NewbornDx™ Advanced Sequencing Evaluation Disorders List

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Skeletal

Abnormal bone and cartilage

Disorder	Associated gene(s)
Alpha-mannosidosis	*MAN2B1*
Camurati-Engelmann disease	*TGFB1*
Cerebrocostomandibular syndrome	*SNRPB*
Chondrocalcinosis	*ANKH*
Chondrodysplasia punctata	*ARSE, EBP, PTH1R*
Chondrodysplasia with joint dislocations	*IMPAD1*
Craniometaphyseal dysplasia	*ANKH*
Eiken syndrome	*PTH1R*
Familial osteochondritis dissecans	*ACAN*
Fibrodysplasia ossificans progressiva	*ACVR1*
Hyperphosphatasia with mental retardation syndrome	*PGAP3, PIGV*
Kabuki syndrome	*KDM6A*
Keutel syndrome	*MGP*
Klippel-Feil syndrome	*GDF3, GDF6, MEOX1, MYO18B*
Lethal restrictive dermopathy	*ZMPSTE24*
Mandibuloacral dysplasia with type B lipodystrophy	*ZMPSTE24*
Marshall-Smith syndrome	*NFIX*
Metaphyseal chondrodysplasia	*PTH1R*
Multiple epiphyseal dysplasia with early-onset diabetes mellitus	*EIF2AK3*
Short-rib thoracic dysplasia	*DYNC2H1, IFT140, IFT80, NEK1, TTC21B, WDR19, WDR35*
Sotos syndrome	*NFIX, NSD1*

Bone fragility

Glycogen storage disease type IV	*GBE1*
Arthrogryposis-renal dysfunction-cholestasis syndrome	*VIPAS39, VPS33B*
Arthrogryposis, mental retardation, and seizures	*SLC35A3*
Bruck syndrome	*PLOD2*
Caffey disease	*COL1A1*
Cerebrooculofacioskeletal syndrome	*ERCC1, ERCC5*
Cockayne syndrome	*ERCC6, ERCC6, ERCC8*
Congenital contractural arachnodactyly	*FBN2*
Congenital contractures, hypotonia, and developmental delay	*NALCN*

Contractures

Distal arthrogryposis	*ECEL1, MYBPC1, MYH3, TNNI2, TNNT3, TPM2*
Ehlers-Danlos syndrome	*B4GALT7, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, FKBP14, PLOD1*
Histiocytosislymphadenopathy plus syndrome	*SLC29A3*
Infantile hypotonia, psychomotor retardation, characteristic facies	*NALCN*

Contractures (cont.)

Disorder	Associated gene(s)
Lethal arthrogryposis, with anterior horn cell disease	*GLE1*
Lethal congenital contracture syndrome	*ADCY6, CNTNAP1, DNM2, ERBB3, GLE1, MYBPC1, PIP5K1C*
Marden-Walker syndrome	*PIEZO2*
Multicentric osteolysis, nodulosis, and arthropathy	*MMP2*
Osteogenesis Imperfecta	*BMP1*
Osteogenesis imperfecta	*COL1A1, COL1A2, CRTAP, FKBP10, IFITM5, P3H1, PPIB, SERPINF1, SERPINH1, SP7, TMEM38B, WNT1*
Osteopathia striata with cranial sclerosis	*AMER1*
Osteopetrosis with renal tubular acidosis	*CA2*
Van den Ende-Gupta syndrome	*SCARF2*
Winchester syndrome	*MMP14*

Hand and foot abnormalities

Adams-Oliver syndrome	*ARHGAP31, DOCK6, EOGT, NOTCH1*
Atelosteogenesis	*SLC26A2*
Bardet-Biedl syndrome	*ARL6, BBIP1, IFT27, LZTFL1, MKKS, MKS1*
Brachydactyly	*BMP2, BMPR1B, HOXD13, PTHLH*
Catel-Manzke syndrome	*TGDS*
Ciliopathy	*CC2D2A, CEP290, IFT172, SDCCAG8*
COACH syndrome	*CC2D2A, TMEM67*
Coffin-Siris syndrome	*ARID1A, ARID1B, SMARCA4, SMARCB1, SMARCE1*
Congenital clubfoot with or without deficiency of long bones, mirror-image polydactyly	*PITX1*
Culler-Jones syndrome	*GLI2*
Diastrophic dysplasia	*SLC26A2*
Hay-Wells syndrome	*TP63*
Joubert syndrome	*CC2D2A, CEP290, CEP41, NPHP1, NPHP4*
Loeys-Dietz syndrome	*SMAD3, TGFB2, TGFB3, TGF-BR1, TGFBR2*
Meckel syndrome	*B9D1, CEP290, EXOC4, NPHP3, TMEM67*
Megalencephaly-capillary syndrome	*PIK3CA*
Multiple synostoses syndrome	*NOG*
Opsismodysplasia	*INPPL1*
Pallister-Hall syndrome	*GLI3*
Pitt-Hopkins-like syndrome	*CNTNAP2*
Postaxial polydactyly	*ZNF141*

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Hand and foot abnormalities (cont.)

Disorder	Associated gene(s)
Rubinstein-Taybi syndrome	EP300
Scalp-ear-nipple syndrome	*KCTD1*
Split-hand/foot malformation	*DLX5, FBXW4*
Split-hand/foot malformation with sensorineural hearing loss	*DLX5*
Syndactyly	*GJA1*
Synpolydactyly	*FBLN1, HOXD13*
Temtamy preaxial brachydactyly syndrome	*CHSY1*
Townes-Brocks syndrome	*SALL1*
Trichorhinophalangeal syndrome	*TRPS1*
Trismus-pseudocamptodactyly syndrome	*MYH8*
Weill-Marchesani syndrome	*ADAMTS10*

Overgrowth

Beckwith-Wiedemann syndrome	*CDKN1C*
Kosaki overgrowth syndrome	*PDGFRB*
Perlman syndrome	*DIS3L2*
Sotos syndrome	*NFIX, NSD1*

Short stature

17-alpha-hydroxylasedeficient congenital adrenal hyperplasia	*CYP17A1*
3-M syndrome	*CCDC8, CUL7, OBSL1*
Aarskog-Scott syndrome	*FGD1*
Achondrogenesis	*COL2A1, SLC26A2, TRIP11*
Achondroplasia	*FGFR3*
Acrocapitofemoral dysplasia	*IHH*
Acrodysostosis with or without hormone resistance	*PDE4D*
Acromesomelic chondrodysplasia	*GDF5*
Acromesomelic dysplasia BMPR1B	
Anauxetic dysplasia POP1	
Auriculocondylar syndrome	*GNAI3, PLCB4*
Brachydactyly	*BMP2, BMPR1B, HOXD13, PTHLH*
Cardiofaciocutaneous syndrome	*BRAF, KRAS, MAP2K1*
Carney complex	*MYH8, PRKAR1A*
Coffin-Lowry syndrome	*RPS6KA3*
Costello syndrome	*HRAS*
COUSIN syndrome	*TBX15*
Dyggve-Melchior-Clausen disease	*DYM*
Ellis-van Creveld syndrome	*EVC, EVC2*
Epiphyseal chondrodysplasia	*NPR2*
Fibrochondrogenesis	*COL11A1*
Floating-Harbor syndrome	*SRCAP*
Geleophysic dysplasia	*ADAMTSL2*
Intrahepatic cholestasis of pregnancy	*ABCB11*
KBG syndrome	*ANKRD11*
Kniest dysplasia	*COL2A1*

Short stature (cont.)

