

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 |

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|--|-----------------------|-----|-----|-----------|---|---|
| 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 |

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|--|-----------------------|---------|-----|-----------|---|----|
| 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 dyschondroostosis (LWD) - SHOX deletion | X:460558-753877 | Het | DEL | 293.32 kb | . | Y |
| Leri-Weill dyschondroostosis (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* |

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|------|---------------------|------|-----|---------|---|----|
| 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.