



ExonArrayDx

Gene list for clinical testing

Exon-level deletion/duplication testing is available for more than 375 clinically significant genes. ExonArrayDx is an oligonucleotide microarray-based (oligo array CGH) test developed by GeneDx to examine one or more genes for full or partial gene deletions or duplications **at the exon level**. The following list contains clinically relevant genes (and their associated disorders) that are available for deletion/duplication testing by ExonArrayDx. Deletions/duplications of these genes have been published in the literature. ExonArrayDx can be ordered for one specific gene or for a custom panel of clinically related genes (up to 10 genes selected by the ordering physician or genetic counselor). Please use the ExonArrayDx submission form to request testing. **This list is searchable—please use the search function in any PDF reader software to find specific entries.**

Genes listed individually by disease name

| Disorder | Gene | Disorder | Gene |
|--|---------------------|---|---------------------|
| 17-alpha-Hydroxylase-Deficient Congenital Adrenal Hyperplasia | CYP17A1 | Anterior segment ocular dysgenesis/Axenfeld-Rieger syndrome | PITX2 |
| 17-beta Hydroxysteroid Dehydrogenase III Deficiency | HSD17B3 | Anterior segment ocular dysgenesis/Axenfeld-Rieger syndrome | FOXC1 |
| 17-Beta-Hydroxysteroid Dehydrogenase X Deficiency | HADH2 | Antithrombin III deficiency (thrombophilia) | Serpinc1 |
| 3-methylcrotonyl-CoA carboxylase deficiency | MCCC1 | Antley-Bixler Syndrome (ABS), POR deficiency | POR |
| 3-methylcrotonyl-CoA carboxylase deficiency | MCCC2 | Aplastic anemia, dyskeratosis congenita, pulmonary fibrosis | TERT |
| 3-Methylglutaconic Aciduria Type 1 | AUH | Arginase Deficiency | ARG1 |
| 46,XY Disorder of Sex Development | NR5A1 | Argininosuccinate Lyase Deficiency | ASL |
| 46,XY Disorder of Sex Development | DHH | Aromatase deficiency, pseudohermaphroditism | CYP19A1 |
| Achromatopsia | CNGA3 | Arylsulfatase A Deficiency | ARSA |
| Achromatopsia | CNGB3 | Ataxia telangiectasia | ATM |
| Achromatopsia | GNAT2 | Atrial septal defect with atrioventricular conduction defects | NKX2-5 |
| ADA Severe Combined Immunodeficiency, Autosomal Recessive (ADA SCID), Adenosine Deaminase Deficiency | ADA | Autism spectrum; atypical Rett; epileptic encephalopathy | CDKL5 |
| Adenomatous polyposis colorectal cancer | MUTYH | Autism susceptibility locus | AHI1 |
| Adrenal hypoplasia congenita, 46,XY disorder of sex development | NROB1 | Autism susceptibility locus | AUTS2 |
| Agammaglobulinemia, X-linked | BTK | Autism susceptibility locus; 16p13.3 deletion syndrome | A2BP1 |
| Alagille syndrome | JAG1 | Autism susceptibility locus; 22q13.3 deletion syndrome | SHANK3 |
| Albright Hereditary Osteodystrophy, PHP1A,PPHP, POH | GNAS | Autoimmune Lymphoproliferative syndrome (ALPS) Type Ia | TNFRSF6/FAS |
| Alpha-L-Fucosidase Deficiency | FUCA1 | Autoimmune Lymphoproliferative syndrome (ALPS) Type IIA and IIB | CASP8 |
| Alport syndrome | COL4A5 | Autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy syndrome (APECED), Autoimmune polyglandular syndrome | AIRE |
| Amyloid polyneuropathy Andrade or Portuguese type | TTR | Autosomal Dominant Mental Retardation 1 | MBD5 |
| Androgen insensitivity syndrome, Testicular feminization syndrome | AR | Autosomal Dominant Mental Retardation 2, Autosomal Recessive Hyper-IgE Recurrent Infection Syndrome | DOCK8 |
| Angelman syndrome | UBE3A | Autosomal hypohidrotic ectodermal dysplasia | EDAR |
| Aniridia | DCDC1 | Autosomal Recessive Leber Congenital Amaurosis (LCA) | AIPL1 |
| Aniridia | ELP4 | Bannayan-Riley-Ruvalcaba syndrome, Cowden syndrome | PTEN |
| Aniridia | PAX6 | Beckwith-Wiedeman Syndrome/Russell-Silver Syndrome | IGF1R |
| Anophthalmia/microphthalmia | BMP4 | Best macular dystrophy | VMD2 (BEST1) |
| Anophthalmia/microphthalmia | CHX10 (VSX2) | Beta Thalassemia | HBB |
| Anophthalmia/microphthalmia | OTX2 | Beta-ketothiolase Deficiency | ACAT1 |
| Anophthalmia/microphthalmia | RAX | Biotinidase deficiency | BTD |
| Anophthalmia/microphthalmia | SIX6 | Birt-Hogg-Dube syndrome | FLCN |
| Anophthalmia/microphthalmia | SOX2 | Blackfan-Diamond syndrome | RPS19 |
| Anophthalmia/microphthalmia STRA6 | STRA6 | Blepharophimosis, ptosis, and epicanthus inversus (BPES) | FOXL2 |
| Anterior segment mesenchymal dysgenesis | PITX3 | Bloom syndrome | RECQL3 (BLM) |
| Anterior segment ocular dysgenesis | FOXE3 | Branchiootic syndrome | SIX1 |
| Anterior segment ocular dysgenesis (microphthalmia) | BCOR | | |





