



Phenotype	Genes	Cytogenetics
Perisylvian polymicrogyria (Polymicrogyria, bilateral perisylvian; BPP)	<i>SRPX2</i>	Xq22.1
10q22q23 deletion	<i>NRG3, GRID1</i>	10q22q23 deletion
11q11–q13.3 duplication/multiple craniosynostoses, congenital heart defect	<i>FGF3, FGF4</i>	11q13.3 duplication
12q14.1q15 microdeletion	<i>GRIP1</i>	12q14.3 deletion
12q24.21–q24.23 microduplication	<i>THRAP2, NOS1, RFC5</i>	12q24.21–q24.23 duplication
13q33q34 deletion/ genital malformation in males/Microcephaly/MR	<i>EFNB2, ARHGEF7</i>	13q33.3q34 deletion
14q11.2 deletion syndrome	<i>SUPT16H, CHD8</i>	14q11.2 deletion
14q12 deletion syndrome	<i>FOXP1B</i>	14q12 deletion
15q21 microdeletion syndrome		15q21.2 deletion
15q24 deletion	<i>HCN4</i>	15q24.1
15q24 microdeletion	<i>PML</i>	15q24.1 deletion
15q26.3 deletion, Severe IUGR, developmental delay, postnatal growth retardation	<i>IGF1R insulin-like growth factor -1 receptor</i>	15q26.3 deletion
16p11.2p12 deletion		16p12.1 deletion
17q21.31 microdeletion/microduplication syndrome	<i>MAPT, CRHR1</i>	17q21.31 deletion
1q21.2 deletion/duplication		1q21.2 deletion/duplication
1q41q42 deletion	<i>DISP1</i>	1q41 deletion
20q13.33 deletion/Autosomal-dominant nocturnal frontal lobe epilepsy, Benign familial neonatal convulsions (type 1), hypotrichosis–lymphedema–telangiectasia	<i>ARFGAP1, CHRNA4, KCNQ2, SOX18</i>	20q13.33 deletion
22q11.2 duplication syndrome reciprocal to DGS deletion	<i>TBX1 T-box 1 ?</i>	22q11.2 duplication
22q13.3 deletion/Autism	<i>SHANK3</i>	22q13.33 deletion
2p15–p16.1 microdeletion		2p15–p16.1 deletion
2q22.3 deletion		2q22.3 deletion
2q22.3q23.3	<i>MBD5, KIF5C</i>	2q23.1
2q35	<i>IHH</i>	2q35

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2q37 deletion/brachydactyly-MR/obesity/Albright hereditary osteodystrophy	<i>CENTG2, GPC1, GPR35, ATSV/KIF1A, STK25</i>	2q37.3 deletion
3p25 deletion	<i>CNTN4</i>	3p25.3 deletion
3q29 deletion syndrome/Autism	<i>PAK2, DLG1</i>	3q29 deletion
5q14.3 deletion	<i>MEF2C</i>	5q14.3
5q21q31 deletion syndrome		5q21q31 deletion
6q24.3q25.1(Diabetes mellitus, insulin-dependent, 5; IDDM5)	<i>SUMO4</i>	6q25.1
7q11.23 duplication (WBS region)		7q11.23 duplication
7q21.13q22.1 deletion/Ectrodactyly/Deafness	<i>CUTL1 (CUX1), FZD1</i>	7q22.13q22.1 deletion
8p23.1 Inverted duplication/deletion		8p22p23 inversion
9q34.3 deletion syndrome	<i>NOTCH1, EHMT1</i>	9q34.3 deletion
Acheiropody/ and preaxial polydactyly	<i>LMBR1</i>	7q36.3 deletion
Action myoclonus-renal failure syndrome (AMRF)	<i>SCARB2</i>	4q21.1 deletion
Adrenal hyperplasia, congenital (CAH) due to 21-alpha hydroxylase deficiency	<i>CYP21A2</i>	6p21.32 deletion
Adrenal hypoplasia congenital (AHC)	<i>NROB1 nuclear receptor family 0 B1 (DAX1)</i>	Xp21.2 deletion
Adrenoleukodystrophy; (ALD)	<i>ABCD1</i>	Xq28 deletion
Alagille syndrome (AGS)	<i>JAG1 jagged 1</i>	20p12.2 deletion
Albinism, ocular type 1	<i>GPR143/OA1</i>	Xp22.2 deletion
All 41 unique subtelomeric regions	Multiple	41 sites
All 43 unique pericentromeric regions	Multiple	43 sites
Allan-Herndon-Dudley syndrome, X-linked mental retardation (XLMR)	<i>SLC16A2</i>	Xq13.2 deletion
Alpha thalassemia-MR syndrome	<i>HBA2 & HBA1</i>	16p13.3 deletion
Alport syndrome, X-linked (ATS)	<i>COL4A5</i>	Xq22.3 deletion
Andersen syndrome	<i>KCNJ2</i>	17q24.3 deletion
Androgen insensitivity syndrome (AIS)	<i>AR</i>	Xq12 deletion
Aneuploidy for 24 chromosomes	Multiple	24 chromosomes
Angelman syndrome (AS)	<i>UBE3A ubiquitin ligase 3A, AS-SRO,</i>	15q11.2-q12 deletion
Angelman syndrome (AS)	AS imprinting center	15q11.2 deletion
Aniridia II (AN2)	<i>PAX6 paired box gene 6</i>	11p13 deletion
Anterior segment mesenchymal dysgenesis (ASMD)/ Cataract	<i>PITX3</i>	10q24.32 deletion
Arthropathy, progressive pseudorheumatoid, of childhood; (PPAC)	<i>WISP3</i>	6q21 deletion
Ataxia	<i>ITPR1</i>	3p26.2 deletion

