

## **Supplementary Material**

10.1302/0301-620X.107B2.BJJ-2024-0213.R1

**Table i.** List of all major congenital anomalies and syndromes (defined in accordance with EUROCAT's classification of malformations: https://eu-rd-platform.jrc.ec.europa.eu/system/files/public/eurocat/Guide\_1.5\_Chapter\_3.3.pdf) that were assessed within the 1994 to 2011 Danish Perthes' disease cohort. The Danish (Classification of diseases and health-related conditions) ICD-10 codes (ICD-10-DK) (listed here: https://medinfo.dk/sks/brows.php?s\_nod=6199) were used to define the major congenital anomalies and syndromes investigated and are listed below. Conditions were assessed hierarchically in the order listed.

Order	Group	Description	Include ICD.10.DK	Exclude minor anomalies
0	DKpop	All infants born in DK	None	
		between 1994 and 2011		
		(subset of patients, born		
		after the implementation of		
		ICD-10, who have passed		
		their twelfth birthday)		
		included in the DNSB		
1	LCPD	Infants born in DK between	DM911, DM912, DM913	
		1994 and 2011 (subset of		
		patients, born after the		
		implementation of ICD-10,		
		who have passed their		

		twelfth birthday) included in the DNSB		
2	CHROM	Down syndrome (trisomy 21)	DQ90	
3	CHROM	Edwards syndrome (trisomy 18) & Patau syndrome (trisomy 13)	DQ91	
4	CHROM	Triploidy and polyploidy	DQ92	
5	CHROM	Monosomies and deletions from the autosomes, not elsewhere classified	DQ93	
6	CHROM	Turner syndrome	DQ96	
7	CHROM	Other sex chromosome abnormalities, female phenotype, not elsewhere classified	DQ97	
8	CHROM	Other sex chromosome abnormalities, male phenotype, not elsewhere classified	DQ98	
9	CHROM	Other chromosome abnormalities, not elsewhere classified	DQ99	
10	GEN	DiGeorge syndrome	DD821	
11	GEN	Alagille syndrome	DQ447B	
12	GEN	Meckel-Gruber syndrome	DQ619A	
13	GEN	Cleidocranial dysostosis	DQ740B	
14	GEN	Craniofacial dysostosis	DQ751	
15	GEN	Mandibulofacial dysostosis	DQ754	
16	GEN	Osteochondrodysplasia with defects of growth of tubular bones and spine	DQ77	

17	GEN	Other	DQ780-DQ789	
		osteochondrodysplasias		
18	GEN	Ehlers-Danlos syndrome	DQ796	
19	GEN	Congenital ichthyosis	DQ800-DQ824	
20	GEN	Neurofibromatosis	DQ850	
		(nonmalignant)		
21	GEN	Tuberous sclerosis	DQ851	
22	GEN	Sturge-Weber(-Dimitri) syndrome	DQ858C	
23	GEN	Other specified congenital malformation syndromes affecting multiple systems	DQ87	DQ870D, DQ870F, DQ870G, DQ870I, DQ872E, DQ872G
24	GEN	Kartagener's syndrome	DQ893F	
25	TERA	Congenital malformation syndromes due to known exogenous causes, not elsewhere classified	DQ86	
26	TERA	Congenital rubella syndrome	DP350	
27	TERA	Congenital cytomegalovirus infection	DP351	
28	TERA	Other congenital viral diseases	DP358	
29	TERA	Congenital toxoplasmosis	DP371	
30	CARD	Congenital malformations of cardiac chambers and connections	DQ20	
31	CARD	Congenital malformations of cardiac septa	DQ21	DQ211C
32	CARD	Congenital malformations of pulmonary and tricuspid valves	DQ22	