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| Branchio-oto-renal syndrome type 1 | EYA1 |
| Brugada syndrome | CACNB2 |
| Brugada syndrome 2 | GPD1L |
| CA4-Related Retinitis Pigmentosa | CA4 |
| Campomelic dysplasia | SOX9 |
| Canavan disease | ASPA |
| Carbamoyl phosphate synthetase I deficiency | CPS1 |
| Carney Complex | PRKAR1A |
| Carnitine palmitoyltransferase I deficiency | CPT1A |
| Carnitine palmitoyltransferase II deficiency | CPT2 |
| Carpenter disease | RAB23 |
| Cartilage hair hypoplasia, Metaphyseal dysplasia without hypotrichosis, Spondylometaphyseal dysplasia, Anauxetic dysplasia | RMRP |
| Cerebral cavernous malformation | CCM1 (KRIT1) |
| Cerebral cavernous malformation | CCM2 |
| Cerebral cavernous malformation 3 | PDCD10 |
| Chanarin-Dorfman Syndrome, Neutral lipid storage disease with ichthyosis (NLSLSD) | ABHD5 |
| CHARGE syndrome | CHD7 |
| Chondrodysplasia punctata | ARSE |
| Choroideremia | CHM |
| Chronic granulomatous disease | CYBA |
| Chronic granulomatous disease | CYBB |
| Chronic granulomatous disease | NCF2 |
| Cleidocranial dysplasia | RUNX2 |
| Cobalamin C deficiency | MMACHC |
| Cockayne syndrome A | ERCC8 |
| Cockayne syndrome B | ERCC6 |
| Coffin-Lowry Syndrome | RSK2 (RPS6KA3) |
| Cohen syndrome | COH1 (VPS13B) |
| Combined pituitary hormone deficiency | PROP1 |
| Combined Saposin Deficiency, Saposin A, B, C Deficiency | PSAP |
| Cone Dystrophy 3 | GUCA1A |
| Cone Rod Dystrophy 4 | PDE6C |
| Cone-rod dystrophy, autosomal dominant | CRX |
| Cone-rod dystrophy, autosomal dominant | GUCY2D |
| Cone-rod dystrophy, autosomal dominant | RDS (PRPH2) |
| Congenital amegakaryocytic | MPL |
| Congenital Disorder of Glycosylation Ib | MPI |
| Congenital Disorder of Glycosylation Ic | ALG6 |
| Congenital insensitivity to pain with anhidrosis (CIPA) | NTRK1 |
| Congenital neutropenia, recessive | HAX1 |
| Congenital recessive ichthyosis | ALOX12B |
| Congenital recessive ichthyosis | ALOXE3 |