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Ataxia teleangiectasia	<i>ATR</i>	3q23 deletion
Atrial septal defect	<i>GATA4</i>	8p23.1 deletion
ATRX, XLMR-Hypotonic facies syndrome, ATR-X, and others	<i>ATRX</i>	Xq21.1 deletion/duplication
Autism		4q32.1 deletion
Autism	<i>DLX5</i>	7q21.3 deletion
Autism	<i>JMJD1C, TRIP8, REEP3</i>	10q21.3 deletion
Autism	<i>NUFIP1</i>	13q14.12 deletion
Autism	<i>NRXN3</i>	14q31.1 deletion
Autism	<i>A2BP1</i>	16p13.2 deletion
Autism	<i>DLG4</i>	17p13.1 deletion
Autism	<i>NUFIP2</i>	17q11.2 deletion
Autism	<i>CNTNAP1</i>	17q21.31 deletion
Autism	<i>ASMT (ASMTL)</i>	Xp22.33 deletion
Autism	<i>DPP10</i>	2q14.1 deletion
Autism	<i>DPP6</i>	7q36.2 deletion
Autism	<i>PCDH9</i>	13q21.32 deletion
Autism	<i>SYBL1</i>	Xq28 deletion
Autism	<i>NRXN1</i>	2p16.3 deletion
Autism	<i>CNTNAP3</i>	9p13.1 deletion
Autism	<i>BDNF</i>	11p14.1 deletion
Autism	<i>NRXN2</i>	11q13.1 deletion
Autism / Asperger syndrome-1, X-linked	<i>NLGN3</i>	Xq13.1 deletion
Autism/Schizophrenia 9 (SCZD9)	<i>DISC1</i>	1q42.2 deletion
Autistic Features 15q11.2-q12	Uncertain	15q11.2-q13 maternal duplication
Autistic features, X-linked, susceptibility to, AUTSX2 (XLMR)	<i>NLGN4</i>	Xp22.32 deletion
Azoospermia factor (AZFa, AZFb)		Yq11.21 deletion
Bartter syndrome type 2	<i>KCNJ1</i>	11q24.3 deletion
Bartter syndrome type 3	<i>CLCNKB</i>	1p36.13 deletion
Bartter syndrome type 4 (Bartter syndrome type 3)?	<i>CLCNKA, CLCNKB, BSND</i>	1p36.13-p32.3
Bartter syndrome, antenatal type 1	<i>SLC12A1</i>	15q21.1 deletion
Bartter with autosomal dominant	<i>CASR</i>	3q21.1 deletion
Basal cell nevus syndrome (BCNS) / Gorlin syndrome (GS)	<i>PTCH1</i> <i>patched Drosophila homolog</i>	9q22.32 deletion

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Beckwith-Wiedemann syndrome (BWS)	<i>IGF2 insulin-like growth factor II, CDKN1C cyclin-dependent kinase inhibitor 1C H19, KCNQ1, p57 (CDKN1C)</i>	11p15.4/15.5 deletion/duplication
Bilateral frontoparietal polymicrogyria/focal or multifocal epilepsy	<i>GPR56</i>	16q13 deletion
Bipolar disorder	<i>IMPA1</i>	8q21.13 deletion
Birk-Barel syndrome	<i>KCNK9</i>	8q24.3
Blepharophimosis, ptosis, and epicanthus inversus (BPES)	<i>FOXL2</i>	3q22.3 deletion
Blepharophimosis/vormian hypoplasia/exotropia/DD	<i>POFUT2</i>	21q22.3 deletion
Börjeson-Forssman-Lehmann syndrome	<i>PHF6</i>	Xq26.2 deletion
BP1/BP2 breakpoint region in 15q11.2		15q11.2 deletion/duplication
Brachydactyly and other skeletal anomalies	<i>GPC5, GPC6</i>	13q31.3 deletion
Brachydactyly, type C (BDC)	<i>GDF5</i>	20q11.22 deletion
Branchiootic syndrome-3 (BOS3)	<i>SIX1</i>	14q23.1 deletion
Branchiootorenal dysplasia syndrome (BOR)/Otofaciocervical (OFC)/Melnick-Fraser	<i>EYA1</i>	8q13.3 deletion
Brunner syndrome/Monoamine oxidase-A deficiency/Antisocial behavior following childhood maltreatment	<i>MAOA</i>	Xp11.3 deletion
Bruton agammaglobulinemia tyrosine kinase	<i>BTK Bruton agammaglobulinemia tyrosine kinase</i>	Xq22.1 deletion
Buschke-Ollendorff syndrome / Osteopoikilosis, short stature and mental retardation	<i>LEMD3</i>	12q14.3 deletion
CADASIL	<i>NOTCH3</i>	19p13.12 deletion
CALC1, behavioral problems and autistic spectrum disorder	<i>CALCA, CALCB</i>	11p15.2 deletion
Campomelic dysplasia	<i>SOX9 SRY- box 9</i>	17q24.3 deletion
Canavan disease	<i>ASPA</i>	17p13.3 deletion
Cardiomyopathy, familial hypertrophic/Congenital heart defect	<i>ACTC</i>	15q14 deletion
Cat eye syndrome (CES)	<i>CECR1, CECR5, CECR6</i>	inv dup(22) (q11.2)
Cerebellar hypoplasia	<i>OPHN1</i>	Xq12 deletion
Cerebral cavernous malformation	<i>CCM2</i>	7p13 deletion
Cerebral cavernous malformations 2	<i>CCM2</i>	7p13 deletion
Ceroid lipofuscinosis, neuronal 8, northern epilepsy variant	<i>CLN8</i>	8p23.3 deletion
Char syndrome	<i>TFAP2B</i>	6p12.3 deletion