33	CARD	Congenital malformations of aortic and mitral valves	DQ23	
34	CARD	Other congenital malformations of heart	DQ24	DQ246
35	CARD	Congenital malformations of great arteries	DQ25	DQ254E
36	CARD	Congenital malformations of great veins	DQ26	DQ261
37	NERV	Anencephaly and similar malformations	DQ00	
38	NERV	Encephalocele	DQ01	
39	NERV	Microcephaly	DQ02	
40	NERV	Congenital hydrocephalus	DQ03	
41	NERV	Other congenital malformations of brain	DQ04	
42	NERV	Spina bifida	DQ05	
43	NERV	Other congenital malformations of spinal cord	DQ06	
44	NERV	Other congenital malformations of nervous system	DQ07	DQ078D, DQ078G
45	NERV	Cyclopia	DQ870D	
46	URO-RENAL	Renal agenesis and other reduction defects of kidney	DQ60	
47	URO-RENAL	Cystic kidney disease	DQ61	DQ610
48	URO-RENAL	Congenital obstructive defects of renal pelvis and congenital malformations of ureter	DQ62	DQ627
49	URO-RENAL	Other congenital malformations of kidney	DQ63	DQ633

50	URO-RENAL	malformations of urinary system	DQ64	
51	URO-RENAL	Prune belly syndrome	DQ794	
52	OFC	Cleft palate	DQ35	DQ357
53	OFC	Cleft lip	DQ36	
54	OFC	Cleft palate with cleft lip	DQ37	
55	LIMB	Congenital deformities of hip	DQ65	DQ653-DQ656, DQ658, DQ659
56	LIMB	Congenital deformities of feet	DQ66	DQ661-DQ669
57	LIMB	Other congenital musculoskeletal deformities	DQ68	DQ680, DQ682A, DQ683-DQ685
58	LIMB	Polydactyly	DQ69	
59	LIMB	Syndactyly	DQ70	
60	LIMB	Reduction defects of upper limb	DQ71	
61	LIMB	Reduction defects of lower limb	DQ72	
62	LIMB	Reduction defects of unspecified limb	DQ73	
63	LIMB	Other congenital malformations of limb(s)	DQ74	DQ740G
64	GENITAL	Congenital malformations of ovaries, fallopian tubes, and broad ligaments	DQ50	DQ501, DQ502, DQ505
65	GENITAL	Congenital malformations of uterus and cervix	DQ51	
66	GENITAL	Other congenital malformations of female genitalia	DQ52	DQ523, DQ525, DQ527
67	GENITAL	Hypospadias	DQ54	DQ544

68	GENITAL	Other congenital malformations of male genital organs	DQ55	DQ552B, DQ552F
69	GENITAL	Indeterminate sex and pseudohermaphroditism	DQ56	
70	GASTRO	Other congenital malformations of tongue, mouth, and pharynx	DQ38	DQ381, DQ382, DQ385B
71	GASTRO	Congenital malformations of oesophagus	DQ39	
72	GASTRO	Other congenital malformations of upper alimentary tract	DQ40	DQ400, DQ401
73	GASTRO	Congenital absence, atresia, and stenosis of small intestine	DQ41	
74	GASTRO	Congenital absence, atresia, and stenosis of large intestine	DQ42	
75	GASTRO	Other congenital malformations of intestine	DQ43	DQ430
76	GASTRO	Congenital malformations of gallbladder, bile ducts, and liver	DQ44	DQ444
77	GASTRO	Other congenital malformations of digestive system	DQ45	DQ458B
78	GASTRO	Congenital diaphragmatic hernia	DQ790	
79	EYE	Congenital malformations of eyelid, lacrimal apparatus, and orbit	DQ10	DQ101, DQ102, DQ103, DQ105

