

Supplementary Table S5. Behavioural-trouble-related CNVs annotated by DECIPHER are covered by CES.

Syndrome	Flag	Phenotype	Status	Location (hg19)	Size (Mb)	Cytoband	Key Gene
1q21.1 recurrent microduplication	suspected	Autism, Intellectual disability	DUP	1:146533376-147883376	1.35	1q21.1	<i>GJA8</i>
2q33.1 deletion syndrome	confirmed	Behavioural/Psychiatric Abnormality, Cleft palate, Feeding difficulties in infancy, High palate, Inguinal hernia, Intellectual disability, Seizures, Short stature	DEL	2:196925121-205206939	8.28	2q33.1	<i>SATB2</i>
Williams-Beuren Syndrome (WBS)	confirmed	Aortic valve stenosis, Everted lower lip vermilion, Intellectual disability, Malar flattening, Microdontia, Short attention span, Short stature	DEL	7:72744455-74142672	1.4	7q11.23	<i>ELN</i>
7q11.23 duplication syndrome	confirmed	Delayed speech and language development, Intellectual disability, Short attention span	DUP	7:72744455-74142672	1.4	7q11.23	<i>ELN</i>
8p23.1 duplication syndrome	confirmed	Abnormal facial shape, Behavioural/Psychiatric Abnormality, Delayed speech and language development, Intellectual disability, Malformation of the heart and	DUP	8:8100055-11764629	3.66	8p23.1	<i>SOX7, GATA4</i>

16p11.2-p12.2 microduplication syndrome	confirmed	great vessels Abnormal facial shape, Autism, Global developmental delay, Intellectual disability, Microcephaly, Short Stature Brachydactyly syndrome, Hoarse voice, Hyperactivity,	DUP	16:21475060- 29284077	7.81	16p11.2- p12.2	<i>PLK1</i>
Smith-Magenis Syndrome	confirmed	Intellectual disability, Muscular hypotonia, Self- mutilation, Short stature, Sleep disturbance, Stereotypic behavior	DEL	17:16773072- 20222149	3.7	17p11.2	<i>ALDH3A2,</i> <i>RAI1,</i> <i>TOP3A</i>
Potocki-Lupski syndrome (17p11.2 duplication syndrome)	confirmed	Autism, Hyperactivity, Short attention span, Short stature	DUP	17:16773072- 20222149	3.45	17p11.2	<i>ALDH3A2,</i> <i>RAI1,</i> <i>TOP3A</i>
22q13 deletion syndrome (Phelan- Mcdermid syndrome)	confirmed	Autism, Delayed speech and language development, Hyperactivity, Intellectual disability, Muscular hypotonia	DEL	22:51045516- 51187844	0.142	22q12	<i>SHANK3</i>