Disorder	Associated gene(s)
Marshall syndrome	*COL11A1*
Meier-Gorlin syndrome	*CDC6, CDT1, ORC1, ORC4, ORC6*
Microcephalic osteodysplastic primordial dwarfism	*PCNT*
Microcephaly, short stature, and polymicro-gyria with seizures	*RTTN*
Multiple joint dislocation syndrome	*B3GAT3*
Nicolaides-Baraitser syndrome	*SMARCA2*
Nijmegen breakage syndrome	*NBN*
Noonan syndrome	*A2ML1, BRAF, KRAS, LZTR1, NRAS, PTPN11, RAF1, RASA2, RIT1, RRAS, SOS1, SOS2*
Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia	*CBL*
Noonan-like syndrome with loose anagen hair	*SHOC2*
Osteoarthritis with mild chondrodysplasia	*COL2A1*
Phosphorylase kinase deficiency of liver and muscle	*PHKB*
Platyspondylic lethal skeletal dysplasia	*COL2A1*
Resistance to insulin-like growth factor I	*IGF1R*
Rhizomelic chondrodysplasia punctata	*AGPS, GNPAT*
Roberts syndrome	*ESCO2*
Rothmund-Thomson syndrome	*RECQL4*
SC phocomelia syndrome	*ESCO2*
Schimke immunoosseous dysplasia	*SMARCAL1*
Schwartz-Jampel syndrome	*HSPG2*
Short stature with nonspecific skeletal abnormalities	*NPR2*
Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis	*POC1A*
Smith-McCort dysplasia	*DYM, RAB33B*
Spondylo-megaepiphysealmetaphyseal dysplasia	*NKX3-2*
Spondylocheirodysplasia	*SLC39A13*
Spondylocostal dysostosis	*DLL3, HES7, MESP2, TBX6*
Spondyloenchondrodysplasia with immune dysregulation	*ACP5*
Spondyloepimetaphyseal dysplasia	*MMP13*
Spondyloepimetaphyseal dysplasia with joint laxity	*KIF22*
Spondyloepiphyseal dysplasia with congenital joint dislocations	*CHST3*
Spondylometaepiphyseal dysplasia	*DDR2*
Spondyloperipheral dysplasia	*COL2A1*
Stickler syndrome	*COL11A1, COL2A1, COL9A1*
Thanatophoric dysplasia	*FGFR3*
Weyers acrofacial dysostosis	*EVC, EVC2*
Wiedemann-Steiner syndrome	*KMT2A*

Skull abnormalities

3MC syndrome	*MASP1*
Apert syndrome	*FGFR2*
Beare-Stevenson syndrome	*FGFR2*

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Skull abnormalities (cont.)

Disorder	Associated gene(s)
Bent bone dysplasia syndrome	*FGFR2*
Carpenter syndrome	*MEGF8, RAB23*
Cleidocranial dysplasia	*RUNX2*
Cranioectodermal dysplasia	*IFT122, IFT43, WDR35*
Craniooosteoarthropathy	*HPGD*
Craniosynostosis	*ERF, TCF12*
Crouzon syndrome	*FGFR2*
Frank-ter Haar syndrome	*SH3PXD2B*
Hydrolethalus syndrome	*HYLS1*
Knobloch syndrome	*COL18A1*
Lenz-Majewski hyperostotic dwarfism	*PTDSS1*
Oculodentodigital dysplasia	*GJA1*
Pycnodysostosis	*CTSK*
Ritscher-Schinzel syndrome	*CCDC22, KIAA0196*
Saethre-Chotzen syndrome	*FGFR2, TWIST1*
Schinzel-Giedion midface retraction syndrome	*SETBP1*
Shprintzen-Goldberg syndrome	*SKI*

Tall stature

Marfan syndrome	*FBN1*
Weaver syndrome	*EZH2*

Upper and/or lower limb defects

Acro-renal-ocular syndrome	*SALL4*
Acrofacial dysostosis	*SF3B4*
Bainbridge-Ropers syndrome	*ASXL3*
Campomelic dysplasia	*SOX9*
Cartilage-hair hypoplasiaanauxetic dysplasia spectrum disorders	*RMRP*
Caudal regression syndrome	*VANGL1*
Chondrodysplasia punctata	*ARSE, EBP, PTH1R*
Desbuquois dysplasia	*CANT1, XYLT1*
Duane-radial ray syndrome	*SALL4*
Eiken syndrome	*PTH1R*
Endocrinecerebroosteodysplasia	*ICK*
Frontometaphyseal dysplasia	*FLNA*
Fuhrmann syndrome	*WNT7A*
Greenberg skeletal dysplasia	*LBR*
Holt-Oram syndrome	*SALL4, TBX5*
Hypophosphatasia	*ALPL*
Langer mesomelic dysplasia	*SHOX*
Larsen syndrome	*FLNB*
Marshall syndrome	*COL11A1*
Mesomelia-synostoses syndrome	*SULF1*
Metaphyseal anadysplasia	*MMP9*
Metaphyseal chondrodysplasia	*PTH1R*
Miller syndrome	*DHODH*
Multicentric carpotarsal osteolysis syndrome	*MAFB*
Multiple epiphyseal dysplasia	*SLC26A2*
Myhre syndrome	*SMAD4*
Nail-patella syndrome	*LMX1B*

Upper and/or lower limb defects (cont.)

Disorder	Associated gene(s)
Omodysplasia	*GPC6*
Otospondylomegaepiphyseal dysplasia	*COL11A2*
Paget disease of bone	*TNFRSF11B*
Peters plus syndrome	*B3GLCT*
Raine syndrome	*FAM20C*
Robinow syndrome	*ROR2, WNT5A*
Schneckenbecken dysplasia	*SLC35D1*
Skeletal disease	*LMBR1*
Small patella syndrome	*TBX4*
Spondylocarpotarsal synostosis syndrome	*FLNB*
Stuve-Wiedemann syndrome/ Schwartz-Jampel syndrome	*LIFR*
Temtamy syndrome	*C12orf57*
Ulnar-mammary syndrome	*TBX3*
Weissenbacher-Zweymuller syndrome	*COL11A2*

Respiratory

Abnormal breathing

Brown-Vialetto-Van Laere syndrome	*SLC52A2, SLC52A3*
Central hypoventilation syndrome	*ASCL1, EDN3, GDNF, PHOX2B, RET*
Hyperekplexia	*SLC6A5*
Neonatal diabetes with cerebellar agenesis	*PTF1A*
Pitt-Hopkins syndrome	*NRXN1, TCF4*
Pitt-Hopkins-like syndrome	*CNTNAP2*

Chronic respiratory infection/disease

Alpha-1-antitrypsin deficiency	*SERPINA1*
Cystic fibrosis	*CFTR*
Keutel syndrome	*MGP*

Developmental abnormalities

Hereditary hemorrhagic telangiectasia	*ACVRL1, ENG*
Primary pulmonary arterial hypertension	*BMPR2, CAV1, KCNK3, SMAD9*
Pulmonary venoocclusive disease	*EIF2AK4*

Pulmonary fibrosis

Hermansky-Pudlak syndrome	*AP3B1, BLOC1S6*
Dyskeratosis congenita	*DKC1, RTEL1, TERT, TINF2*

Respiratory distress

Alveolar capillary dysplasia with misalignment of pulmonary veins	*FOXF1*
Ciliary dyskinesia	*ARMC4, C21orf59, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, DRC1, DYX1C1, HYDIN, LRRC6, NME8, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, ZMYND10*
Hereditary C1 esterase inhibitor deficiency	*SERPING1*

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Respiratory distress (cont.)