| Disorder | Gene |
|--|-------------------|
| Congenital recessive ichthyosis | NIPAL4 (Ichthyin) |
| Congenital Stationary Night Blindness, Type 1B | GRM6 |
| Congenital Stationary Night Blindness, Type 1C | TRPM1 |
| Congenital Stationary Night Blindness, Type 2B | CABP4 |
| Corneal dystrophy | CYP4V2 |
| Costello syndrome | HRAS |
| Craniofrontonasal syndrome | EFNB1 |
| Currarino syndrome | HLXB9 |
| Darier disease | ATP2A2 |
| Dent disease | CLCN5 |
| DiGeorge/velocardiofacial syndromes | TBX1 |
| DiGeorge-like phenocopy | CRKL |
| Dihydropteridine reductase deficiency | QDPR |
| Duane-radial ray syndrome | SALL4 |
| Dyskeratosis congenita | DKC1 |
| Dyskeratosis congenita | TERC (hTR) |
| Dyskeratosis Congenita | TINF2 |
| Ectodermal Dysplasia, Odonto-onycho-dermal dysplasia (OODD), Schopf-Schulz-Passarge Syndrome (SSPS) | WNT10A |
| EDN3-Related Hirschsprung Disease | EDN3 |
| Ehlers-Danlos syndrome type 4 | COL3A1 |
| Ellis-van Creveld Syndrome | EVC |
| Epidermolysis bullosa, dystrophic type | COL7A1 |
| Epidermolysis bullosa, junctional type | COL17A1 |
| Epidermolysis bullosa, junctional type | LAMA3 |
| Epidermolysis bullosa, junctional type | LAMC2 |
| Epidermolysis bullosa, junctional with pyloric atresia (JEB-PA) | ITGB4 |
| Escobar Syndrome/Multiple Pterygium Syndrome | CHRNA3 |
| Ethylmalonic aciduria | ETHE1 |
| Fabry Disease | GLA |
| Familial Atrial Fibrillation | KCNA5 |
| Familial exudative vitreoretinopathy | FZD4 |
| Familial exudative vitreoretinopathy | LRP5 |
| Familial gastric cancer | CDH1 |
| Familial hypercholesterolemia | LDLR |
| Familial hypocalciuric hypercalcemia, Neonatal severe primary hyperparathyroidism; Autosomal Dominant Hypocalcemia, Familial isolated hypoparathyroidism | CASR |
| Fanconi Anemia | FANCC |
| Feingold syndrome | MYCN |
| Focal dermal hypoplasia (Goltz syndrome) | PORCN |
| FSCN2-Related Retinitis Pigmentosa | FSCN2 |
| Fundus albipunctatus | RDH5 |





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| Galactokinase deficiency | GALK1 |
| Galactosemia (galactosyltransferase deficiency) | GALT |
| Glanzmann thrombasthenia | ITGB3 |
| Glutaric aciduria type I | GCDH |
| Glutaric aciduria type II (Multiple acyl-CoA dehydrogenase def.) | ETFA |
| Glutaric aciduria type II (Multiple acyl-CoA dehydrogenase def.) | ETFB |
| Glutaric aciduria type II (Multiple acyl-CoA dehydrogenase def.) | ETFDH |
| Glutathione synthetase deficiency | GSS |
| Glycogen storage disease type IV | GBE1 |
| Gonadal dysgenesis | SRY |
| Gorlin syndrome | PTCH1 |
| Growth hormone insensitivity syndrome | GHR |
| GTP cyclohydrolase 1 deficiency; dopa-responsive dystonia | GCH1 |
| Guanidinoacetate Methyltransferase Deficiency | GAMT |
| GUCA1B-Related Retinitis Pigmentosa | GUCA1B |
| Hailey-Hailey disease (HHD; familial benign chronic pemphigus) | ATP2C1 |
| Harlequin ichthyosis (HI), Lamellar ichthyosis type 2 (LI-2) | ABCA12 |
| Hemophilia A | F8 |
| Hemophilia B | F9 |
| Hereditary angioedema | C1NH |
| Hereditary hemorrhagic telangiectasia | ENG |
| Hereditary leiomyomatosis and renal cell cancer, Fumarate hydratase deficiency | FH |
| Hereditary multiple exostoses | EXT1 |
| Hereditary multiple exostoses | EXT2 |
| Hereditary paraganglioma-pheochromocytoma syndromes | SDHB |
| Hereditary paraganglioma-pheochromocytoma syndromes | SDHC |
| Hereditary paraganglioma-pheochromocytoma syndromes | SDHD |
| Hereditary polyposis coli (colon cancer) | APC |
| Hereditary schwannomatosis/malignant rhabdoid tumor | SMARCB1 |
| Hermansky-Pudlak syndrome | HPS1 |
| Hermansky-Pudlak Syndrome 2 | AP3B1 |
| HMG co-A lyase deficiency | HMGCL |
| Holocarboxylase synthetase deficiency | HLCS |
| Holoprosencephaly | SHH |
| Holoprosencephaly | SIX3 |
| Holoprosencephaly | ZIC2 |
| Holt-Oram syndrome | TBX5 |
| Homocystinuria (cystathionine beta-synthase deficiency) | CBS |
| Hydrocephalus, X-linked | L1CAM |
| Hyper-IgD syndrome (HIDS) | MVK |
| Hyper-IgM syndrome type 1 | CD40LG |
| Hyperparathyroidism-jaw tumor syndrome | HRPT2 (CDC73) |
| Hypomyelination, Global cerebral | SLC25A12 |

