D1810 which are accepted as outside Q-codes	**Group O:** non-syndrome: outside malformation chapter (teratogenic syndromes without a major congenital anomaly are included here)
	Exclude all cases with only minor, unspecified and invalid codes	only minor codes, and/or 7584 and/or 7597 and/or 7599	only minor codes, and/or Q95, and/or Q897 and/or Q899	**Group X**: minor, unspecified and invalid codes
Step 1	Exclude all cases with a genetic disorder code	2377, 27910, 751653, 755551, 75581, 75601, 75604, 7564, 75650-75659, 75685, 7571, 75730, 75732-75736, 7580-7583, 7585-7589, 75934, 7595, 75961, 7598 except 75862, 759801, 759844, 759895	D821, Q4471, Q6190, Q7402, Q7484, Q751, Q754, Q7581, Q77, Q780-Q789, Q796, Q800-Q824, Q8282, Q8283, Q850, Q851, Q8581, Q87, Q8934, Q90-Q93, Q96-Q99, except Q8703, Q8704, Q8706, Q8708, Q8724, Q8726	**Group G**: genetic disorders (genetic syndromes, hereditary skin disorders, skeletal dysplasias and chromosomal anomalies)
Step 2	Exclude all cases with a teratogenic syndrome code and at least one other accepted major IC9/BPA or ICD10/BPA code	7607, 7608, 7710, 7711, 77121	Q86, P350, P351, P354, P358, P371	**Group T**: teratogenic syndromes resulting in major malformations
Step 3	Exclude all cases with the VACTERL association code	759895	Q8726	**Group M**: potential multiple congenital anomalies
	Exclude all cases with the OAVS / Goldenhar syndrome code	75606	Q8704	

Steps*	Rule	ICD9-BPA codes**	ICD10-BPA codes**	Transfer to:
	Exclude all cases with polycystic kidney codes	75311-75313	Q611-Q613	
Step 4	Exclude all cases with codes only for neural tube defects	740, 741, 7420	Q00, Q01, Q05	**Group N**: isolated NTD
Step 5	Exclude all cases with codes only for congenital heart defects	745, 746, 7470-7474	Q20-Q26	**Group A**: isolated cardiac
Step 6	Exclude all cases with codes only for congenital anomalies of kidney and urinary tract (CAKUT)	75261, 753, 75672	Q60-Q64, Q794	**Group R**: isolated renal
Step 7	Exclude all cases with only one accepted major ICD9-BPA or ICD10-BPA code			**Group I**: isolated other
Step 8	Exclude all cases with the code for cystic hygroma (2281 or D1810) and only one other accepted major ICD9/BPA or ICD10/BPA code	If (740, 741, 7420) and 2281	If (Q00, Q01, Q05) and D1810	**Group N**: isolated NTD
		If (745, 746, 7470-7474) and 2281	If (Q20-Q26) and D1810	**Group A**: isolated cardiac
		If (75261, 753, 75672) and 2281	If (Q60-Q64, Q794) and D1810	**Group R**: isolated renal
		If only 2281, or 2281 and one other accepted major ICD9/BPA code	If only D1810, or D1810 and one other accepted major ICD10/BPA code	**Group I**: isolated other
Step 9	Exclude all cases with codes only for nervous system anomalies, not NTD	7421-7429, 759801	Q02-Q04, Q06-Q07, Q8703	**Group I**: isolated other
Step 10	Exclude all cases with codes only for eye anomalies	743	Q10-Q15	**Group I**: isolated other
Step 11	Exclude all cases with codes only for ear anomalies	7440-7443	Q16-Q17	**Group I**: isolated other