Disorder	Associated gene(s)
Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa	*ITGA3*
Myopathy, lactic acidosis, and sideroblastic anemia	*YARS2, YARS2*
Pulmonary surfactant metabolism dysfunction	*ABCA3, SFTPB, SFTPC, SFTPD, ACADL*
Respiratory distress syndrome in premature infants	*SFTPD*

Miscellaneous

Asthma, drug response	*CRHR1*
Elevated extracellular superoxide dismutase	*SOD3*

Metabolic

Amino acid metabolism disorder

3-methylglutaconic aciduria	*CLPB, DNAJC19, OPA3*
Biotinidase deficiency	*BTD*
Branched-chain ketoacid dehydrogenase kinase deficiency	*BCKDK*
Combined D-2- and L-2- hydroxyglutaric aciduria	*SLC25A1*
Congenital glutamine deficiency	*GLUL*
D-2-hydroxyglutaric aciduria	*D2HGDH*
Dihydrolipoamide dehydrogenase deficiency	*DLD*
Ethylmalonic encephalopathy	*ETHE1*
Glycine N-methyltransferase deficiency	*GNMT*
Glutamate formiminotransferase deficiency	*FTCD*
Guanidinoacetate methyltransferase deficiency	*GAMT*
Hereditary folate malabsorption	*SLC46A1*
Histidinemia	*HAL*
Holocarboxylase synthetase deficiency	*HLCS*
Homocystinuria	*CBS, MTHFR*
Homocystinuria-megaloblastic anemia	*MTR, MTRR*
Hypermethioninemia with Sadenosylhomo-cysteine hydrolase deficiency	*AHCY*
Hyperlysinemia	*AASS*
Isobutyryl-CoA dehydrogenase deficiency	*ACAD8*
Isolated persistent hypermethioninemia	*MAT1A*
Isovaleric acidemia	*IVD*
L-arginine:glyci amidinotransferase deficiency	*GATM*
Lysinuric protein intolerance	*SLC7A7*
Maple syrup urine disease	*BCKDHA, BCKDHB, DBT, DLD*
Methylmalonic aciduria and homocystinuria	*ABCD4, LMBRD1, MMACHC, MMADHC*
Methylmalonic acidemia and homocysteinemia, cblX type	*HCFC1*
Mevalonic aciduria	*MVK*
Non-ketotic hyperglycinemia	*AMT, GCSH, GLDC*
Phenylketonuria	*PAH*
Phosphoglycerate dehydrogenase deficiency	*PHGDH*
Phosphoserine phosphatase deficiency	*PSPH*
Riboflavin deficiency	*SLC52A1*
Tyrosinemia, type I	*FAH*

Carbohydrate metabolism disorder

Disorder	Associated gene(s)
Epimerase-deficienct galactosemia	*GALE*
Fructose 1,6 bisphosphatase deficiency	*FBP1*
Galactokinase deficiency	*GALK1*
Galactosemia	*GALT*
Glycogen storage disease	*AGL, ALDOA, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LDHA, PFKM, PHKA2, PHKG2, PRK-AG2, PYGL, SLC37A4*
Hereditary fructose intolerance	*ALDOB*
Lipoyltransferase deficiency	*LIPT1*
McArdle disease	*PYGM*
Phosphoglycerate kinase deficiency	*PGK1*

Cholesterol disorder

Desmosterolosis	*DHCR24*
Lathosterolosis	*SC5D*

Fatty acid oxidation disorder

Carnitine palmitoyltransferase deficiency, type I	*CPT1A, CPT1B*
Carnitine-acylcarnitine translocase deficiency	*SLC25A20*
Glutaric acidemia, type II	*ETFA, ETFB, ETFDH*
Medium chain acyl-coenzyme A dehydrogenase deficiency	*ACADM*
Short chain acyl-CoA dehydrogenase deficiency	*ACADS*
Succinyl-CoA:3-oxoacid CoA transferase deficiency	*OXCT1*
Systemic primary carnitine deficiency	*SLC22A5*
Very long chain acylcoenzyme A dehydrogenase deficiency	*ACADVL*

Lipid storage

Cerebrotendinous xanthomatosis	*CYP27A1*

Lysosomal storage disease

Aspartylglucosaminuria	*AGA*
Fabry Disease	*GLA*
Frontotemporal lobar degeneration with ubiquitinpositive inclusions	*GRN*
Fucosidosis	*FUCA1*
Galactosialidosis	*CTSA*
Gaucher disease	*GBA*
Hexosaminidase A deficiency	*HEXA*
I2-mannosidosis	*MANBA*
Inclusion body myopathy and Nonaka myopathy	*GNE*
Lysosomal acid lipase deficiency	*LIPA*
Metachromatic leukodystrophy	*ARSA, PSAP*
Mucolipidosis, type IV	*MCOLN1*
Mucolipidosis, type II	*GNPTAB*
Mucolipidosis, type III	*GNPTG*
Mucopolysaccharidosis type I	*IDUA*

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Lysosomal storage disease (cont.)

Disorder	Associated gene(s)
Mucopolysaccharidosis type II	*IDS*
Mucopolysaccharidosis type III	*GNS*
Mucopolysaccharidosis type III	*HGSNAT, NAGLU, SGSH*
Mucopolysaccharidosis type IV	*GLB1*
Mucopolysaccharidosis type VII	*GUSB*
Multiple sulfatase deficiency	*SUMF1*
Neuronal ceroid lipofuscinosis	*CLN3, CLN5, CLN6, CLN8, CTSD, MFSD8, PPT1, TPP1*
Niemann-Pick disease	*NPC1, NPC2, SMPD1*
Primary progressive aphasia	*GRN*
Sandhoff disease	*HEXB*
Schindler disease	*NAGA*
Sialidosis	*NEU1*
Sialuria	*GNE*
Tay-Sachs disease	*HEXA*
Wolman disease	*LIPA*

Mitochondrial

2,4-dienoyl-CoA reductase deficiency	*DECR1, NADK2*
3-hydroxy-3-methylglutaryl-CoA synthase deficiency	*HMGCS2*
3-Methylglutaconic acidur with deafness, encephalopathy, and Leigh-like Syndrome	*SERAC1*
3-methylcrotonyl-CoA carboxylase deficiency	*MCCC1, MCCC2*
Acyl-CoA dehydrogenase deficiency	*ACAD9*
Alpers-Huttenlocher syndrome	*POLG*
Alpha-ketoglutarate dehydrogenase deficiency	*OGDH*
Coenzyme Q10 deficiency	*ADCK3, COQ2, COQ6, COQ9, PDSS1, PDSS2*
Combined mitochondrial complex deficiency	*GFER*
Combined oxidative phosphorylation deficiency	*AARS2, AIFM1, ATP5A1, C12orf65, EARS2, ELAC2, FARS2, GFM1, GTPBP3, LYRM4, MARS2, MRPL3, MRPL44, MRPS16, MRPS22, MTFMT, MTO1, NARS2, PNPT1, RMND1, SFXN4, SURF1, TARS2, TSFM, TUFM, VARS2*
Cytosolic phosphoenolpyruvate carboxykinase-1 deficiency	*PCK1*
Encephalopathy due to defective mitochondrial and peroxisomal fission	*DNM1L*
Fatal infantile cardioencephalomyopathy	*COA5*
Hereditary myopathy with lactic acidosis	*ISCU, PNPLA8*
Infantile striatonigral degeneration	*NUP62*
Leigh syndrome	*BCS1L, COX4I1, LRPPRC, NDUFA10, NDUFA12, NDUFA2, NDUFA4, NDUFA9, NDUFAF6, NDUFS7, SURF1*
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	*HADHA*

Mitochondrial (cont.)

Disorder	Associated gene(s)
Mitochondrial complex I deficiency	*FOXRED1, NDUFA1, NDUFA11, NDUFA13, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF7, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS8, NDUFV1, NDUFV2, NUBPL*
Mitochondrial complex II deficiency	*SDHAF1, SDHD*
Mitochondrial complex III deficiency	*BCS1L, CYC1, TTC19, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRQ*
Mitochondrial complex IV deficiency	*APOPT1, COX10, COX14, COX15, COX20, COX6B1, FASTKD2, PET100, SCO1, TACO1*
Mitochondrial complex V deficiency	*ATP5E, ATPAF2, TMEM70*
Mitochondrial disease	*MFF*
Mitochondrial DNA depletion syndrome	*C10orf2, DGUOK, FBXL4, MGME1, MPV17, RRM2B, SLC25A4, SUCLA2, SUCLG1, TK2*
Mitochondrial myopathy and sideroblastic anemia	*PUS1*
Mitochondrial phosphate carrier deficiency	*SLC25A3*
Mitochondrial pyruvate carrier deficiency	*MPC1*
Mitochondrial recessive ataxia syndrome	*POLG*
Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency	*ECHS1*
Multiple mitochondrial dysfunctions syndrome	*IBA57, NFU1, BOLA3*
PEPCK deficiency	*PCK2*
Progressive external ophthalmoplegia	*DNA2, POLG2,SLC25A4*
Progressive leukoencephalopathy	*AARS2*
Pyruvate carboxylase deficiency	*PC*
Pyruvate dehydrogenase deficiency	*DLAT*
Pyruvate dehydrogenase E1-subunit deficiency	*PDHA1, PDHB*
Pyruvate dehydrogenase E3-binding protein deficiency	*PDHX*
Pyruvate dehydrogenase lipoic acid synthetase deficiency	*LIAS*