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Charcot-Marie-Tooth disease type 1A (CMT1A)	<i>PMP22</i> peripheral myelin protein 22	17p12 duplication
CHARGE syndrome	<i>CHD7</i> Chromo-domain helicase DNA-binding 7	8q12.2 deletion
Chondrodysplasia punctata, X-linked recessive	<i>CDPX1 (ARSE)</i>	Xp22.33 deletion
Chondrodysplasia, grebe tybe	<i>CDMP1 (GDF5)</i>	20q11.22 deletion
Chondrodysplasias (Stickler syndrome/Spondyloepiphyseal dysplasia)	<i>COL2A1</i>	12q13.11 deletion
Choroideremia	<i>CHM</i>	Xq21.2 deletion
Choroideremia	<i>CHM</i>	Xq21.2
Chromosome 10q deletion syndrome		10q26 deletion
Chromosome 18p deletion syndrome		18p11.3 deletion
Chromosome 18q deletion syndrome		18q23 deletion
Chromosome Xp11.3 deletion syndrome/Retinitis pigmantosa X-linked RP2/ XLMR	<i>RP2</i>	Xp11.3 deletion
Chronic pancreatitis	<i>SPINK1</i>	5q32 deletion
Cleft lip/palate	<i>ESR1</i>	6q25.1 deletion
Cleft lip/palate	<i>FGFR2</i>	10q26.13 deletion
Cleft palate, isolated (CPI)	<i>SATB2</i>	2q33.1 deletion
Cleft plate, congenital heart defect	<i>GREM1, GJD2 (CX36), MEIS2</i>	15q13.3 deletion
Cleidocranial dysplasia (CCD)	<i>RUNX2 runt-related transcription factor 2</i>	6p12.3 deletion
Coffin-Lowry syndrome	<i>RPS6KA3/RSK2</i>	Xp22.12 deletion
Coloboma	<i>SNAP25</i>	20p12.2 deletion
Corneal dystrophies	<i>TGFBI</i>	5q31.2
Cornelia de Lange syndrome (CDLS)	<i>NIPBL nipped-B-like</i>	5p13.2 deletion
Cornelia de Lange syndrome (CDLS), X-linked	<i>SMC1A/SMC1L1</i>	Xp11.22 deletion
Cowden (CD) & Bannayan-Riley-Ruvalcaba syndrome (BRRS)	<i>PTEN</i>	10q23.31 deletion
Craniofrontal dysplasia	<i>CFND</i>	Xq11.2q13.1
Cranio metaphyseal dysplasia autosomal dominant (CMDD)	<i>ANKH</i>	5p15.2 deletion
Craniosynostosis	<i>SOX6</i>	11p15.2 deletion
Craniosynostosis (CRS2)	<i>MSX2</i>	5q35.2 deletion
Creatine deficiency syndrome / X-linked mental retardation (XLMR)	<i>SLC6A8</i>	Xq28 deletion
Cri-du-Chat syndrome (cry/speech/face critical regions)	Multiple (<i>TERT, EGR1</i>)	5p15.2 -p13.3 deletion
Currarino syndrome (Sacral agenesis)	<i>HLXB9</i>	7q36.3 deletion

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Cystinosis, Nephropathic (CTNS)	<i>CTNS</i>	17p13.2-p13.3 deletion
Dandy-Walker syndrome (DWS)	<i>ZIC1, ZIC4 zinc finger protein of the cerebellum 1, 4</i>	3q24 deletion
Danon disease	<i>LAMP2</i>	Xq24 deletion
Deafness, X-linked 2 (DNFX2)	<i>POU3F4</i>	Xq21.1 deletion
Dent disease	<i>CLCN5</i>	Xp11.22 deletion
Diabetes insipidus, nephrogenic, X-linked	<i>AVPR2</i>	Xq28 deletion
Diabetes mellitus, transient neonatal, IUGR	<i>ZAC (PLAGL1)</i>	6q24.2 paternal deletion
Diaphragmatic hernia	<i>SOX7</i>	8p23.1 deletion
Diaphragmatic hernia, congenital	<i>NR2F2</i>	15q26.1-q26.2 deletion
DiGeorge syndrome 2 (DGS2)		10p14 deletion
DiGeorge/velocardiofacial syndrome DGS1/VCFS / Autism	<i>GNB1L TBX1 T-box 1</i>	22q11.2 deletion
Dosage sensitive sex reversal (DSS)	<i>NROB1 nuclear receptor family 0 B1 (DAX1)</i>	Xp21.2 duplication
Down syndrome critical regions	<i>DSCR1, DSCR2, DSCR3, DSCR4, DSCR5, DSCR8, DSCR9, DSCR10,</i>	21q22.12-q22.13/q22.2?
Duchenne/Becker muscular dystrophy (DMD/BMD)	<i>DMD</i>	Xp21.1 deletion/duplication
Dyggve-Melchior-Clausen syndrome (DMC) (autosomal recessive)	<i>DYM</i>	18q21.1 deletion/duplication
Dyskeratosis congenita, X-linked	<i>DKC1</i>	Xq28 deletion
Ectodermal dysplasia, anhidrotic	<i>EDAR</i>	2q13 deletion
Ectodermal dysplasia, X-linked hypohidrotic	<i>EDA</i>	Xq13.1 deletion
Epilepsy	<i>KCND2</i>	7q31.31 deletion
Epilepsy	<i>EFHC2</i>	Xp11.3 deletion
Epilepsy idiopathic generalized/Migraine/Spinocerebellar ataxia 6	<i>CACNA1A</i>	19q21.2 deletion
Epilepsy, Benign neonatal (EBN2)	<i>KCNQ3</i>	8q24.22 deletion
Epilepsy, idiopathic generalized (EIG6)	<i>CACNA1H</i>	16p13.3 deletion
Epilepsy, juvenile absence	<i>GRIK1 (kainate ionotropic)</i>	21q21.3 deletion
Epilepsy, juvenile absence (JAE)	<i>GRIK1</i>	21q21.3 deletion
Epilepsy, juvenile absence (JAE) juvenile myoclonic (EJM1)	<i>EFHC1, c6orf33, LMPB1</i>	6p12.2 deletion
Epilepsy, juvenile myoclonic	<i>CACNB4</i>	2q22-q23 deletion
Epilepsy, juvenile myoclonic	<i>CLCN2</i>	3q27.1 deletion
Epilepsy, juvenile myoclonic	<i>CHRNA7</i>	15q13.3 deletion
Epilepsy, lateral temporal lobe, autosomal dominant (ADLTE)	<i>LGI1</i>	10q23.33 deletion