Steps*	Rule	ICD9-BPA codes**	ICD10-BPA codes**	Transfer to:
Step 12	Exclude all cases with codes only for respiratory anomalies	7480, 74833-74835, 7484, 74850, 74852, 74858, 7486, 7488	Q300, Q32-Q34	**Group I**: isolated other
Step 13	Exclude all cases with codes only for oro-facial clefts	7490, 7491, 7492	Q35-Q37	**Group I**: isolated other
Step 14	Exclude all cases with codes only for small intestinal atresia	75111, 75112	Q411-Q418	**Group I**: isolated other
Step 15	Exclude all cases with codes only for genital anomalies	7520-7524, 75260, 7527-7529	Q50-Q52, Q54-Q56, except Q518	**Group I**: isolated other
Step 16	Exclude all cases with codes only for limb anomalies	7543-7548, 755	Q65-Q74	**Group I**: isolated other
Step 17	Exclude all cases with code for conjoined twins	7594	Q894	**Group I**: isolated other
	Exclude all cases with code for cyclopia	759801	Q8703	
Step 18	Exclude all cases with the code for balanced chromosomal rearrangements (7584 or Q95) and only one other accepted major ICD9/BPA or ICD10/BPA code	If (740, 741, 7420) and 7584	If (Q00, Q01, Q05) and Q95	**Group N**: isolated NTD
		If (745, 746, 7470-7474) and 7584	If (Q20-Q26) and Q95	**Group A**: isolated cardiac
		If (75261, 753, 75672) and 7584	If (Q60-Q64, Q794) and Q95	**Group R**: isolated renal
		If only one other accepted major ICD9/BPA code and 7584	If only one other accepted major ICD10/BPA code and Q95	**Group I**: isolated other
Step 19	Exclude all known sequences or combinations of anomalies without other anomaly codes *(NB: Any one of these codes may be used more than once – disregard duplicate codes):*			
	Spina bifida – central nervous system anomalies – talipes – hip dislocation – congenital hydronephrosis	741 coded with (7422-7429 and/or 7545	Q05 coded with (Q03, Q04, Q06-Q07 and/or	**Group N**: isolated NTD

Steps*	Rule	ICD9-BPA codes**	ICD10-BPA codes**	Transfer to:
		and/or 7543 and/or 75320)	Q66 and/or Q65 and/or Q620)	
	Anencephaly – adrenal anomaly	740 coded with 7591	Q00 coded with Q891	**Group N**: isolated NTD
	Bilateral renal agenesis/dysplasia – lung hypoplasia – talipes	75300 coded with (74851 and/or 7545)	Q601/Q606 coded with (Q336 and/or Q66)	**Group R**: isolated renal
	Gastroschisis/omphalocele – anomalies of intestinal fixation – small intestinal atresia	75671/75670 coded with (7514 and/or 7511)	Q793/Q792 coded with (Q433 and/or Q41)	**Group I**: isolated other
	Ano-rectal atresia and stenosis – rectovaginal fistula	7512 coded with 75242	Q42 coded with Q522	**Group I**: isolated other
	Diaphragmatic hernia – lung hypoplasia – anomalies of intestinal fixation	75661 coded with (74851 and/or 7514)	Q790 coded with (Q336 and/or Q433)	**Group I**: isolated other
	NTD – Arnold Chiari malformation – tethered cord	No ICD9/BPA code for Arnold-Chiari malformation or tethered cord	Q01/Q05 coded with (Q070 and/or Q068)	**Group N**: isolated NTD
	Amniotic band sequence code and at least one other accepted major ICD9/BPA or ICD10/BPA code	All cases with the code 76280 and at least one accepted major ICD9/BPA code	All cases with the code Q7980 and at least one accepted major ICD10/BPA code	**Group I**: isolated other
	Poland anomaly – symbrachydactyly – aplasia pectoral muscle	75680 coded with (7551 and/or 76581)	Q7982 coded with (Q70 and/or Q7480 and/or Q7981)	**Group I**: isolated other
	Caudal regression sequence	No ICD9/BPA code for caudal regression sequence	All cases with the code Q8980	**Group I**: isolated other

Steps*	Rule	ICD9-BPA codes**	ICD10-BPA codes**	Transfer to:
	Sirenomelia sequence	All cases coded with 759844 (sirenomelia/caudal regression)	All cases coded with Q8724	**Group I**: isolated other
	Pierre Robin sequence	All cases coded with 75603-as the only code or with 7490	All cases coded with Q8708 as the only code or with Q35	**Group I**: isolated other
	Holoprosencephaly/arhinencephaly – oro-facial clefts – brain anomalies	74226 coded with (7490-7492 and/or 7421-7424)	Q042/Q041 coded with (Q35-Q37 and/or Q02-Q04)	**Group I**: isolated other
	Annular pancreas – atresia/stenosis of duodenum	75172 coded with 75110	Q451 coded with Q410	**Group I**: isolated other
Step 20	The remaining cases are **group M**: potential multiple congenital anomalies. Manual evaluation of all remaining cases will take place by the geneticists of the EUROCAT coding and classification committee. All cases will be manually allocated to either the multiple congenital anomaly group, or one of the other groups.			**Group M:** potential multiple congenital anomalies

Notes:

***This is a hierarchical procedure

**For 3 and 4 digit codes mentioned here, the coding also includes the codes with more digits. Only accepted ICD9/BPA and ICD10/BPA codes are valid for the process after step 2.

For the manual evaluation of group M the following is needed:
Output group M cases as individual case lists with text description of anomalies as well as codes plus variables: local ID number, registry, year of birth, type of birth, twin, gestational age, birth weight, karyotype (including written text), postmortem examination, when discovered.

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

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