Neurotransmitter disease

Pyruvate dehydrogenase phosphatase deficiency	*PDP1*
Trifunctional protein deficiency	*HADHA, HADHB*
6-pyruvoyltetrahydropterin synthase deficiency	*PTS*
Aromatic L-amino acid decarboxylase deficiency	*DDC*
BH4-deficient hyperphenylalaninemia	*GCH1, PCBD1, QDPR*
Dystonia	*SLC2A1*
GABA-transaminase deficiency	*ABAT*
Glucose transporter deficiency	*SLC2A1*
GTP cyclohydrolase 1- deficient dopa-responsive dystonia	*GCH1*
Pyridoxamine 5-prime-phosphate oxidase deficiency	*PNPO*

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Organic aciduria

Disorder	Associated gene(s)
17-beta-hydroxysteroid dehydrogenase X deficiency	*HSD17B10*
2-methylbutyrylglycinuria	*ACADSB*
3-hydroxyisobutryl-CoA hydrolase deficiency	*HIBCH, HMGCL*
3-methylglutaconyl-CoA hydratase deficiency	*AUH*
Barth syndrome	*TAZ*
Glutaric acidemia, type I	*GCDH*
Glutaric acidemia, type III	*SUGCT*
Malonyl-CoA decarboxylase deficiency	*MLYCD*
Methylmalonic aciduria	*CD320, MMAA, MMAB, MUT*
Propionic acidemia	*PCCA, PCCB*

Peroxisomal disorder

Infantile refsum disease	*PEX12, PEX5*
Neonatal adrenoleukodystrophy	*PEX1, PEX10, PEX12, PEX13, PEX5, PEX6*
Peroxisome biogenesis disorder	*PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7*
Peroxisomal fatty acyl-CoA reductase disorder	*FAR1*
Refsum disease	*PHYH*
Rhizomelic chondrodysplasia	*PEX7*
Zellweger syndrome	*PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX3, PEX5, PEX6*

Urea cycle disorder

Argininemia	*ARG1*
Argininosuccinic aciduria	*ASL*
Carbamoylphosphate synthetase I deficiency	*CPS1*
Citrin deficiency	*SLC25A13*
Citrullinemia, type I	*ASS1*
Citrullinemia, type II	*SLC25A13*
N-acetylglutamate syntha deficiency	*NAGS*
Ornithine aminotransferase deficiency	*OAT*
Ornithine transcarbamylase deficiency	*OTC*

Miscellaneous

5-oxoprolinase deficiency	*OPLAH*
Combined malonic and methylmalonic aciduria	*ACSF3*
Congenital disorder of glycosylation	*ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, B4GALT1, COG1, COG4, COG5, COG6, COG7, COG8, DDOST, DOLK, DPAGT1, DPM1, DPM2, DPM3, MGAT2, MOGS, MPDU1, MPI, NGLY1, NHEJ1, PGM1, PGM3, PMM2, RFT1, RPN2, SLC35A1, SLC35A2, SLC35C1, SRD5A3, SSR4, STT3A, STT3B, TMEM165*
D-glyceria aciduria	*GLYCTK*
Fumarate hydrotase deficiency	*FH*
Glutathione synthetase deficiency	*GSS*
Homocystinuria	*CBS, MTHFR*

Miscellaneous (cont.)

Disorder	Associated gene(s)
Hyperornithinemia- hyperammonemia-homocitrullinemia syndrome	*SLC25A15*
Hyperprolinemia	*ALDH4A1, PRODH*
Lysyl hydroxylase deficiency	*PLOD3*
Methylmalonyl-CoA epimerase deficiency	*MCEE*
Molybdenum cofactor deficiency	*GPHN, MOCS1, MOCS2*
Orotic aciduria	*UMPS*
Sialic acid storage disorder	*SLC17A5*

Muscle

Dystonia/abnormal movements

Familial dyskinesia	*ADCY5*
Infantile parkinsonism-dystonia	*SLC6A3*
Paramyotonia congenita	*SCN4A*

Hypotonia

Glycogen storage disease type IV	*GBE1*
Allan-Herndon-Dudley syndrome	*SLC16A2*
Arts syndrome	*PRPS1*
Beta-ureidopropionase deficiency	*UPB1*
Bethlem myopathy	*COL12A1*
Centronuclear myopathy	*BIN1, CCDC78, SPEG*
Charcot-Marie-Tooth disease	*DNM2, EGR2, FGD4, GARS, GDAP1, IGHMBP2, MPZ, NAGLU, PMP22, PRPS1, PRX, SURF1*
Combined mitochondrial complex deficiency	*GFER*
Ehlers-Danlos syndrome	*B4GALT7, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, FKBP14, PLOD1*
Griscelli syndrome	*MYO5A, RAB27A*
Klippel-Feil syndrome	*GDF3, GDF6, MEOX1, MYO18B*
Koolen-De Vries syndrome	*KANSL1*
L-2-hydroxyglutaric aciduria	*L2HGDH*
Lethal congenital contracture syndrome	*ADCY6, CNTNAP1, DNM2, ERBB3, GLE1, MYBPC1, PIP5K1C*
Mitochondrial disease	*MFF*
Multiple congenital anomalies-hypotonia-seizure syndrome	*PIGA, PIGN, PIGT*
Myoadenylate deaminase deficiency	*AMPD1*
Non-syndromic hearing loss	*ACTG1, MYH9, PNPT1, PRPS1, SIX1, SIX2, TBC1D24, TJP2*
Opitz-Kaveggia syndrome	*MED12*
Phosphoribosylpyrophosphate synthetase superactivity	*PRPS1*
Phosphorylase kinase deficiency of liver and muscle	*PHKB*
Prader-Willi syndrome	*SNRPN*
Prader-Willi-like syndrome	*MAGEL2*

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Hypotonia (cont.)

Disorder	Associated gene(s)
Sialic acid storage disorder	*SLC17A5*
Smith-Magenis syndrome	*RAI1, SMS*
Spastic paraplegia	*AP4B1, AP4E1, AP4M1, AP4S1, C12orf65, ERLIN2, HSPD1, IBA57, KIF1A, KIF5A, PLP1, TECPR2*
Spinal muscular atrophy	*ASAH1, BICD2, DYNC1H1, IGHMBP2, TRPV4, UBA1*

Muscle atrophy/dystrophy

Glycogen storage disease type IV	*GBE1*
Becker muscular dystrophy	*DMD*
Beta-ketothiolase deficiency	*ACAT1*
Congenital muscular dystrophy	*CHKB, ITGA7, LAMA2, LMNA, POMT1, POMT2*
Duchenne muscular dystrophy	*DMD*
Emery-Dreifuss muscular dystrophy	*SYNE1, SYNE2, TMEM43*
Fukuyama congenital muscular dystrophy	*FKRP*
Generalized congenital lipodystrophy	*PTRF*
Limb-girdle muscular dystrophy	*DAG1, DYSF, PLEC, TCAP, TRIM32*
Miyoshi muscular dystrophy	*DYSF*
Muscular dystrophy-dystroglycanopathy	*DAG1, FKRP, FKTN, GMPPB, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, TMEM5, B3GALNT2, B4GAT1*
Spinal muscular atrophy	*ASAH1, BICD2, DYNC1H1, IGHMBP2, TRPV4, UBA1*
Spinocerebellar ataxia	*ATP2B3, GRM1, ITPR1, SPTBN2*
Ullrich congenital muscular dystrophy	*COL6A1, COL6A2, COL6A3*

Muscle weakness

Brown-Vialetto-Van Laere syndrome	*SLC52A2, SLC52A3*
Congenital myasthenic syndrome	*AGRN, ALG14, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, COLQ, DOK7, GFPT1, LRP4, MUSK, RAPSN, SNAP25*
Fetal akinesia deformation sequence	*RAPSN*
Hyperkalemic periodic paralysis	*SCN4A*
Marinesco-Sjögren syndrome	*SIL1*
Schwartz-Jampel syndrome	*HSPG2*
Wieacker-Wolff syndrome	*ZC4H2*

Myopathy

Acute recurrent myoglobinuria	*LPIN1*
CAP myopathy	*TPM2, TPM3*
Central core disease	*RYR1*
Congenital myopathy	*ACTA1, CNTN1, HACD1*
Congenital myopathy with fiber type disproportion	*TPM3*
Congenital neuromuscular disease with uniform type 1 fiber	*RYR1*
Distal arthrogryposis	*ECEL1, MYBPC1, MYH3, TNNI2, TNNT3, TPM2*

Myopathy (ccont.)