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Epilepsy, nocturnal frontal lobe (type 3)	<i>CHRNA2</i>	1q21.3 deletion
Epilepsy, nocturnal frontal lobe (type 4)	<i>CHRNA2</i>	8p21.2 deletion
Epilepsy, nocturnal frontal lobe, type2/ Autosomal-dominant nocturnal frontal lobe epilepsy (ADNFLE)	<i>CHRNA3, CHRNA4, CHRNA5,</i>	15q25.1 deletion
Epilepsy, progressive myoclonus	<i>NHLRC1</i>	6p22.3 deletion
Epilepsy, X-linked	<i>SYN1</i>	Xp11.23/11.3 deletion
Episodic ataxia (EA1), partial epilepsy	<i>KCNA1</i>	12p13.32 deletion
Fabry disease	<i>GLA</i>	Xq22.1 deletion
Faciogenital dysplasia	<i>FGD1</i>	Xp11.22 deletion
Faciogenital dysplasia/Aarskog-Scott syndrome	<i>FGD1</i>	Xp11.22 deletion
Feingold syndrome	<i>MYCN</i>	2p24.3 deletion
FGFR3 (Achondroplasia; ACH)	<i>FGFR3</i>	4p16.3
FGS2		Xq28 deletion
Focal dermal hypoplasia (Goltz syndrome), XLMR	<i>PORCN</i>	Xp11.23 deletion
Forebrain defects	<i>TDGF1 (CRIPTO)</i>	3p21.31 deletion
Fragile-X mental retardation syndrome (FMR1)	<i>FMR1</i>	Xq27.3 deletion
Generalized epilepsy with febrile seizures plus (GEFS+)/ Epilepsy, childhood absence (CAE)	<i>GABRG2</i>	5q34 deletion
Generalized epilepsy with febrile seizures-3 plus (type 1)	<i>SCN1B</i>	19q13.11 deletion
Glucose transport defect / Epilepsy, autosomal dominant	<i>GLUT1 (SLC2A1)</i>	17p13.1/1p34.2 deletion
Gonadal dysgenesis, XY female type/Sex reversal X/Y translocations, (Sex determination male)	<i>SRY sex-determining region Y</i>	Yp11.31 translocation/deletion
Granulomatous disease, chronic X-linked	<i>CYBB</i>	Xp11.4 deletion
Greig cephalo-polysyndactyly syndrome (GCPS)	<i>GLI3 GLI-Kruppel family 3</i>	7p14.1 deletion
Hand-foot-uterus syndromes	<i>HOXA13</i>	7p15.2 deletion
Hemophilia A, (Factor 8, hemophilia)	<i>F8</i>	Xq28 deletion
Hereditary neuropathy with pressure palsies (HNPP)	<i>PMP22 peripheral myelin protein 22</i>	17p12 deletion
Heterotaxy	<i>ACVR2B</i>	3p22.2 deletion
Heterotaxy	<i>EBAF (LEFTY2)</i>	1q42.12 deletion
Heterotaxy, visceral 2	<i>CFC1</i>	2q21.1 deletion
Heterotopia, periventricular, X-linked dominant/ Otopalatodigital syndrome type 1 (OPD)/multifocal epilepsy in females	<i>FLNA</i>	Xq28 deletion
Hirschsprung disease	<i>RET</i>	10q12.11 deletion
Hirschsprung disease (HSCR2)	<i>EDNRB</i>	13q22.3 deletion

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Holoprosencephaly 1 (HPE1)	<i>TMEM1 transmembrane protein1,LSS</i>	21q22.3 deletion
Holoprosencephaly 1 (HPE1)	<i>SIM2</i>	21q22.13 deletion
holoprosencephaly 2(HPE2)	<i>SIX3 sine oculis homolog 3</i>	2p21 deletion
Holoprosencephaly 3 (HPE3)	<i>SHH sonic hedgehog</i>	7q36.3 deletion
Holoprosencephaly 4 (HPE4)	<i>TGIF transforming growth factor-β induced factor</i>	18p11.31 deletion
Holoprosencephaly 5 (HPE5)	<i>ZIC2 zinc finger protein cerebellum 2</i>	13q32.3 deletion
Holoprosencephaly 6 (HPE6)		2q37.1q37.3 deletion
Holoprosencephaly 7 (HPE7)	<i>PTCH1 patched Drosophila homolog</i>	Missense gain-of-fuction mutations /duplication? duplications?
Holoprosencephaly 9 (HPE9)/Pituitary anomalies with holoprosencephaly	<i>GLI2</i>	2q14.2 deletion
Holt-Oram syndrome	<i>TBX5</i>	12q24.21 deletion
Hunter/Mucopolysaccharidosis, type II (MPS2)	<i>IDS</i>	Xq28 deletion
Hyperekplexia and epilepsy/X-linked mental retardation (XLMR)	<i>ARHGEF9</i>	Xq11.1 deletion
Hyperglycerolemia (Glycerol kinase deficiency)	<i>GK glycerol kinase</i>	Xp21.2 deletion
Hypertension with CHD	<i>BMPR2</i>	2q33.1 deletion
Hypoparathyroidism, sensorineural deafness, and renal disease (HDR)	<i>GATA3 GATA-binding protein 3</i>	10p14 deletion duplication
Hypophosphatemic rickets, X-linked dominant	<i>PHEX</i>	Xp22.11 deletion
Hypotonia-Cystinuria syndrome/ with mitochondrial disease	<i>PPM1B, SLC3A1, PREPL</i>	2p21 deletion
Ichthyosis, X-linked (steroid sulfatase deficiency)	<i>STS steroid sulfatase deficity</i>	Xp22.31 deletion
Immunodeficiency-centromeric Instability-Facial anomalies syndrome	<i>DNMT3B</i>	20q11.21 deletion
Incontinentia pigmenti (IP)	<i>IKBKG (NEMO)</i>	Xq28 deletion
Infantile spasm syndrome, X-linked (ISSX), West, Proud, XLAG, Partington, multifocal epilepsy	<i>ARX</i>	Xp21.3 deletion
Iridogoniodysgenesis anomaly, Axenfeld-Rieger syndrome, 6p25.3 deletion syndrome	<i>FKHL7 (FOXC1)</i>	6p25.3 deletion
Jacobsen syndrome		11q24-q25 deletion
Jacobsen syndrome (JBS)/ Trigonocephaly 11q deletion		11q24.1 deletion
Johanson-Blizzard syndrome (JBS)	<i>UBR1</i>	15q15.2 deletion
Juvenile myoclonic epilepsy	<i>GABRA1</i>	5q34 deletion
Kallmann syndrome 1 (KAL1)	<i>KAL1 Kallmann syndrome 1</i>	Xp22.31 deletion
Kartagener syndrome	<i>DNAI1</i>	9p13.3 deletion

