microdeletion 1p36
microdeletion 1p36 (GABRD) microduplication 1p36
microduplication 1p34.1
microdeletion 1p32.2
microdeletion 1p21.3
microdeletion 1q21.1 microduplication 1q21.1
thrombocytopenia-absent radius syndrome/TAR 1q21.1
deletion 1q21.1 (GJA5) duplication 1q21.1 (GJA5)
microdeletion 1q24q25 - - 1q24.3q25.1
microdeletion 1q24.3
Van der Waude syndrome/VWS1 - 1q32.2-q41
microdeletion 1q41–42
corpus callosum agenesis microdeletion 1q44
microduplication 2p25.3
Feingold syndrome/FS - 2p24.3
hypotonia-cystinuria syndrome/HCS - 2p21
holoprosencephaly 2/HPE2 - 2p21
microduplication 2p21
NRXN1 microdeletion NRXN1 microduplication 2p16.3
microdeletion 2p15–16.1
microdeletion 2p14-p15
microdeletion 2p11.2-p12
microdeletion 2q11.2
mesomelic dysplasia/MMD - 2q11.2
microdeletion 2q11.2q13
microduplication 2q11.2q13
nephronophthisis 1/NPHP1 microduplication 2q11.2q13
microdeletion 2q13 microduplication 2q13 - 2q13
autism-dyslexia microdeletion 2q14.3
microduplication 2q14.3
Mowat–Wilson syndrome/MWS - 2q22.3
microdeletion 2q23.1
microdeletion 2q23.3q24.1
microdeletion 2q24.3 neonatal epilepsy
microduplication 2q14.3
synpolydactyly 1/SPD1 microduplication 2q31.1
microdeletion 2q31.2-q32.3
microdeletion 2q33.1
brachydactyly-mental retardation syndrome/BDMR 2q37
distal 3p deletion - 3p25-p26
Von Hippel Lindau disease/VHL - 3p25-p26
microdeletion 3p21.31
microdeletion 3p14.1p13
microdeletion 3p11.1p12.1
proximal 3q microdeletion syndrome 3q13.11-q13.12
microdeletion 3q13.31
blepharophimosis, ptosis, and epicanthus inversus syndrome/BPES 3q23
Dandy–Walker syndrome/DWS - 3q24
microdeletion 3q27.3q29
microdeletion 3q29 microduplication 3q29
Wolf–Hirschhorn syndrome/WHS microduplication 4p16.3
microduplication 4p16.1

microdeletion 4p15.3
microdeletion 4q21.21q21.22
microdeletion 4q21
microdeletion 4q21.2q21.3
Parkinson disease/PARK14q22.1
Rieger type 1/RIEG1 - 4q25 - 4q32.1-q32.2 Triple/Duplication syndrome
Cri–du-Chat syndrome/CdCS - 5p15.2-p15.33
Cornelia de Lange syndrome/CDLS NIPBL microduplication 5p13.2
spinal muscular atrophy/SMA - 5q13.2
microdeletion 5q14.3 - 5q14.3
microdeletion 5q14.3-q15
familial adenomatous polyposis/FAP - 5q22.2
dominant leukodystrophy/ADLD 5q23.2
PITX1 microdeletion - 5q31.1
microdeletion 5q31.3
Pseudo trisomy 13 syndrome 5q35.1
microdeletion 5q35.1
parietal foramina/PFM - 5q35.2
Sotos syndrome microduplication 5q35
microdeletion 6p
microdeletion 6p22.3
adrenal hyperplasia/AH - 6p21.32
microdeletion 6p21.31
microdeletion 6q13–14
Prader–Willi like - 6q16.2
mellitus 1/TNDM1 6q24.2
microdeletion 6q25.2-q25.3
PARK2 microdeletion PARK2 microduplication 6q26
microdeletion 6q27 anosmia Chondroma/CHDM
Saethre–Chotzen syndrome/SCS - 7p21.1
Greig cephalopolysyndactyly/GCPS - 7p14.1
Williams–Beuren syndrome/WBS microduplication 7q11.23
WBS-distal deletion (RHBDD2,HIP1) 7q11.23
split hand/foot malformation 1/SHFM1 7q21.3
microdeletion 7q22.1-q22.3
autism/dyslexia microdeletion 7q31.1