Disorder	Associated gene(s)
Hereditary myopathy with lactic acidosis	*ISCU, PNPLA8*
King-Denborough syndrome	*RYR1*
McArdle disease	*PYGM*
Minicore myopathy	*RYR1*
Myofibrillar myopathy	*CRYAB*
Myopathy, areflexia, respiratory distress, and dysphagia	*MEGF10*
Myopathy, lactic acidosis, and sideroblastic anemia	*YARS2*
Myopathy, proximal, with early respiratory muscle involvement	*TTN*
Myopathy, X-linked, with excessive autophagy	*VMA21*
Myotonia congenita	*CLCN1*
Myotubular myopathy	*MTM1*
Native American myopathy	*STAC3*
Nemaline myopathy	*ACTA1, KLHL40, KLHL41, LMOD3, NEB, TNNT1, TPM2, TPM3*
Polyglucosan body myopathy with or without immunodeficiency	*RBCK1*
Proximal myopathy and ophthalmoplegia	*MYH2*
Visceral myopathy	*ACTG2*
Myosin storage myopathy	*MYH7*
Liang distal myopathy	*MYH7*

Integumentary

Abnormal growths/lesions

Basal cell nevus syndrome	*PTCH1, PTCH2*
Buschke-Ollendorff syndrome	*LEMD3*
Coproporphyria	*CPOX*
Erythrokeratodermia variabilis with erythema gyratum repens	*GJB4*
Harderoporphyria	*CPOX*
Infantile capillary hemangioma	*ANTXR1*
MEDNIK syndrome	*AP1S1*
Prolidase deficiency	*PEPD*
Tuberous sclerosis	*TSC1, TSC2*

Abnormal hair and hair growth

Acrodermatitis enteropathica	*SLC39A4*
Cantu syndrome	*ABCC9, KCNJ8*
Ectodermal dysplasia	*EDA, EDAR, EDARADD, HOXC13, NFKBIA*
Hypotrichosis with juvenile macular dystrophy	*CDH3*
Hypotrichosis-lymphedema-telangiectasia syndrome	*SOX18*
Monilethrix	*KRT81, KRT83, KRT86*
Schopf-Schulz-Passarge syndrome	*WNT10A*
Trichodentoosseous syndrome	*DLX3*
Trichorhinophalangeal syndrome	*TRPS1*

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Abnormal hair and hair growth (cont.)

Disorder	Associated gene(s)
Trichothiodystrophy	*GTF2H5*
Wiedemann-Steiner syndrome	*KMT2A*

Abnormal palms and feet

Arrhythmogenic right ventricular dysplasia, mild palmoplantar keratoderma and woolly hair	*DSC2*
Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome	*SNAP29*
Congenital erythroderma with palmoplantar keratoderma, hypotrichosis, and hyper IgE	*DSG1*
Craniooosteoarthropathy	*HPGD*
Meleda disease	*SLURP1*
Naxos disease	*JUP*
Odontoonychodermal dysplasia	*WNT10A*
Pachyonychia congenita	*KRT16*
Palmoplantar keratoderma	*KRT16, KRT9*

Abnormal pigmentation

Carney complex	*MYH8, PRKAR1A*
Carpenter syndrome	*MEGF8, RAB23*
Chediak-Higashi syndrome	*LYST*
Dowling-Degos disease	*KRT5*
Dyskeratosis congenita	*DKC1, RTEL1, TERT, TINF2*
Griscelli syndrome	*MYO5A, RAB27A*
Hermansky-Pudlak syndrome	*AP3B1, BLOC1S6*
Legius syndrome	*SPRED1*
Neurofibromatosis, type 1	*NF1*
Oculocutaneous albinism	*TYR*
Piebaldism	*SNAI2*
Tietz albinism-deafness syndrome	*MITF*
Waardenburg syndrome	*EDN3, EDNRB, MITF, SNAI2, SOX10*

Abnormal skin appearance

Congenital symmetric circumferential skin creases	*TUBB*
Focal facial dermal dysplasia	*CYP26C1*
Hennekam lymphangiectasia-lymphedema syndrome	*FAT4*
Hereditary hemorrhagic telangiectasia	*ACVRL1, ENG*
Hereditary lymphedema	*FLT4, VEGFC*
Lethal restrictive dermopathy	*ZMPSTE24*
Linear skin defects with multiple congenital anomalies	*HCCS*
Mandibuloacral dysplasia with type B lipodystrophy	*ZMPSTE24*
Poikiloderma with neutropenia	*USB1*
Popliteal pterygium syndrome	*IRF6, RIPK4*
Psoriasis	*IL20RA*
Pyogenic sterile arthritis, pyoderma gangrenosum, and acne	*PSTPIP1*

Absence of skin

Disorder	Associated gene(s)
Adams-Oliver syndrome	*ARHGAP31, DOCK6, EOGT, NOTCH1*
Focal dermal hypoplasia	*PORCN*
Netherton syndrome	*SPINK5*
Rothmund-Thomson syndrome	*RECQL4*
Scalp-ear-nipple syndrome	*KCTD1*

Blistering

Congenital erythropoietic porphyria	*UROS*
Epidermolysis bullosa	*CD151, COL17A1, COL7A1, DSP, DST, EXPH5, ITGA6, ITGB4, KRT14, KRT5, LAMA3, LAMB3, LAMC2, PLEC*
Incontinentia pigmenti	*IKBKG*
Kindler syndrome	*FERMT1*
Laryngoonychocutaneous syndrome	*LAMA3*
Nephropathy with pretibial epidermolysis bullosa and deafness	*CD151*

Dry skin

Chanarin-Dorfman syndrome	*ABHD5*
CHIME syndrome	*PIGL*
Congenital ichthyosis	*ABCA12, ALOX12B, ALOXE3, CERS3, CYP4F22, FLG, KRT1, KRT10, KRT2, LIPN, NIPAL4, PNPLA1, SLC27A4, ST14, STS, TGM1*
Craniofaciocutaneous syndrome	*MAP2K2*
Ectodermal dysplasia/skin fragility syndrome	*PKP1*
Ichthyosis	*ELOVL4*
Keratitis-ichthyosis-deafness syndrome	*GJB2*
Keratosis linearis with ichthyosis congenita and sclerosing keratoderma	*POMP*
Peeling skin syndrome	*CHST8, TGM5*
Sjögren-Larsson syndrome	*ALDH3A2*

Loose skin

Cutis laxa	*ALDH18A1, ATP6V0A2, EFEMP2, FBLN5, LTBP4, PYCR1*
Ehlers-Danlos syndrome	*B4GALT7, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, FKBP14, PLOD1*
Geroderma osteodysplasticum	*GORAB*
Williams syndrome	*ELN*

Skin sensitivity to UV light

Bloom syndrome	*BLM*
Porphyria	*ALAD, ALAS2, UROD*
UV-sensitive syndrome	*UVSSA*
Xeroderma pigmentosum	*ERCC2, ERCC3, ERCC4, ERCC5, XPA, XPC*

Venous abnormalities

Capillary-arteriovenous malformation	*RASA1*
Multiple cutaneous and mucosal venous malformations	*TEK*
Van Maldergem syndrome	*FAT4*

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Miscellaneous

Disorder	Associated gene(s)
Congential insensitivity to pain with anhidrosis	*NTRK1*

Immune

Abnormal immune system cells

Histiocytosis- lymphadenopathy plus syndrome	*SLC29A3*
Hyper-IgE syndrome	*DOCK8, STAT3*
Hypereosinophilic syndrome	*PDGFRA*