| Disorder | Gene |
|--|----------------|
| Hypoparathyroidism, deafness, and renal disease | GATA3 |
| Hypophosphatemic rickets (autosomal dominant); Pseudo-vitamin D deficiency rickets (autosomal recessive) | CYP27B1 |
| Hypophosphatemic Rickets, Autosomal Recessive 1 | DMP1 |
| Hypophosphatemic rickets, X-linked | PHEX |
| IL7R Severe Combined Immunodeficiency (autosomal recessive) | IL7R |
| Immunodeficiency syndrome with Hyper IgM type 2 | AICDA |
| Inclusion body myopathy, hereditary (HIBM) | GNE |
| IRAK4 deficiency | IRAK4 |
| Isobutyryl-CoA dehydrogenase deficiency | ACAD8 |
| Isolated Persistent Hypermethioninemia | MAT1A |
| Isovaleric acidemia | IVD |
| Jalili syndrome | CNNM4 |
| Joubert syndrome | CEP290 |
| Joubert Syndrome | INPP5E |
| Junctional epidermolysis bullosa | LAMB3 |
| Juvenile polyposis syndrome; pancreatic cancer | SMAD4 |
| Kallmann syndrome | FGFR1 |
| KLHL7-Related Retinitis Pigmentosa | KLHL7 |
| Krabbe disease | GALC |
| Lamellar ichthyosis | TGM1 |
| L-Arginine:Glycine Amidinotransferase Deficiency | GATM |
| LCA5-Related Leber Congenital Amaurosis | LCA5 |
| Leber congenital amaurosis, autosomal recessive | CRB1 |
| Leber congenital amaurosis, autosomal recessive | RDH12 |
| Leber congenital amaurosis, autosomal recessive | RPGRIP1 |
| Leber congenital amaurosis, autosomal recessive | TULP1 |
| Leri-Weill dyschondrosteosis; short stature | SHOX |
| Li-Fraumeni syndrome | TP53 |
| Li-Fraumeni syndrome type 2 | CHEK2 |
| Limb-girdle muscular dystrophy type 2A | CAPN3 |
| Long QT syndrome | AKAP9 |
| Long QT syndrome | KCNE3 |
| Long QT syndrome 1 | KCNQ1 |
| Long QT Syndrome 10 | SCN4B |
| Long QT Syndrome 12 | SNTA1 |
| Long QT syndrome 2 | KCNH2 |
| Long QT syndrome 3 | SCN5A |
| Long QT syndrome 5 | KCNE1 |
| Long QT syndrome 6 (acquired susceptibility) | KCNE2 |
| Long QT syndrome 7 | KCNJ2 |
| Long-chain acyl-CoA dehydrogenase deficiency, Mitochondrial trifunctional protein deficiency | HADHA |





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| Lowie syndrome | OCRL |
| Lowie syndrome | OCRL1 |
| Lysosomal acid lipase deficiency | LIPA |
| Malonic aciduria | MLYCD |
| Maple syrup urine disease | BCKDHA |
| Maple syrup urine disease | BCKDHB |
| Maple syrup urine disease | DBT |
| Maple syrup urine disease, type III | DLD |
| Marfan syndrome, Shprintzen-Goldberg craniosynostosis, Weill-Marchesani syndrome | FBN1 |
| Medium-chain Acyl-CoA Dehydrogenase (MCAD) deficiency | ACADM |
| MERTK-Related Retinitis Pigmentosa | MERTK |
| Metaphyseal chondrodysplasia | COL10A1 |
| Methylmalonic acidemia - cbl A type | MMAA |
| Methylmalonic acidemia - cbl B type | MMAB |
| Methylmalonic acidemia - mutase deficiency | MUT |
| Mitochondrial trifunctional protein deficiency | HADHB |
| MMADHC-Related Methylmalonic Aciduria | MMADHC |
| Molybdenum cofactor deficiency | MOCS2 |
| Morquio B syndrome | GLB1 |
| Morquio syndrome A (mucopolysaccharidosis type IVA) | GALNS |
| Mucopolidosis I | NEU1 |
| Mucopolidosis IV | MCOLN1 |
| Mucopolysaccharidosis Type I (Hurler Syndrome, Hurler-Scheie Syndrome, Scheie Syndrome) | IDUA |
| Mucopolysaccharidosis Type IIIA | SGSH |
| Mucopolysaccharidosis Type IIIB | NAGLU |
| Mucopolysaccharidosis Type IIIC | HGSNAT |
| Mucopolysaccharidosis Type IIID | GNS |
| Mucopolysaccharidosis Type VI | ARSB |
| Multiple endocrine neoplasia type 1 | MEN1 |
| Multiple Sulfatase Deficiency | SUMF1 |
| Multiple Synostoses Syndrome | NOG |
| N-Acetylglutamate Synthase Deficiency | NAGS |
| Nail-patella syndrome | LMX1B |
| Nemaline myopathy | NEB |
| Nephrogenic diabetes insipidus | AVPR2 |
| Nephronophthisis | NPHP1 |
| Nephropathic cystinosis | CTNS |
| Netherton syndrome (NTS), Ichthyosis linearis circumflexa | SPINK5 |
| Neurofibromatosis type 2 | NF2 |
| Niemann Pick Type A and B | SMPD1 |
| Niemann Pick type C | NPC1 |