speech-language-disorder 1/SPCH1 - 7q31
holoprosencephaly 3/HPE3 - 7q36.3
polysyndactyly syndrome/TPTS 7q36.3
Currarino syndrome/CS - 7q36.3
microdeletion 8p23.1 microduplication 8p23.1
microdeletion 8p21.2
microdeletion 8p12p21
microduplication 8q11.23
CHARGE syndrome microduplication 8q12
microdeletion 8q12.3q13.2
mesomelia-synostoses syndrome/MSS 8q13
microdeletion 8q21.11
nablus mask-like facial syndrome/NMLFS 8q21.3-q22.1
microdeletion 8q22.2q22.3
Langer–Giedion syndrome/LGS - 8q24.11
sex reversal syndrome 4/SRXY4 - 9p24.3

monosomy 9p syndrome - 9pter-p22.3
microduplication 9q21.11
microdeletion 9q22.3 PTCH1 microduplication 9q22.3
holoprosencephaly 7/HPE7 - 9q22.32
nail-patella syndrome/NPS - 9q33.3
early infantile epileptic encephalopathy 4/EIEE4 9q34.11
microdeletion 9q34 (EHMT1) microduplication 9q34
subtelomere deletion 9q - 9q34.3
hypoparathyroidism, sensorineural deafness, and renal disease/HDRS 10p15
Di George syndrome 2/DGS2 - 10p12.31
microdeletion 10q22-q23 (NRG3,GRID1)
juvenile polyposis syndrome/JPS - 10q23.2-q23.3
10q24.32
microdeletion 10q25q26
Beckwith–Wiedemann syndrome/BWS—Silver Russell syndrome/SRS microdeletion microduplication 11p15.5
WAGR syndrome microduplication 11p13
Potocki–Shaffer syndrome/PSS - 11p11.2
11q12.2q12.3
microdeletion 11q14.1 - - 11q14.1-q14.2
Jacobsen syndrome/JBS - 11q23.3-qter
microduplication 12p13.31 - 12p13.31
microdeletion 12q14
nasal speech-hypothyroidism microdeletion/NSH 12q15-q21.1
Noonan syndrome 1/NS1 - 12q24.1
microdeletion 13q12 (CRYL1) microduplication 13q12
spastic ataxia Charlevoix–Saguenay/SACS 13q12.12
microdeletion 13q12.3-q13.1
retinoblastoma/RB1 - 13q14.2
Hirschsprung disease 2/HSCR2 - 13q22
holoprosencephaly5/HPE5 - 13q32.3
microdeletion 14q11.2 - 14q11.2
congenital Rett variant/CRV microduplication 14q12
microdeletion 14q22-q23
autism spherocytosis microdeletion/ASC 14q23.2-q23.3
microdeletion 14q32.2
microdeletion 15q11.2 (NIPA1) microduplication 15q11.2
Angelman syndrome Typ1/AS1 microduplication 15q11.2-q13.1
Angelman syndrome Typ2/AS2 microduplication 15q11.2-q13.1
Prader–Willi syndrome Typ 1/microduplication 15q11.2-q13.1
Prader–Willi syndrome Typ 2/microduplication 15q11.2-q13.1
microdeletion 15q13.3 (CHRNA7) microduplication 15q13.3
microdeletion 15q14 - - 15q14
deafness and male infertility syndrome/DMIS 15q15.3
microdeletion 15q21
microdeletion 15q24 (BBS4,NPTN,NE01)
microdeletion 15q24 microduplication 15q24
orofacial clefting/OC - 15q24.3-q25.2
microdeletion 15q25
microdeletion 15q26.1
Fryns syndrome/FNS - 15q26.2
microdeletion 15q26.2-qter
ATR-16-syndrome - 16p13.3

tuberous sclerosis microdeletion
syndrome/PKDTS tuberous sclerosis microduplication 16p13.3
Rubinstein–Taybi syndrome 1/RSTS1Rubinstein–Taybimicroduplication 16p13.3
microdeletion 16p13.1 (MYH11) microduplication 16p13.1
microdeletion 16p11.2-p12.2 microduplication 16p11.2-p12.2
microdeletion 16p12.1 (EEF2K,CDR2)