Immunodeficiency

Agammaglobulinemia	*BLNK, BTK, CD79A, CD79B, IGHM, IGLL1, LRRC8A*
Ataxia-telangiectasia	*ATM*
Common variable immunodeficiency	*CD19, CD81, ICOS, IL21, MS4A1, NFKB1, TNFRSF13C*
Dyskeratosis congenita	*DKC1, RTEL1, TERT, TINF2*
Gastrointestinal defects and immunodeficiency syndrome	*TTC7A*
Griscelli syndrome	*MYO5A, RAB27A*
Immunodeficiency	*CARD11, CD247, IKBKB, IL21R, IRF8, LAMTOR2, LCK, LRBA, PIK3CD, PIK3R1, PNP, STAT1, TRAC*
Immunodeficiency-centromeric instability-facial anomalies syndrome	*DNMT3B, ZBTB24*
Infantile-onset multisystem autoimmune disease	*STAT3, ZAP70*
Lymphoproliferative syndrome	*CD27, XIAP*
Neutrophil immunodeficiency syndrome	*RAC2*
Polyglucosan body myopathy with or without immunodeficiency	*RBCK1*
Reticular dysgenesis	*AK2*
Schimke immunoosseous dysplasia	*SMARCAL1*
Severe combined immunodeficiency	*ADA, CD3D, CD3E, DCLRE1C, IL2RG, IL7R, JAK3, LIG4, PTPRC, RAG1, RAG2, ZAP70*

Neutropenia

Autoimmune lymphoproliferative syndrome	*CASP10, CTLA4, FAS, FASLG, PRKCD*
Carpenter syndrome	*MEGF8, RAB23*
Cohen syndrome	*VPS13B*
Congenital severe neutropenia	*G6PC3*
Hereditary neutrophilia	*CSF3R*
Severe congenital neutropenia	*ELANE, GFI1, HAX1, JAGN1, VPS45, WAS*

Recurrent fevers/recurrent infections

Autoinflammation, antibody deficiency, and immune dysregulation syndrome	*PLCG2*
B-cell expansion with NFKB and T-cell anergy	*CARD11*
Chediak-Higashi syndrome	*LYST*
Congenital erythroderma with palmoplantar keratoderma, hypotrichosis, and hyper IgE	*DSG1*

Recurrent fevers/recurrent infections (cont.)

Disorder	Associated gene(s)
Congenital isolated asplenia	*RPSA*
Familial hemophagocytic lymphohistiocytosis	*PRF1, STXBP2, UNC13D*
Familial Mediterranean fever	*MEFV*
Interleukin 2 receptor alpha chain deficiency	*IL2RA*
IRAK4 deficiency	*IRAK4*
Muckle-Wells syndrome	*NLRP3*
Natural killer cell and glucocorticoid deficiency with DNA repair defect	*MCM4*
Recurrent isolated invasive pneumococcal disease	*IRAK4*
T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations	*STK4*
WHIM syndrome	*CXCR4*
Wiskott-Aldrich syndrome	*WAS*

Hematologic

Anemia

Acute intermittent porphyria	*HMBS*
Anemia	*GATA1*
Beta-thalassemia	*HBB*
Congenital dyserythropoietic anemia	*C15orf41, CDAN1, SEC23B*
Dehydrated hereditary stomatocytosis	*PIEZO1*
Diamond-Blackfan anemia	*RPL11, RPL15, RPL26, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS28, RPS29, RPS7, TSR2*
Distal renal tubular acidosis	*SLC4A1*
Elliptocytosis	*SPTA1*
Erythropoietic protoporphyria	*FECH*
Familial thrombotic thrombocytopenia purpura	*ADAMTS13*
Fanconi anemia	*BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, RAD51C, SLX4, UBE2T, XRCC2*
Ghosal hematodiaphyseal syndrome	*TBXAS1*
Glucose-6-phosphate dehydrogenase deficiency	*G6PD*
Glutathione synthetase deficiency	*GSS*
Glycogen storage disease	*AGL, ALDOA, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LDHA, PFKM, PHKA2, PHKG2, PRKAG2, PYGL, SLC37A4*
Hemolytic anemia	*CD59, TPI1*
Hereditary lymphadema	*PIEZO1*
Hereditary persistence of fetal hemoglobin	*KLF1*
Homocystinuria-megaloblastic anemia	*MTR, MTRR*
Majeed syndrome	*LPIN2*
Megaloblastic anemia	*CUBN, DHFR*
Orotic aciduria	*UMPS*
Porphyria	*ALAD, ALAS2, UROD*
Pyruvate kinase deficiency	*PKLR*
Sickle cell disease, hemoglobin trait	*HBB*

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Anemia (cont.)

Disorder	Associated gene(s)
Sideroblastic anemia	*ABCB7, ALAS2, SLC25A38, TRNT1*
Spherocytosis	*ANK1, EPB42, SPTB*
Thiamine-responsive megaloblastic anemia syndrome	*SLC19A2*
Transcobalamin II deficiency	*TCN2*
Xeroderma pigmentosum	*ERCC2, ERCC3, ERCC4, ERCC5, XPA, XPC*

Heme/hemoglobinopathy

Acute intermittent porphyria	*HMBS*
Alpha-thalassemia intellectual disability syndrome	*ATRX*
Beta-thalassemia	*HBB*
Familial thrombotic thrombocytopenia purpura	*ADAMTS13*
Porphyria	*ALAD, ALAS2, UROD*
Sickle cell disease, hemoglobin trait	*HBB*
Sideroblastic anemia	*ABCB7, ALAS2, SLC25A38, TRNT1*

Hemophilia (bleeding disorder)

Bernard-Soulier syndrome	*GP1BA, GP1BB, GP9*
Combined factor V and VIII deficiency	*LMAN1, MCFD2*
Congenital afibrinogenemia	*FGA, FGB, FGG*
Epstein syndrome	*MYH9*
Factor VII deficiency	*F7*
Factor X deficiency	*F10*
Factor XI deficiency	*F11*
Factor XIII subunit deficiency	*F13A1, F13B*
Familial visceral amyloidosis	*FGA*
Fechtner syndrome	*MYH9*
Hemophilia	*F8, F9*
Macrothrombocytopenia and progressive sensorineural deafness	*MYH9*
May-Hegglin anomaly	*MYH9*
Non-syndromic hearing loss	*ACTG1, MYH9, PNPT1, PRPS1, SIX1, SIX2, TBC1D24, TJP2*
Platelet-type bleeding disorder	*GP6, ITGA2B, ITGB3, RASGRP2*
Pseudo-von Willebrand disease, platelet-type	*GP1BA*
Sebastian syndrome	*MYH9*
von Willebrand disease	*VWF*

Hyperbilirubinemia

Alpha-thalassemia intellectual disability syndrome	*ATRX*
Beta-thalassemia	*HBB*
Dubin-Johnson syndrome	*ABCC2*
Familial thrombotic thrombocytopenia purpura	*ADAMTS13*
Intrahepatic cholestasis of pregnancy	*ABCB11*
Pyruvate kinase deficiency	*PKLR*
Sickle cell disease, hemoglobin trait	*HBB*

Immune

Disorder	Associated gene(s)
Agammaglobulinemia	*BLNK, BTK, CD79A, CD79B, IGHM, IGLL1, LRRC8A*
Agammaglobulinemia and isolated hormone deficiency	*BTK*
Congenital severe neutropenia	*G6PC3*
Dehydrated hereditary stomatocytosis	*PIEZO1*
Hereditary lymphadema	*PIEZO1*
Leukocyte adhesion deficiency	*FERMT3*
Lymphoproliferative syndrome	*CD27, XIAP*
Methemoglobinemia	*CYB5R3*
Severe congenital neutropenia	*ELANE, GFI1, HAX1, JAGN1, VPS45, WAS*
Shwachman-Diamond Syndrome	*SBDS*

