BeSHG

COLLEGE GENETICS

Table of susceptibility loci in context of invasive prenatal testing

prepared by the BeSHG Prenatal Committee on 17.09.2022

chr	start in Mb (hg19)	stop in Mb (hg 19)	size in kb	CNV	gene	phenotype	morph. anomaly	return?	OMIM	update May 2017	update December 2021	update June / September 2022
1	146.57	147.39	820	distal 1q21.1 dup	*GJA5 (CX40)*	ID, DD, ASD, schizophrenia	macrocephaly, CHD	YES	612475	YES		YES
1	146.57	147.39	820	distal 1q21.1 del	*GJA5 (CX40)*	ID, DD, ASD, SZ, facial dysmorphism	microcephaly, CHD, renal and urinary tract anomalies	YES	612474	YES		YES
1	171.81	172,38(?)	57	1q24.3 del	*DNM3*	ID	IUGR, microcephaly, brachydactyly	YES				YES
2	50	51.11	1110	2p16.3 del (exon 6-24 del)	*NRXN1*	ID, ASD, SZ, DD, dysmorphic features	none	YES	614332		added to YES (pubmed ID 31932357 and discussion in consortium 18/06/2020)	YES
15	31.13	32.48	1350	15q13.3 del	*CHRNA7*	DD, ID, ASD, epilepsy, SZ	microcephaly, CHD	YES	612001	YES		YES
15	99.36	102.52	3160	15q26 del	*IGF1R*	MR	IUGR	YES		YES		
16	28.74	28.96	220	16p11.2 distal del	*SH2B1*	obesity, DD, ID, SZ	none	YES	613444	YES		YES
16	29.59	30.19	600	16p11.2 proximal dup	*TBX6*	ASD, ID, DD, SZ, anorexia	microcephaly	YES	614671	moved to YES since actionable; penetrance del and dup comparable		YES
16	29.59	30.19	600	16p11.2 proximal del	*TBX6*	ID, DD, ASD, obesity, SZ, speech delay	macrocephaly, vertebra	YES	611913	YES		YES
17	34.82	36.21	1390	17q12 deletion syndrome RCAD (renal cysts & diabetes)	*TCF2*	facial dysmorphy, genital abnormalities, ID, DD, ASD, MODY	renal anomalies	YES	614527	YES		YES
22	19.02	20.29	1270	22q11.2 dup	*TBX1*	ASD, ID, DD, dysmorphic features	microcephaly, CHD	YES	608363	YES		YES
1	144.97	146.61	1640	1q21.1 dup	*HFE2*	DD, ASD	CHD	NO		NO		NO
2	50	51.11	1110	2p16.3 del (whole gene, intronic, exon 1-5)	*NRXN1*	ID, ASD, SZ, DD, dysmorphic features	none	NO	614332	NO	NO in case of whole gene del, intronic del or exon 1-5 del (pubmed ID 31932357 and discussion in consortium 18/06/2020)	NO
2	110.87	110.98	110	2q13 dup	*NPHP1*	ASD, ID	none	NO		NO		NO
2	111.4	113	1600	2q13del		ID, DD, dysmorphic features	CHD			NO (Govaerts 2017)		NO
3	1.7	2.8	1100	3p26.3 del	*CNTN4*	ASD				NO (Govaerts 2017)		NO
3	195.7	197.30	1600	3q29 dup		MR, DD	none	NO		NO		NO (note: coordinates corrected)
10	49	52.4	3400	10q11.22q11.23 del		ID, DD				NO (Govaerts 2017)		NO
10	49	52.4	3400	10q11.22q11.23 del		ID, DD				NO (Govaerts 2017)		NO
13	20.81	21.01	1200	13q12 dup	*CRYL1*			NO		NO		NO
15	22.8	23.09	290	15q11.2 dup	*NIPA1*	DD, motor delay, speech delay, ASD	none	NO		NO (likely benign)		NO
15	22.8	23.09	290	15q11.2 del	*NIPA1*	ID, DD, epilepsy	CHD	NO	615656	NO (likely benign)		NO
15	31.13	32.48	1350	15q13.3 dup	*CHRNA7*	ADHD, ID, DD, ASD	none	NO		NO (likely benign)		NO
16	14.98	16.48	1500	16p13.11 dup	*MYH11*	ID, ASD, SZ, ADHD	aorta dilatation	NO		NO		NO
16	14.98	16.48	1500	16p13.11 del	*MYH11*	ID, DD, ASD, epilepsy	microcephaly	NO		NO		NO
16	21.94	22.46	520	16p12.2 dup	*EEF2K, CDR2*			NO		NO (likely benign)		NO
16	21.94	22.46	520	16p12.2 del	*EEF2K, CDR2*	DD, speech dealy	cranofacial and skeletal abnormalities, CHD	NO	136570	NO		NO
16	28.74	28.96	220	16p11.2 distal dup	*SH2B1*	anorexia, ID, DD, ASD, SZ	none	NO		NO		NO
17	34.73	36.22	1500	17q12 dup	*HNF1B*	DD	none	NO		NO		
22	21.91	23.65	1740	22q11.2 distal dup		DD, epilepsy, dysmorphic features	none	NO		moved back to NO (not enough evidence for pathogenicity)		NO