| Disorder | Gene |
|--|----------------------|
| Niemann Pick type C | NPC2 |
| Nijmegen breakage syndrome | NBS1 (NBN) |
| NR2E3-Related Retinitis Pigmentosa | NR2E3 |
| NRL-Related Retinitis Pigmentosa | NRL |
| NYX-Related X-Linked Congenital Stationary Night Blindness | NYX |
| Ocular coloboma | GDF6 |
| Oculocutaneous albinism | OCA2 |
| Oguchi Disease 2 | GRK1 |
| Optic atrophy 1 | OPA1 |
| Optic nerve hypoplasia/septo optic dysplasia | HESX1 |
| Ornithine transcarbamylase deficiency | OTC |
| Oro-facial-digital syndrome | CXORF5 (OFD1) |
| Osler-Rendu-Weber syndrome type 2 | ACVRL1 |
| Osteogenesis imperfecta type 1 and 4 | COL1A1 |
| Osteogenesis imperfecta | COL1A2 |
| Overgrowth Syndrome | RNF135 |
| Peters-plus syndrome | B3GALTL |
| Peutz-Jeghers syndrome | STK11 |
| Phenylketonuria | PAH |
| Pituitary dwarfism | LHX3 |
| Pituitary dwarfism type 1, short stature | GH1 |
| Pompe disease (glycogen storage disease type II) | GAA |
| Primary carnitine deficiency (carnitine uptake defect) | SLC22A5 |
| Primary congenital glaucoma | CYP1B1 |
| Primary open angle glaucoma | MYOC |
| Primary open angle glaucoma | OPTN |
| PROM1-Related Retinitis Pigmentosa | PROM1 |
| Propionic acidemia | PCCA |
| Propionic acidemia | PCCB |
| Pseudachondroplasia | COMP |
| Pseudohypoparathyroidism Type IB | STX16 |
| Renal cell carcinoma | FHIT |
| Renal coloboma syndrome | PAX2 |
| Renal cysts and diabetes syndrome | HNF1B |
| Retinal Cone Dystrophy 3 | KCNV2 |
| Retinitis pigmentosa 46 | IDH3B |
| Retinitis pigmentosa, autosomal dominant | PRPF31 |
| Retinitis pigmentosa, autosomal dominant | PRPF8 |
| Retinitis pigmentosa, autosomal dominant | RP1 |
| Retinitis pigmentosa, autosomal dominant | PRPF3 |
| Retinitis pigmentosa, autosomal dominant | RHO |
| Retinitis pigmentosa, autosomal recessive | CNGA1 |