Platelet disorder

Bernard-Soulier syndrome	*GP1BA, GP1BB, GP9*
Chediak-Higashi syndrome	*LYST*
Dyskeratosis congenita	*DKC1, RTEL1, TERT, TINF2*
Epstein syndrome	*MYH9*
Familial platelet disorder	*RUNX1*
Fechtner syndrome	*MYH9*
Glanzmann thrombasthenia	*ITGA2B, ITGB3*
Gray platelet syndrome	*NBEAL2*
Isolated giant platelet disorder	*GP1BB*
Leukemia, acute	*DICER1, PBX1, RUNX1*
Macrothrombocytopenia and progressive sensorineural deafness	*MYH9*
May-Hegglin anomaly	*MYH9*
Non-syndromic hearing loss	*ACTG1, MYH9, PNPT1, PRPS1, SIX1, SIX2, TBC1D24, TJP2*
Platelet-type bleeding disorder	*GP6, ITGA2B, ITGB3, RASGRP2*
Pseudo-von Willebrand disease, platelet-type	*GP1BA*
Scott syndrome	*ANO6*
Sebastian syndrome	*MYH9*
Thrombocythemia	*MPL*
Thrombocytopenia	*GATA1, MASTL, WAS*
Thrombocytopenia absent radius syndrome	*RBM8A*
Transcobalamin II deficiency	*TCN2*

Thrombophilia (clotting disorder)

Dysprothrombinemia	*F2*
Factor V Leiden thrombophilia	*F5*
Hereditary antithrombin deficiency	*SERPINC1*
Hypoprothrombinemia	*F2*
Thrombophilia	*F2, PROC, PROS1*

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Genitourinary

Genital abnormalities

Disorder	Associated gene(s)
17-alpha-hydroxylase-deficient congenital adrenal hyperplasia	*CYP17A1*
46XY sex reversal	*NR5A1*
Aarskog-Scott syndrome	*FGD1*
Bardet-Biedl syndrome	*ARL6, BBIP1, IFT27, LZTFL1, MKKS, MKS1*
Caudal regression syndrome	*VANGL1*
Congenital adrenal hyperplasia	*CYP11B1*
Congenital severe neutropenia	*G6PC3*
FG syndrome	*CASK, FLNA*
Fraser syndrome	*FRAS1, FREM2, GRIP1*
Frontometaphyseal dysplasia	*FLNA*
Hypergonadotropic hypogonadism	*LHCGR*
Hypogonadotropic hypogonadism	*FGF8, KISS1, KISS1R, NSMF, PROKR2, TAC3, TACR3*
McKusick-Kaufman syndrome	*MKKS*
Opitz GBBB syndrome	*MID1, SPECC1L*
Orofacial cleft	*BMP4*
Perrault syndrome	*CLPP, HARS2, HSD17B4, LARS2*
Robinow syndrome	*ROR2, WNT5A*
Townes-Brocks syndrome	*SALL1*
Wilms tumor-aniridia-genital anomalies-retardation syndrome	*WT1*

Liver disease

Alagille syndrome	*JAG1, NOTCH2*
Arthrogryposis-renal dysfunction-cholestasis syndrome	*VIPAS39, VPS33B*
Ciliopathy	*CC2D2A, CEP290, IFT172, SDCCAG8*
COACH syndrome	*CC2D2A, TMEM67*
Congenital bile acid synthesis defect	*ABCD3, AKR1D1, AMACR, CYP7B1, HSD3B7, SLC10A2*
Glycogen storage disease	*AGL, ALDOA, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LDHA, PFKM, PHKA2, PHKG2, PRKAG2, PYGL, SLC37A4*
Hajdu-Cheney syndrome	*NOTCH2*
Joubert syndrome	*CC2D2A, CEP290, CEP41, NPHP1, NPHP4*
Meckel syndrome	*B9D1, CEP290, EXOC4, NPHP3, TMEM67*
Nephronophthisis	*DCDC2, INVS, NPHP1, NPHP3, NPHP4, WDR19*
Osteopetrosis with renal tubular acidosis	*CA2*
Renal-hepatic-pancreatic dysplasia	*NPHP3*
Senior-Loken syndrome	*CEP290, IQCB1, NPHP1, NPHP4, SDCCAG8, WDR19*

Renal disease

Disorder	Associated gene(s)
Acute recurrent myoglobinuria	*LPIN1*
Adenine phosphoribosyltransferase deficiency	*APRT*
Alagille syndrome	*JAG1, NOTCH2*
Arthrogryposis-renal dysfunction-cholestasis syndrome	*VIPAS39, VPS33B*
Bardet-Biedl syndrome	*ARL6, BBIP1, IFT27, LZTFL1, MKKS, MKS1*
Bartter syndrome	*BSND, CLCNKB, SLC12A1*
Branchiootorenal spectrum disorders	*EYA1, SIX5*
CAKUT with or without VACTERL	*TRAP1*
Central hypoventilation syndrome	*ASCL1, EDN3, GDNF, PHOX2B, RET*
Ciliopathy	*CC2D2A, CEP290, IFT172, SDCCAG8*
Congenital anomalies of kidney and urinary tract	*DSTYK*
Congenital cardiac defects	*FOXH1, GATA4, GATA6, HAND2, NKX2-5, NR2F2, TAB2, ZIC3*
Cystinosis	*CTNS*
Cystinuria	*SLC3A1, SLC7A9*
Dent disease	*CLCN5*
Diabetes mellitus and renal disease	*LHX1*
Distal renal tubular acidosis with progressive nerve deafness	*ATP6V1B1*
Epidermolysis bullosa	*CD151, COL17A1, COL7A1, DSP, DST, EXPH5, ITGA6, ITGB4, KRT14, KRT5, LAMA3, LAMB3, LAMC2, PLEC*
Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosi	*COX4I2*
Familial Mediterranean fever	*MEFV*
Familial thrombotic thrombocytopenia purpura	*ADAMTS13*
Fanconi syndrome	*EHHADH, SLC34A1*
Fanconi-Bickel syndrome	*SLC2A2*
Hajdu-Cheney syndrome	*NOTCH2*
Heterotaxy syndrome	*ACVR2B, CRELD1, NODAL, ZIC3*
Hyperuricemia, pulmonary hypertension, renal failure, and alkalosi	*SARS2*
Hypomagnesemia	*CLDN19, CNNM2, TRPM6*
Hypoparathyroidism, sensorineural deafness, and renal disease	*GATA3*
Hypophosphatemic nephrolithiasis/osteoporosis	*SLC34A1*
Hypophosphatemic rickets	*CLCN5, PHEX, SLC34A3*
Idiopathic infantile hypercalcemia	*SLC34A1*
Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa	*ITGA3*
Joubert syndrome	*CC2D2A, CEP290, CEP41, NPHP1, NPHP4*
Kabuki Syndrome	*KMT2D*

Renal disease (cont.)

Disorder	Associated gene(s)
Leber congenital amaurosis	*AIPL1, CEP290, CRB1, CRX, GDF6, GUCY2D, IMPDH1, IQCB1, KCNJ13, LCA5, LRAT, NMNAT1, RPE65, RPGRIP1, TULP1*
Lowe syndrome	*OCRL*
Meckel syndrome	*B9D1, CEP290, EXOC4, NPHP3, TMEM67*
Medullary thyroid carcinoma	*RET*
Multicentric carpotarsal osteolysis syndrome	*MAFB*
Multiple endocrine neoplasia, type 2	*RET*
Nephrolithiasis	*CLCN5*
Nephronophthisis	*DCDC2, INVS, NPHP1, NPHP3, NPHP4, WDR19*
Nephropathic cystinosis	*CTNS*
Nephropathy with pretibial epidermolysis bullosa and deafness	*CD151*
Nephrotic syndrome	*ARHGDIA, DGKE, NPHS1*
Non-nephropathic ocular cystinosis	*CTNS*
Osteopetrosis with renal tubular acidosis	*CA2*
Pallister-Hall syndrome	*GLI3*
Pheochromocytoma	*RET, SDHB, SDHD*
Pierson syndrome	*LAMB2*
Polycystic kidney disease	*PKD2, PKHD1*
Primary hyperoxaluria	*AGXT, GRHPR*
Renal agenesis	*RET*
Renal coloboma syndrome	*PAX2*
Renal cysts and diabetes syndrome	*HNF1B*
Renal hypodysplasia	*PAX2*
Renal tubular acidosis, proximal, with ocular abnormalities	*SLC4A4*
Renal-hepatic-pancreatic dysplasia	*NPHP3*
Robinow syndrome	*ROR2, WNT5A*
Senior-Loken syndrome	*CEP290, IQCB1, NPHP1, NPHP4, SDCCAG8, WDR19*
Sensorineural deafness with mild renal dysfunction	*BSND*
Townes-Brocks syndrome	*SALL1*
Urofacial syndrome	*HPSE2*
VACTERL association	*ZIC3*
Wilms tumor-aniridia-genital anomalies-retardation syndrome	*WT1*
Xanthinuria	*XDH*

