Supplementary Table S6. DD-related CNV annotated by DECIPHER.

Syndrome	Location(hg19)	Genotype	Status	Size	Grade	CES covered
1p36 microdeletion syndrome	1:10001-12840259	Het	DEL	12.83 Mb	1	Y
1q21.1 susceptibility locus for Thrombocytopenia-Absent Radius (TAR) syndrome	1:145386506-145748067	Het	DEL	361.56 kb	3	Y
1q21.1 recurrent microdeletion (susceptibility locus for neurodevelopmental disorders)	1:146533376-147883376	Het	DEL	1.35 Mb	3	Y
1q21.1 recurrent microduplication (possible susceptibility locus for neurodevelopmental disorders)	1:146533376-147883376	Het	DUP	1.35 Mb	3	Y
2p21 Microdeletion Syndrome	2:44410451-44589584	Het	DEL	179.13 kb	.	Y
2p15-16.1 microdeletion syndrome	2:59285696-61819815	Het	DEL	2.53 Mb	.	Y
2q33.1 deletion syndrome	2:196925121-205206939	Het	DEL	8.28 Mb	1	Y
2q37 monosomy	2:239969863-240322643	Het	DEL	352.78 kb	1	Y
3q29 microdeletion syndrome	3:195726835-197344663	Het	DEL	1.62 Mb	.	Y
3q29 microduplication syndrome	3:195726835-197344663	Het	DUP	1.62 Mb	.	Y
Wolf-Hirschhorn Syndrome	4:1569197-2110236	Het	DEL	541.04 kb	1	Y
Cri du Chat Syndrome (5p deletion)	5:10001-12533304	Het	DEL	12.52 Mb	1	Y
Familial Adenomatous Polyposis	5:112043201-112181936	Het	DEL	138.74 kb	.	Y
Adult-onset autosomal dominant leukodystrophy (ADLD)	5:126112314-126172712	Het	DUP	60.40 kb	.	Y
Sotos syndrome	5:175724636-177052116	Het	DEL	1.33 Mb	1	Y
Williams-Beuren Syndrome (WBS)	7:72744455-74142672	Het	DEL	1.40 Mb	1	Y
7q11.23 duplication syndrome	7:72744455-74142672	Het	DUP	1.40 Mb	.	Y
Split hand/foot malformation 1 (SHFM1)	7:96318078-96339203	Het	DEL	21.13 kb	.	Y
8p23.1 duplication syndrome	8:8100055-11764629	Het	DUP	3.66 Mb	.	Y

8p23.1 deletion syndrome	8:8100055-11764629	Het	DEL	3.66 Mb	.	Y
8q21.11 Microdeletion Syndrome	8:77226464-77766239	Het	DEL	539.78 kb	.	Y
9q subtelomeric deletion syndrome	9:140513443-140730578	Het	DEL	217.14 kb	1	Y
WAGR 11p13 deletion syndrome	11:31806339-32457087	Het	DEL	650.75 kb	.	Y
Potocki-Shaffer syndrome	11:43994800-46052450	Het	DEL	2.06 Mb	1	Y
12p13.33 Microdeletion Syndrome	12:1080000-1346471	Het	DEL	266.47 kb	.	N
12q14 microdeletion syndrome	12:65071919-68645525	Het	DEL	3.57 Mb	.	Y
Angelman syndrome (Type 1)	15:22749354-28438266	Het	DEL	5.69 Mb	1	Y
Prader-Willi syndrome (Type 1)	15:22749354-28438266	Het	DEL	5.69 Mb	1	Y
Prader-Willi Syndrome (Type 2)	15:23619912-28438266	Het	DEL	4.82 Mb	1	Y
Angelman syndrome (Type 2)	15:23619912-28438266	Het	DEL	4.82 Mb	1	Y
15q13.3 microdeletion syndrome	15:30910306-32445407	Het	DEL	1.54 Mb	.	Y
15q24 recurrent microdeletion syndrome	15:74412643-75972911	Het	DEL	1.56 Mb	.	Y
15q26 overgrowth syndrome	15:99357970-102521392	Het	DUP	3.16 Mb	.	Y
ATR-16 syndrome	16:60001-834372	Het	DEL	774.37 kb	1	Y
Rubinstein-Taybi Syndrome	16:3775055-3930121	Het	DEL	155.07 kb	1	Y
16p13.11 recurrent microdeletion (neurocognitive disorder susceptibility locus)	16:14986684-16486684	Het	DEL	1.50 Mb	.	Y
16p13.11 recurrent microduplication (neurocognitive disorder susceptibility locus)	16:14986684-16486684	Het	DUP	1.50 Mb	.	Y
16p11.2-p12.2 microduplication syndrome	16:21475060-29284077	Het	DUP	7.81 Mb	.	Y
16p11.2-p12.2 microdeletion syndrome	16:21512062-30199854	Het	DEL	8.69 Mb	.	Y
Recurrent 16p12.1 microdeletion (neurodevelopmental susceptibility locus)	16:21946524-22467284	Het	DEL	520.76 kb	.	N
16p11.2 microduplication syndrome	16:29606852-30199855	Het	DUP	593.00 kb	.	Y
Miller-Dieker syndrome (MDS)	17:1-2588909	Het	DEL	2.59 Mb	1	Y