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Kartagener syndrome	<i>DNAH5</i>	5p15.2 deletion
Langer-Giedion syndrome (LGS)/trichorhinophalangeal syndrome type II (TRPS2)	<i>TRPS1 zinc finger transcription TRPS1 & EXT1 exostosin 1</i>	8q23.3-q24.11 deletion
Leri-Weill dyschondrosteosis (LWD) / short stature	<i>SHOX/SHOXY short stature homeobox</i>	Xp22.33 / Yp11.32
Lesch-Nyhan syndrome (LNS)	<i>HPRT1 (HPRT)</i>	Xq26.2 deletion
Leukodystrophy due to mitochondrial complex 1 deficiency	<i>NDUFV1</i>	11q13.2 deletion
Leukodystrophy with microdeletion 11q14.3		11q14.2q14.3 deletion
Leukodystrophy, adult onset autosomal dominant (ADLD)	<i>LMNB1</i>	5q23.2 deletion
Lissencephaly/Autism /multifocal epilepsy	<i>RELN</i>	7q22.1 deletion
Loeys-Dietz syndrome (LDS)	<i>TGFBR1</i>	9q22.33 deletion
Loeys-Dietz syndrome, type 2B (LDS2B)/Marfan syndrome, type II	<i>TGFBR2</i>	3p24.1 deletion
Lowe oculocerebrorenal syndrome	<i>OCRL</i>	Xq25 deletion
LUBS (MRXSL) / MECP2 duplication	<i>MECP2 methyl-CpG-binding protein-2</i>	Xq28 duplication
Lymphedema hereditary, II	<i>FOXC2</i>	16q24.1 deletion
Macrocephaly/seizures	<i>SYT14</i>	1q32.2 deletion
Marfan syndrome (MFS)	<i>FBN1</i>	15q21.1 deletion
MASA syndrome, X-linked mental retardation (XLMR)	<i>L1CAM</i>	Xq28 deletion
McCune-Albright syndrome	<i>GNAS</i>	20q13.32 deletion
McKusick-Kaufman syndrome (MKKS)	<i>MKKS</i>	20p12.2 deletion
Menkes disease (MNK), Cutis laxa / Occipital horn syndrome (OHS)	<i>ATP7A</i>	Xq21.1 deletion
Mental retardation	<i>ARHGAP11A, CHRM5</i>	15q13.3 deletion
Mental retardation X-linked with isolate growth hormone deficiency (MRGH)	<i>SOX3 SRY- box 3</i>	Xq27.1 deletion or duplication
Mental retardation, X-linked 21 (MRX21)	<i>IL1RAPL1</i>	Xp21.3 deletion
Mental retardation, X-linked, syndromic, Turner type/X-linked mental retardation (XLMR)	<i>HUWE1</i>	Xp11.22 deletion
Mental retardation, X-linked, with Epilepsy (XMRE)	<i>ATP6AP2</i>	Xp11.4 deletion
Metachromatic leukodystrophy	<i>ARSA</i>	22q13.33 deletion
Micophthalmia, syndromic 7 (MCOPS7)/Microphthalmia with linear skin defects (MLS)	<i>HCCS</i>	Xp22.2 deletion
Microcephaly, congenital heart disease	<i>NPM1</i>	5q35.1 deletion
Microcephaly/agenesis corpus callosum	<i>AKT3</i>	1q44 deletion
Microphthalmia syndromic (MCOPS2)	<i>BCOR</i>	Xp11.4 deletion
Microphthalmia syndromic 3 (MCOPS3)	<i>SOX2</i>	3q26.33 deletion