80	EYE	Anophthalmos,	DQ11	
		microphthalmos, and		
		macrophthalmos		
81	EYE	Congenital lens	DQ12	
		malformations		
82	EYE	Congenital malformations	DQ13	DQ135
		of anterior segment of eye		
83	EYE	Congenital malformations	DQ14	
		of posterior segment of eye		
84	EYE	Other congenital	DQ15	
		malformations of eye		
85	EFN	Congenital malformations	DQ16	
		of ear causing impairment		
		of hearing		
86	EFN	Other congenital	DQ17	DQ170-DQ175, DQ179
		malformations of ear		
87	EFN	Other congenital	DQ18	DQ180-DQ182, DQ184-DQ187, DQ189
		malformations of face and		
		neck		
88	ABW	Omphalocele	DQ792	
89	ABW	Gastroschisis	DQ793	
90	ABW	Other congenital	DQ795	
		malformations of		
		abdominal wall		
91	RESP	Choanal atresia	DQ300	
92	RESP	Congenital malformations	DQ32	DQ320, DQ322
		of trachea and bronchus		
93	RESP	Congenital malformations	DQ33	DQ331, DQ336
		of lung		
94	RESP	Other congenital	DQ34	
		malformations of		
		respiratory system		
95	OTHER	Cystic hygroma	DD181A	

96	OTHER	Other congenital	DQ750	
		malformations of skull and		
		face bones		
97	OTHER	Poland's syndrome	DQ798S	
98	OTHER	Moebius syndrome	DQ870G	
99	OTHER	Pierre Robin sequence	DQ870I	
100	OTHER	Sirenomelia syndrome	DQ872E	
101	OTHER	VACTERL association	DQ872G	
102	OTHER	Congenital malformations	DQ890	
		of spleen		
103	OTHER	Situs inversus	DQ893	
104	OTHER	Conjoined twins	DQ894	

ABW, abdominal wall defect; CARD, congenital heart defects; CHROM, chromosomal anomalies; DK, Denmark; DKpop, Danish population; DNSB, Danish Neonatal Screening Biobank; EFN, congenital ear, face, and neck anomalies; EYE, congenital eye anomalies; GASTRO, congenital gastrointestinal anomalies; GEN, genetic syndromes; GENITAL, genital anomalies; ICD-10, International Classification of Diseases, Tenth Revision; LCPD, Perthes' disease; LIMB, congenital limb anomalies; NERV, congenital anomalies of the nervous system; OFC, orofacial clefts; OTHER, other congenital anomalies; RESP, congenital malformation of the respiratory system; TERA, teratogenic syndromes; URO-RENAL, congenital anomalies of the kidneys and urinary tract.

**Table ii.** List of autism spectrum disorders and attention deficit hyperactivity disorders that were assessed within the 1994 to 2011 Danish Perthes' disease cohort. The Danish (Classification of diseases and health-related conditions) ICD-10 codes (ICD-10-DK) (listed here: https://medinfo.dk/sks/brows.php?s\_nod=6199) were used to define the autism spectrum disorders and attention deficit hyperactivity disorders investigated and are listed below. Conditions were assessed hierarchically in the order listed.

Order	Group	Description	ICD.10.DK
0	DKpop	All infants born in DK between 1994 and 2011 (subset of patients, born after the implementation of ICD-10, who have passed their twelfth birthday) included in the DNSB	None
1	LCPD	Infants born in DK between 1994 and 2011 (subset of patients, born after the implementation of ICD-10, who have passed their twelfth birthday) included in the DNSB	DM911, DM912, DM913
2	ASD	Childhood autism	DF840
3	ASD	Atypical autism	DF841
4	ASD	Rett syndrome	DF842
5	ASD	Other childhood disintegrative disorder	DF843
6	ASD	Overactive disorder associated with mental retardation and stereotyped movements	DF844
7	ASD	Asperger's syndrome	DF845
8	ASD	Other pervasive developmental disorders	DF848
9	ASD	Pervasive developmental disorder, unspecified	DF849
10	ADHD	Disturbance of activity and attention - Attention deficit	DF900
11	ADHD	Hyperkinetic conduct disorder	DF901
12	ADHD	Other hyperkinetic disorders	DF908
13	ADHD	Hyperkinetic disorder, unspecified	DF909

DK, Denmark; DNSB, Danish Neonatal Screening Biobank; ICD-10, International Classification of Diseases, Tenth Revision; LCPD, Perthes' disease.