microduplication 16p12.2
16q11.2 distal microdeletion
microduplication (SH2B1) 16q11.2
microdeletion 16p11.2 (TBX6) microduplication 16p11.2
microdeletion 16q11.2-q12.1
microdeletion 16q21-q22
microdeletion 16q12.1-q12.2
microdeletion 16q24.1
FANCA deletion - 16q24.3
Miller–Dieker syndrome/MDLS Miller–Dieker microduplication 17p13.3
microdeletion 17p13.3 (YWHAE) microduplication 17p13.3
microdeletion 17p13.1
hereditary liability to pressure palsies/HNPP Charcot–Marie–Tooth 1A/ 17p12
Smith–Magenis syndrome/SMS Potocki–Lupski syndrome/17p11.2
neurofibromatosis 1/NF1 microduplication NF1 17q11
microdeletion 17q11.2-q12
microdeletion 17q12a
renal cysts and diabetes syndrome/microduplication 17q12b
Van Buchem disease/VBCH - 17q12-q21
microdeletion 17q21.3 (MAPT) microduplication 17q21.31
microdeletion 17q21.31-q21.32
microdeletion 17q22-q23.2
microduplication 17q23.1
microdeletion 17q24.2-q24.3
carney complex syndrome 1/CNC1 - 17q24.2-q24.3
microduplication 17q24.3
holoprosencephaly 4/HPE4 - 18p11.31
proximal 18q microdeletion - 18q12.3-q21.1
Pitt–Hopkins syndrome/PTHS - 18q21.1
microdeletion 18q22.3-q23
microdeletion 19p13.2
Sotos like microduplication 19p13.2
microdeletion 19p13.13 microduplication 19p13.13
microdeletion 19p13.12
microdeletion 19p13.11
microdeletion 19q13.11
Diamond–Blackfan anemia/DBA - 19q13.2
microdeletion 20p12.3
Alagille syndrome 1/ALGS1 - 20p12
microdeletion 20q13.13-q13.2
Albright hereditary osteodystrophy/20q13.32
microdeletion 20q13.33
microdeletion 21q21.1
microduplication 21q21.3
platelet disorder/PD - 21q22.12
Down syndrome/DS21q22.13

Cat-Eye syndrome/CES 22p11.1-q11.21
Di George syndrome/CATCH22/microduplication 22q11.2
distal microdeletion 22q11.2 (BCR,MAPK1) distal microduplication 22q11.2 (BCR, MAPK1)
neurofibromatosis 2 microdeletion syndrome 22q12.2
Phelan–McDermid syndrome microduplication 22q13 (SHANK3)
Leri–Weill dyschondrosteosis/LWD - Xp22.33
X-Linked autism-2/AUTSX2 - Xp22.32-p22.31
Steroid sulphatase deficiency/STS - Xp22.31
Kallmann syndrome 1/KAL1 - Xp22.31
MIDAS syndrome - Xp22.2
Nance–Horan syndrome/NHS - Xp22.13
microdeletion Xp22.11 - Xp22.11
X-linked congenital adrenal hypoplasia/AHC DAX1 microduplication Xp21.2
complex glycerol kinase/CGK - Xp21.2
muscular dystrophy Duchenne/Xp21.2
Xp11.3 deletion syndrome
Goltz syndrome/GS - Xp11.23
dehydrogenase X/HSD Xp11.22
microduplication Xq12q13.1
X inactivation specific transcript/XIST Xq13.2
Bruton agammaglobulinemia/XLA - Xq22.1
microdeletion Xq22.2 Pelizaeus–Merzbacher microduplication/PMD Xq22.2
microdeletion Xq22.3q23
lymphoproliferative syndrome 1/XLP1 Xq25
X-linked hypopituitarism/SRXX3 Xq27.1
fragile site mental retardation 1/FMR1 Xq27.3
microdeletion Xq28
Rett syndrome/RS MECP2 microduplication Xq28
sex-determining region Y/SRY - Yp11.31
AZFa microdeletion - Yq11.21
AZFb microdeletion - Yq11.22 - q11.223
AZFb+c microdeletion - Yq11.221-q11.23
AZFc microdeletion - Yq11.223-q11.23