Phenotype	Genes	Cytogenetics
Microphthalmia syndromic 6 (MCOPS6)/Anophthalmia, pituitary hypoplasia, and ear anomalies	<i>BMP4</i>	14q22.2 deletion
Microtia	<i>DRD5 (CNV)</i>	4p16.1 deletion
Miller-Dieker lissencephaly syndrome (MDLS)	<i>LIS1, YWHAE</i>	17p13.3 deletion
Mohr-Tranebjaerg syndrome	<i>TIMM8A</i>	Xq22.1 deletion
Monogenic audiogenic seizure susceptibility	<i>MASS1 (GPR98, VLGR1)</i>	5q14.3 deletion
Monosomy 1p36 (Facial Clefting Anomalies, Generalized epilepsy with febrile seizures-3 plus (type 5), Cranial Suture Closure, Seizures)	<i>DVL1, SKI, GABRD, MMP23, KCNAB2</i>	1p36 deletion and/or duplication/triplication
Monosomy 9p syndrome		9p22.3p23
Mowat-Wilson syndrome	<i>ZEB2/ZFHX1B</i>	2q22.3 deletion
Multicentric osteolysis, nodulosis, and arthropathy (MONA)	<i>MMP2</i>	16q12.2 deletion
Muscle-eye-brain disease	<i>POMGNT1</i>	1p34.1 deletion
Myoclonic epilepsy of Lafora	<i>EPM2A</i>	6q24.3 deletion
Myoclonic epilepsy, juvenile/juvenile absence epilepsy	<i>BRD2</i>	6p21.32 deletion
Myoclonic epilepsy, neonatal, with suppression-burst pattern	<i>SLC25A22</i>	11p15.5 deletion
Nail-patella syndrome (NPS)	<i>LMX1B LIM-homeo box factor 1β</i>	9q33.3 deletion
Nance-Horan	<i>NHS</i>	Xp22.13 deletion
Nebulette	<i>NEBL</i>	10p12.31 deletion
Nephronophthisis 1/Related Joubert Syndrome (NPHP1)	<i>NPHP1, nephrocystin</i>	2q13 homozygous deletion
Neurofibromatosis I (NF1)	<i>NF1 neurofibromin 1</i>	17q11.2 deletion
Neurofibromatosis II (NF2)	<i>NF2 neurofibromin 2</i>	22q12.2 deletion
Neuromotor development delay, cerebellar ataxia, and epilepsy	<i>AF9/MLLT3</i>	9p21.3 deletion
Nievergelt syndrome	<i>LAF4/AFF3</i>	2q11.2
Noonan syndrome (NS1)	<i>PTPN11</i>	12q24.13 duplication
Noonan syndrome (NS4)	<i>SOS1</i>	2p22.1 deletion
Noonan syndrome 5 (NS5)	<i>RAF1</i>	3p25.1 deletion
Norrie disease	<i>NDP</i>	Xp11.3 deletion
Oculopharyngeal muscular dystrophy	<i>PABPN1</i>	14q11.2
Opitz BBB syndrome	<i>MID1</i>	Xp22.2 deletion
Ornithine transcarbamylase deficiency	<i>OTC</i>	Xp11.4 deletion
Orofacial cleft	<i>SUMO1</i>	2q32.2-q33 deletion
Orofaciodigital syndrome (OFD1)	<i>CXORF5 (OFD1)</i>	Xp22.2 deletion
Osler-Rendu-Weber syndrome (Hereditary hemorrhagic teleangiectasia)	<i>ACVRL1</i>	12q13.13 deletion

Phenotype	Genes	Cytogenetics
Osler-Rendu-Weber syndrome 2 (ORW2)	<i>ALK1 (ACVRL1)</i>	12q13.13 deletion
Osteogenesis imperfecta type IV	<i>COL1A1</i>	17q21.33 deletion
Osteogenesis imperfecta congenital	<i>COL1A2</i>	17q21.33 deletion
Oto-dental syndrome (dental and inner-ear disease/ocular coloboma)	<i>FGF3/FADD</i>	11q13.3 deletion
Overgrowth	<i>GPR51/GABBR2</i>	9q22.33 deletion
Ovotestes and male-to female sex-reversal XY	<i>NR5A1 (SF1)</i>	11q13.1 (9q33 deletion)?
Pallister-Killian syndrome		12p triplication
Pancreatic agenesis, congenital	<i>IPF1 (PDX1)</i>	13q12.2 deletion
Papillorenal syndrome (Renal-coloboma syndrome)	<i>PAX2</i>	10q24.31 deletion
Partial anodontia, orofacial clefting/Witkop syndrome	<i>MSX1</i>	4p16.2 deletion
Pelizaeus-Merzbacher disease (PMD)	<i>PLP1 proteolipid protein 1</i>	Xq22.2 duplication or deletion
Persistent Mullerian duct syndrome (PMDS) / Sex ambiguity	<i>AMH/MIS</i>	19p13.3 deletion
Phosphoglycerate kinase deficiency	<i>PGK1</i>	Xq21.1 deletion
Phosphoribosylpyrophosphate synthetase superactivity	<i>PRPS1</i>	Xq22.3 deletion
Pitt-Hopkins syndrome	<i>TCF4</i>	18q21.2 deletion
Pituitary hypoplasia, Oculoauriculovertebral spectrum	<i>SIX6</i>	14q23.1(14q22.2-q22.3 del)?
Polycystic kidney disease / Tuberous sclerosis (PKDTS)	<i>PKD1 polycystin 1, TSC2 tuberin</i>	16p13.3 deletion
Potocki-Lupski syndrome (PTLS)	<i>RAI1 retinoic acid-induced gene 1</i>	17p11.2 duplication
Potocki-Shaffer syndrome	<i>ALX4 Aristaless-like-4 and/or EXT2 exostosin 2</i>	11p11.2 deletion
Prader –Willi syndrome (PWS)	<i>PWS-SRO, SNRPN, HBII-85 C/D box, NDN necdin, all snoRNAs , snoRNA cluster</i>	15q11.2-q12 deletion
Prader –Willi syndrome (PWS) imprinting center	PWS imprinting center	15q11.2 deletion
Prader-Willi syndrome-like/Obesity	<i>SIM1</i>	6q16.3 deletion
Pseudoachondroplasia (PSACH)	<i>COMP</i>	19p13.11 deletion
Pseudoxanthoma elasticum, forme fruste	<i>ABCC6</i>	16p13.11 deletion
Pyruvate dehydrogenase deficiency	<i>PDHA1</i>	Xp22.12 deletion
Renal cysts and diabetes (RCAD)/epilepsy	<i>TCF2</i>	17q12 deletion/duplication
Renpenning syndrome 1 (RENS1)/Sutherland-Haan XLMR syndrome/Golabi-Ito-Hall syndrome	<i>PQBP1</i>	Xp11.23 deletion
Retinoblastoma (RB1)	<i>RB1 retinoblastoma 1</i>	13q14.2 deletion
Retinoschisis, X-juvenile, juvenile (RS1)	<i>RS1</i>	Xp22.13 deletion