Charcot-Marie-Tooth syndrome type 1A (CMT1A)	17:14097915-15470903	Het	DUP	1.37 Mb	1	Y
Hereditary Liability to Pressure Palsies (HNPP)	17:14097915-15470903	Het	DEL	1.37 Mb	1	Y
Smith-Magenis Syndrome	17:16773072-20222149	Het	DEL	3.45 Mb	1	Y
Potocki-Lupski syndrome (17p11.2 duplication syndrome)	17:16773072-20222149	Het	DUP	3.45 Mb	.	Y
NF1-microdeletion syndrome	17:29107097-30263321	Het	DEL	1.16 Mb	1	Y
RCAD (renal cysts and diabetes)	17:34815072-36215917	Het	DEL	1.40 Mb	.	Y
17q21.31 recurrent microdeletion syndrome (Koolen de Vries syndrome)	17:43705166-44294406	Het	DEL	589.24 kb	1	Y
Early-onset Alzheimer disease with cerebral amyloid angiopathy	21:27252860-27543446	Het	DUP	290.59 kb	.	Y
Cat-Eye Syndrome (Type I)	22:1-16971860	Hom	DUP	16.97 Mb	.	Y
22q11 deletion syndrome (Velocardiofacial / DiGeorge syndrome)	22:19009792-21452445	Het	DEL	2.44 Mb	1	Y
22q11 duplication syndrome	22:19009792-21452445	Het	DUP	2.44 Mb	3	Y
22q11.2 distal deletion syndrome	22:21917117-23722445	Het	DEL	1.81 Mb	.	Y
22q13 deletion syndrome (Phelan-Mcdermid syndrome)	22:51045516-51187844	Het	DEL	142.33 kb	1	Y
Leri-Weill dyschondrostosis (LWD) - SHOX deletion	X:460558-753877	Het	DEL	293.32 kb	.	Y
Leri-Weill dyschondrostosis (LWD) - SHOX deletion	X:751878-867875	Het	DEL	116.00 kb	.	N
Steroid sulphatase deficiency (STS)	X:6455812-8133195	Hemi	DEL	1.68 Mb	.	Y
Xp11.22-p11.23 Microduplication	X:48334549-52117661	Unknown	DUP	3.78 Mb	.	Y
Xp11.22-linked intellectual disability	X:53401070-53683275	Unknown	DUP	282.21 kb	.	Y
Pelizaeus-Merzbacher disease	X:103031438-103047547	Unknown	DUP	16.11 kb	.	Y
Xq28 (MECP2) duplication	X:153287263-153363188	Unknown	DUP	75.93 kb	.	Y
Xq28 Microduplication	X:153624563-153881853	Unknown	DUP	257.29 kb	.	Y
AZFa	Y:14352761-15154862	Hemi	DEL	802.10 kb	.	Y*
AZFb+AZFc	Y:19964826-27793830	Hemi	DEL	7.83 Mb	.	Y*

AZFb	Y:20118045-26065197	Hemi	DEL	5.95 Mb	.	Y*
AZFc	Y:24977425-28033929	Hemi	DEL	3.06 Mb	.	Y*

Grade*:** provided by the DECIPHER indicating the possibility of a second contributing diagnosis ought to consider for all if phenotype differs from the "usual". ***Grade 1: Pathogenic anomaly; ***Grade 2***: Likely pathogenic anomaly; ***Grade 3***: Susceptibility locus; ***"."***: no grade provided by the DECIPHER.

***Y*:** Covered by CES target; ***N*:** Not covered by CES target; ***Y**:** Covered by CES target but not included in CNV calling.