chr	start in Mb (hg19)	stop in Mb (hg 19)	size in kb	CNV	gene	phenotype	morph. anomaly	return?	OMIM	update May 2017
1	146,57	147,39	820	distal 1q21.1 dup	*GJA5 (CX40)*	ID, DD, ASD, schizophrenia	macrocephaly, CHD	YES	612475	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
1	171,81	172,38(?)	57	1q24.3 del	*DNM3*	ID	IUGR, microcephaly, brachydactyly	YES		
15	31,13	32,48	1350	15q13.3 del	*CHRNA7*	DD, ID, ASD, epilepsy, SZ	microcephaly, CHD	YES	612001	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
16	29,59	30,19	600	16p11.2 proximal dup	*TBX6*	ASD, ID, DD, SZ, anorexia	microcephaly	~~NO~~ YES	614671	moved to YES since actionable; penetrance del and dup comparable
16	29,59	30,19	600	16p11.2 proximal del	*TBX6*	ID, DD, ASD, obesity, SZ, speech delay	macrocephaly, vertebra	YES	611913	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
22	19,02	20,29	1270	22q11.2 dup	*TBX1*	ASD, ID, DD, dysmorphic features	microcephaly, CHD	YES	608363	YES
1	144,97	146,61	1640	1q21.1 dup	*HFE2*	DD, ASD	CHD	NO		NO
2	50	51,11	1110	2p16.3 del	*NRXN1*	ID, ASD, SZ, DD, dysmorphic features	none	NO	614332	NO
2	110,87	110,98	110	2q13 dup	*NPHP1*	ASD, ID	none	NO		NO
3	197,2	198,84	1600	3q29 dup		MR, DD	none	NO		NO
13	20,81	21,01	1200	13q12 dup	*CRYL1*	?	?	NO		NO
15	22,8	23,09	290	15q11.2 dup	*NIPA1*	DD, motor delay, speech delay, ASD	none	NO		NO (likely benign)
15	22,8	23,09	290	15q11.2 del	*NIPA1*	ID, DD, epilepsy	CHD	NO	615656	NO (likely benign)
15	31,13	32,48	1350	15q13.3 dup	*CHRNA7*	ADHD, ID, DD, ASD	none	NO		NO (likely benign)
16	14,98	16,48	1500	16p13.11 dup	*MYH11*	ID, ASD, SZ, ADHD	aorta dilatation	NO		NO
16	14,98	16,48	1500	16p13.11 del	*MYH11*	ID, DD, ASD, epilepsy	microcephaly	NO		NO
16	21,94	22,46	520	16p12.2 dup	*EEF2K, CDR2*	?	?	NO		NO (likely benign)
16	21,94	22,46	520	16p12.2 del	*EEF2K, CDR2*	DD, speech dealy	cranofacial and skeletal abnormalities, CHD	NO	136570	NO

16	28,74	28,96	220	16p11.2 distal dup	*SH2B1*	anorexia, ID, DD, ASD, SZ	none	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	~~YES~~ NO		moved back to NO (not enough evidence for pathogenicity)

Govaerts 2017:

2	111,4	113	1600	2q13del		ID, DD, dysmorphic features	CHD			NO
3	1,7	2,8	1100	3p26.3 del	*CNTN4*	ASD				NO
10	49	52,4	3400	10q11.22q11.23 del		ID, DD				NO