Phenotype	Genes	Cytogenetics
Retinoschisis, X-linked juvenile (RS1)	<i>XLRS1</i>	Xp22.13 deletion
Rett syndrome (RTT)	<i>MECP2 methyl-CpG-binding protein-2</i>	Xq28 deletion
Rett-like, Infantile spasm syndrome, X-linked (ISSX)	<i>CDKL5</i>	Xp21.3 deletion
Rieger syndrome, type 1 (RIEG1)	<i>PITX2</i>	4q25 deletion
Robinow syndrome/brachydactyly type B1 (BDB1)	<i>ROR2</i>	9q22.31 deletion
Rubinstein-Taybi Syndrome	<i>EP300</i>	22q13.2
Rubinstein-Taybi syndrome (RSTS)	<i>TRAP1, DNASE1, CREBBP, CREB_binding protein?</i>	16p13.3 deletion
Sacral/anorectal malformation syndrome		6q25.3 deletion
Saethre-Chotzen syndrome (SCS)	<i>TWIST1</i>	7p21.1 deletion
Schizencephaly / multifocal epilepsy	<i>EMX2</i>	10q26.11 deletion
Schizophrenia & epilepsy	<i>CNTNAP2</i>	7q36.1 deletion
Schwartz-Jampel syndrome, type1 (SJS1)	<i>HSPG2</i>	1p36.12 deletion
Seizures	<i>DGKD</i>	2q37.1 deletion
Seizures, benign familial neonatal-infantile	<i>SCN2A</i>	2q23-q24.3 deletion
Severe myoclonic epilepsy of infancy (SMEI) or Dravet syndrome; Generalized epilepsy with febrile seizures plus; GEFS+	<i>SCN1A</i>	2q24.3 deletion
Sex reversal, autosomal dominant 2 (SRA2)		9p24.3 deletion
Shah-Waardenburg syndrome	<i>EDNRB</i>	13q22.3 deletion
Shah-Waardenburg syndrome	<i>EDN3</i>	20q13.32 deletion
Short stature, pituitary and cerebellar defects, & small sella turcica	<i>LHX4</i>	1q25.2 deletion
Siderius type X-linked mental retardation / Cleft lip/palate	<i>PHF8</i>	Xp11.22 deletion
Simpson-Golabi-Behmel syndrome type 1;(SGBS1)	<i>GPC3</i>	Xq26.2 deletion
Smith-Lemli-Opitz syndrome (SLOS)	<i>DHCR7</i>	11q13.4 deletion
Smith-Magenis syndrome (SMS)	<i>RAI1 retinoic acid-induced gene 1</i>	17p11.2 deletion
Sotos syndrome	<i>NSD1 nuclear receptor binding Su-var</i>	5q35.3 deletion
Speech delay/Autism	<i>FOXP2</i>	7q31.1 deletion
Split hand/foot malformation 1 with hearing loss		7q21.3 deletion
Split hand/foot malformation 4 (SHFM4)	<i>TP73L (TP63)</i>	3q28 deletion
Split-hand/foot malformation (SHFM5)	<i>DLX1/DLX2</i>	2q31.1 deletion
Split-hand/foot malformation-3 (SHFM3)		10p14 duplication

Phenotype	Genes	Cytogenetics
Split-hand/split-foot malformation 1 (SHFM1)	<i>SHFM1</i>	7q21.3
Split-hand/split-foot malformation 3 (SHFM3)	<i>SHFM3/FBXW4</i>	10q24.32 deletion
Spondylocostal dysostosis autosomal recessive; (SCDO1)	<i>DLL3</i>	19q13.2 deletion
Stickler syndrome, type II (STL2)	<i>COL11A1</i>	1p21.1 deletion
Stickler syndrome, type II (STL2)	<i>COL11A1</i>	
Stocco dos Santos XLMR	<i>SHROOM4 (KIAA1202)</i>	Xp11.22 deletion
Susceptibility to adolescent-onset idiopathic generalized epilepsy	<i>ME2</i>	18q21.2 deletion
Symphalangisms proximal, Multiple synostoses syndrome 1, Stapes ankylosis with broad thumb and toes	<i>NOG</i>	17q22 deletion
Synpolydactyly (SPD1); HOXD deletion syndrome	<i>HOXD13</i>	2q31-q32 deletion
Telangiectasia, hereditary hemorrhagic, of rendu, osler, and weber; (HHT)	<i>ENG</i>	9q34.11 deletion
Tetralogy of Fallot / ASD secundum with atrioventricular conduction defects Microcephaly	<i>NKX2-5 (CSX)</i>	5q35.2 duplication
Tetralogy of Fallot/diaphragmatic hernia	<i>ZFPM2/FOG2</i>	8q23.1 deletion
Thrombocytopenia–Absent Radius Syndrome		1q21.1 deletion
Thrombocytopenia-absent radius syndrome (TAR)		1q21.1 deletion
Timothy syndrome	<i>CACNA1C</i>	12p13.33 deletion
Tooth agenesis, selective 3 (STHAG3) Hypodontia/Oligodontia 3	<i>PAX9</i>	14q13.3 deletion
Townes-Brocks syndrome	<i>SALL1</i>	16q12.1 deletion
Treacher-Collins-Franceschetti syndrome (TCOF)	<i>TCOF1</i>	5q33.1 deletion
Trichorhinophalangeal syndrome I (TRPS1)	<i>TRPS1 zinc finger transcription TRPS1</i>	8q23.3 deletion
Trigonocephaly (9p deletion)		9p23 deletion
Triphalangeal thumb Syndrome (Polydactyly, preaxial II; PPD2)	ZRS regulator of <i>SHH</i>	7q36.3
Tuberous sclerosis 1 (TSC1)/Autism/multifocal epilepsy	<i>TSC1 hamartin</i>	9q34.13
Tuberous sclerosis 2 (TSC2)/Autism/multifocal epilepsy	<i>TSC2 tuberin</i>	16p13.3 deletion
Ulnar-mammary syndrome	<i>TBX3</i>	12q24.21 deletion/duplication
Van Buchem disease	<i>SOST</i>	17q21.31
Van Buchem disease(Hyperostosis corticalis generalisata)	<i>SOST</i> and downstream	17q21.31
Van der Woude syndrome (VWS)	<i>IRF6</i>	1q32.2 deletion
Vascular endothelial growth factor (VEGF)	<i>VEGFA (VEGF)</i>	6p21.1 deletion
Von Hippel-Lindau syndrome	<i>VBP1</i>	Xq28 deletion
Von Hippel-Lindau syndrome (VHL)	<i>VHL</i>	3p25.3 deletion
Waardenburg syndrome, type 1	<i>PAX3</i>	2q36.1 deletion