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| Retinitis pigmentosa, autosomal recessive | PDE6A |
| Retinitis pigmentosa, autosomal recessive | PDE6B |
| Retinitis pigmentosa, autosomal recessive | RPE65 |
| Retinitis pigmentosa, autosomal recessive | USH2A |
| Retinitis pigmentosa, autosomal recessive | CNGA1 |
| Retinitis pigmentosa, autosomal recessive | EYS |
| Retinitis pigmentosa, X-linked | RP2 |
| Retinitis pigmentosa, X-linked | RPGR |
| Retinoblastoma | RB1 |
| Retinoschisis, X-linked | RS1 (XLR51) |
| Rett syndrome/male progressive neurodevelopmental syndrome | MECP2 |
| Roberts syndrome | ESCO2 |
| ROM1-Related Retinitis Pigmentosa | ROM1 |
| Rothmund-Thompsen syndrome | RECQL4 |
| Rubenstein-Taybi syndrome | CREBBP |
| Saethre-Chotzen syndrome | TWIST1 |
| Severe combined immune deficiency | JAK3 |
| Severe combined immune deficiency | RAG1 |
| Severe combined immune deficiency | RAG2 |
| Severe combined immune deficiency with radiation sensitivity | DCLRE1C |
| Severe combined immunodeficiency (autosomal recessive), Omenn syndrome | DCLRE1C |
| Short stature, pituitary and cerebellar defects, small sella turcica | LHX4 |
| Short/Branched Chain Acyl-CoA dehydrogenase (SBCAD) deficiency | ACADSB |
| Short-chain Acyl-CoA Dehydrogenase (SCAD) Deficiency | ACADS |
| Simpson-Golabi-Behmel syndrome type 1 | GPC4 |
| Simpson-Golabi-Behmel syndrome type 1; Wilms' tumor | GPC3 |
| Sjogren-Larsson syndrome (SLS) | ALDH3A2 |
| SMC1A-Related Cornelia de Lange Syndrome | SMC1A |
| Smith-Lemli-Opitz syndrome | DHCR7 |
| Smith-Magenis syndrome | RAI1 |
| Sotos syndrome | NSD1 |
| SPATA7-Related Leber Congenital Amaurosis | SPATA7 |
| Spinal muscular atrophy with respiratory distress type 1 (SMARD1) | IGHMBP2 |
| Split-hand/foot malformation | DLX5 |
| Spondyloepiphyseal Dysplasia Tarda, X-Linked | TRAPPC2 |
| Stargardt Disease, Cone Rod Dystrophy, Autosomal Recessive Retinitis Pigmentosa | ABCA4 |
| Stargardt like macular dystrophy, autosomal dominant | ELOVL4 |
| Stickler syndr., Kniest dyspl., spondyloepiphyseal dyspl. congenita | COL2A1 |
| Synpolydactyly type 1/syndactyly | HOXD13 |
| Tay-Sachs disease (hexosaminidase A deficiency) | HEXA |
| Tetralogy of Fallot | ZFPM2 |
| Thoracic Aortic Aneurysms and Dissections | ACTA2 |

| Disorder | Gene |
|---|-----------------------|
| Townes-Brock syndrome | SALL1 |
| Triple-A syndrome (Allgrove syndrome; Achalasia-Addisonianism-Alacrima) | AAAS |
| Tyrosine hydroxylase deficiency, Autosomal recessive infantile Parkinsonism | TH |
| Tyrosinemia type II | TAT |
| Tyrosinemia type I | FAH |
| Tyrosinemia type III | HPD |
| Ulnar-mammary syndrome | TBX3 |
| Van der Woude syndrome | IRF6 |
| Ventricular tachycardia, catecholaminergic polymorphic 1 | RYR2 |
| Very Long-chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency | ACADVL |
| von Hippel Lindau syndrome | VHL |
| Waardenburg syndrome type IIA | MITF |
| Waardenburg syndrome type IIE | SOX10 |
| Waardenburg syndrome type I, III | PAX3 |
| WAGR; Denys-Drash syndrome; Frasier syndrome | WT1 |
| Werner syndrome | RECQL2 (WRN) |
| Williams-Beuren syndrome | ELN |
| Williams-Beuren syndrome | MAGI2 |
| Wolfram syndrome 1; diabetes insipidus and mellitus with optic atrophy and deafness | WFS1 |
| Xeroderma pigmentosum | XPC |
| X-linked Hypohidrotic ectodermal dysplasia | EDA1 |
| X-linked lymphoproliferative syndrome | SH2D1A |
| X-linked mental retardation | GDI1 |
| X-linked mental retardation | MAGT1 |
| X-linked mental retardation (Stocco dos santos syndrome) | KIAA1202 |
| X-linked mental retardation 94 | GRIA3 |
| X-linked mental retardation, JARID1C-related | SMCX (JARID1C) |
| X-linked mental retardation, syndromic, Turner type | HUWE1 |
| X-linked MR (spasms, epileptic encephalopathy, atypical Rett) | ARX |
| X-Linked Severe Combined Immunodeficiency | IL2RG |
| XY sex reversal and gonadal dysgenesis | DMRT1 |
| XY sex reversal and gonadal dysgenesis | WNT4 |