Phenotype	Genes	Cytogenetics
Waardenburg syndrome, type IIA (WS2A)	<i>MITF</i> (<i>Microphthalmia-associated transcription factor</i>)	3p14.1 deletion
Waardenburg-Shah syndrome	<i>SOX10</i>	22q13.1 deletion
Walker-Warburg syndrome	<i>POMT1</i>	9q34.13 deletion
Williams-Beuren syndrome (WBS)	<i>ELN elastin, LIMK1 LIM kinase 1, RFC2, CYLN2</i>	7q11.23 deletion
Wilms tumor 1 (WT1)	<i>WT1 Wilms tumor 1 gene</i>	11p13 deletion
Wilms tumor -aniridia genitourinary anomalies--mental retardation syndrome (WAGR)	<i>PAX6 & WT1</i>	11p13 deletion
Wolff-Parkinson-White	<i>BMP2 Bone morphogenetic proteins2</i>	20p12.3 deletion
Wolf-Hirschhorn syndrome (WHS)	Multiple (<i>LETM1, WFS1 WHCR1/2</i>)	4p16.3 deletion
X-inactivation, familial skewed, XIST deficiency	<i>XIST</i>	Xq13.2 usually ring X chromosome
X-linked heterotaxy, ZIC3	<i>ZIC3 zinc finger protein cerebellum 3</i>	Xq26.3 deletion
X-linked lymphoproliferative syndrome (XLP1)	<i>SH2D1A</i>	Xq25 deletion
X-linked mental retardation (XLMR)	<i>CASK</i>	Xp11.4 deletion
X-linked mental retardation (XLMR)	<i>HSD17B10/HADH2</i>	Xp11.22 deletion
X-linked mental retardation (XLMR)	<i>ELK1</i>	Xp11.23 deletion
X-linked mental retardation (XLMR)	<i>SLC38A5</i>	Xp11.23 deletion
X-linked mental retardation (XLMR)	<i>ZNF41</i>	Xp11.3 deletion
X-linked mental retardation (XLMR)	<i>VCX3A</i>	Xp22.31 deletion
X-linked mental retardation (XLMR)	<i>KLF8</i>	Xp11.21 deletion
X-linked mental retardation (XLMR)	<i>GDI1</i>	Xq28 deletion
X-linked mental retardation (XLMR)	<i>NXF5</i>	Xq22.1 deletion
X-linked mental retardation (XLMR)	<i>ZNF261 (ZMYM3)</i>	Xq13.3 deletion
X-linked mental retardation (XLMR)	<i>KIAA2022</i>	Xq13.3 deletion
X-linked mental retardation (XLMR)	<i>ACSL4</i>	Xq22.3 deletion
X-linked mental retardation (XLMR)	<i>PAK3</i>	Xq22.3 deletion
X-linked mental retardation (XLMR)	<i>AGTR2</i>	Xq23 deletion
X-linked mental retardation (XLMR)	<i>ZDHH9</i>	Xq25 deletion
X-linked mental retardation (XLMR)	<i>FMR2 (AFF2)</i>	Xq28 deletion
X-linked mental retardation (XLMR) (MRX45)	<i>ZNF81</i>	Xp11.23 deletion

Phenotype	Genes	Cytogenetics
X-linked mental retardation (XLMR) (MRX46)	<i>ARHGEF6</i>	Xq26.3 deletion
X-linked mental retardation (XLMR) (MRX46)	<i>ARHGEF6</i>	Xq28 deletion
X-linked mental retardation (XLMR) (MRX58)	<i>TSPAN7 (TM4SF2)</i>	Xp11.4 deletion
X-linked mental retardation (XLMR) (MRX59)	<i>AP1S2</i>	Xp22.2 deletion
X-linked mental retardation (XLMR) (MRX9)	<i>FTSJ1</i>	Xp11.23 deletion
X-linked mental retardation (XLMR) (MRX90)/Autism	<i>DLG3</i>	Xq13.1 deletion
X-linked mental retardation (XLMR) (MRX91)	<i>ZDHHC15</i>	Xq13.3 deletion
X-linked mental retardation (XLMR) (MRX94)	<i>GRIA3</i>	Xq25 deletion
X-linked mental retardation (XLMR) with short stature, small testes muscle wasting, and tremor	<i>CUL4B</i>	Xq24 deletion
X-linked mental retardation (XLMR), syndromic, Chrystianson type	<i>SLC9A6</i>	Xq26.3 deletion
X-lissencephaly/multifocal spasms (LISX)	<i>DCX</i>	Xq22.3 deletion
XLMR / Snyder-Robinson syndrome	<i>SMS spermine synthase gene</i>	Xp22.11 deletion
XLMR/Autism	<i>JARID1C/JARID1D</i>	Xp11.2/Y11.222 deletion
Xp11.3 deletion with mental retardation(XLMR)	<i>ZNF674</i>	Xp11.3 deletion
Xq/Yq pseudoautosomal		Xq28 deletion

*OMIM - Online Mendelian Inheritance of Man (<http://www.ncbi.nlm.nih.gov/omim/>)

**PMID - PubMed (<http://www.ncbi.nlm.nih.gov/entrez/>)

***GeneTests - (<http://www.genetests.org/>)

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***Percent Detection Rates are based on the rates cited in the Selected PMID