Gastrointestinal

Developmental anomalies

Heterotaxy syndrome	*ACVR2B, CRELD1, NODAL, ZIC3*
Kabuki syndrome	*KDM6A*
Pallister-Hall syndrome	*GLI3*
Townes-Brocks syndrome	*SALL1*
VACTERL association	*ZIC3*

Failure to thrive

Disorder	Associated gene(s)
Cardiofaciocutaneous syndrome	*BRAF, KRAS, MAP2K1*
Chylomicron retention disease	*SAR1B*
Homocystinuria-megaloblastic anemia	*MTR, MTRR*
Noonan syndrome	*A2ML1, BRAF, KRAS, LZTR1, NRAS, PTPN11, RAF1, RASA2, RIT1, RRAS, SOS1, SOS2*
Smith-Magenis syndrome	*RAI1, SMS*

Gastric

Congenital diaphragmatic hernia	*SLIT3*
Gastrointestinal defects and immunodeficiency syndrome	*TTC7A*
Meconium ileus, diarrhea	*GUCY2C*
Phosphorylase kinase deficiency of liver and muscle	*PHKB*

Pancreas

Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosi	*COX4I2*
Hereditary pancreatitis	*CTRC*
Mitchell-Riley syndrome	*RFX6*
Neonatal diabetes with cerebellar agenesis	*PTF1A*
Pancreatic agenesis	*PDX1, PTF1A*
Pancreatic lipase deficiency	*PNLIP*

Large and small intestines

Acrodermatitis enteropathica	*SLC39A4*
Congenital diarrhea	*DGAT1, GUCY2C, NEUROG3, SLC26A3*
Congenital short bowel syndrome	*CLMP*
Gastrointestinal defects and immunodeficiency syndrome	*TTC7A*
Goldberg-Shprintzen megacolon syndrome	*KIAA1279*
Hirschsprung disease	*RET*
Immunodysregulation, polyendocrinopathy, and enteropathy	*FOXP3*

Liver

Glycogen storage disease type IV	*GBE1*
Alagille syndrome	*JAG1, NOTCH2*
Arthrogryposis-renal dysfunction-cholestasis syndrome	*VIPAS39, VPS33B*
Congenital bile acid synthesis defect	*ABCD3, AKR1D1, AMACR, CYP7B1, HSD3B7, SLC10A2*
Crigler-Najjar syndrome	*UGT1A1*
Fanconi-Bickel syndrome	*SLC2A2*
Fatty liver disease	*MGAT1*
Glycogen storage disease	*AGL, ALDOA, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LDHA, PFKM, PHKA2, PHKG2, PRKAG2, PYGL, SLC37A4*
Hepatic bile acid conjugation	*SLC27A5*
Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis	*CLDN1*

Liver (cont.)

Disorder	Associated gene(s)
Infantile liver failure syndrome	*LARS*
Infantile transient liver failure	*TRMU*
Intrahepatic cholestasis of pregnancy	*ABCB11*
Neonatal cholestasis	*NR1H4*
Omenn syndrome	*RAG1*
Phosphorylase kinase deficiency of liver and muscle	*PHKB*
Progressive familial intrahepatic cholestasis	*ABCB11, ABCB4, TJP2, ATP8B1*
Transient familial neonatal hyperbilirubinemia	*UGT1A1*

Spleen

Intrahepatic cholestasis of pregnancy	*ABCB11*
Omenn syndrome	*RAG1*

Eye

Abnormal eye movement

Acro-renal-ocular syndrome	*SALL4*
Ataxia with oculomotor apraxia and hypoalbuminemia	*APTX*
Blepharophimosis, epicanthus inversus, and ptosis	*FOXL2*
Congenital fibrosis of extraocular muscles	*KIF21A, TUBB3*
Congenital nystagmus	*FRMD7*
Duane retraction syndrome	*CHN1*
Duane-radial ray syndrome	*SALL4*
Gaze palsy, horizontal, with progressive scoliosis	*ROBO3*
Griscelli syndrome	*MYO5A, RAB27A*
Holt-Oram syndrome	*SALL4, TBX5*
Joubert syndrome	*AHI1, ARL13B, C5orf42, CSPP1, INPP5E, KIF7, PDE6D, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237*
Mosaic variegated aneuploidy syndrome	*BUB1B*
Oculocutaneous albinism	*TYR*
Pelizaeus-Merzbacher disease	*PLP1*
Progressive external ophthalmoplegia	*DNA2, POLG2, SLC25A4*
Proximal myopathy and ophthalmoplegia	*MYH2*
Spastic paraplegia	*AP4B1, AP4E1, AP4M1, AP4S1, C12orf65, ERLIN2, HSPD1, IBA57, KIF1A, KIF5A, PLP1, TECPR2*

Anterior chamber dysgenesis

Aniridia, Peter anomaly	*PAX6*
Axenfeld-Rieger syndrome	*FOXC1, PITX2*
Branchiooculofacial syndrome	*TFAP2A*
Congenital glaucoma	*CYP1B1*
Klippel-Feil syndrome	*GDF3, GDF6, MEOX1, MYO18B*

Anterior chamber dysgenesis (cont.)

Disorder	Associated gene(s)
Lenz microphthalmia syndrome	*NAA10*
Linear skin defects with multiple congenital anomalies	*HCCS*
Martsolf syndrome	*RAB3GAP2*
Microphthalmia	*ABCB6, BCOR, BMP4, GDF3, PRSS56, RARB, SHH, SOX2, STRA6*
Oculofaciocardiodental syndrome	*BCOR*
Odgen syndrome	*NAA10*
Peters plus syndrome	*B3GLCT*
Warburg micro syndrome	*RAB18, RAB3GAP1, RAB3GAP2, TBC1D20*

Coloboma

CHARGE syndrome	*CHD7, SEMA3E*
CHIME syndrome	*PIGL*
Colobomatous macrophthalmia with microcornea	*SIX2*
Donnai-Barrow syndrome	*LRP2*
Kahrizi syndrome	*SRD5A3*
Manitoba oculotrichoanal syndrome	*FREM1*

Lens abnormality

Ayme-Gripp syndrome	*MAF*
Cataracts	*AGK, CRYAA, CRYBA1, CRYGC, CRYGD, EPHA2, NHS*
Cockayne syndrome	*ERCC6, ERCC8*
Combined mitochondrial complex deficiency	*GFER*
Congenital cataracts, facial dysmorphism, and neuropathy	*CTDP1*
Congenital cataracts, hearing loss, and neurodegeneration	*CCS, SLC33A1*
Congenital primary aphakia	*FOXE3*
Donnai-Barrow syndrome	*LRP2*
Lowe syndrome	*OCRL*
Marfan syndrome	*FBN1*
Marinesco-Sjögren syndrome	*SIL1*
Microcephaly, congenital cataract, and psoriasiform dermatitis	*MSMO1*
Nance-Horan syndrome	*NHS*
Sengers syndrome	*AGK*
Warburg micro syndrome	*RAB18, RAB3GAP1, RAB3GAP2, TBC1D20*
Weill-Marchesani syndrome	*ADAMTS10*

Optic nerve disease

AICA-ribosiduria due to ATIC deficiency	*ATIC*
Arts syndrome	*PRPS1*
Bosch-Boonstra-Schaaf optic atrophy syndrome	*NR2F1*
Charcot-Marie-Tooth disease	*DNM2, EGR2, FGD4, GARS, GDAP1, IGHMBP2, MPZ, NAGLU, PMP22, PRPS1, PRX, SURF1*
GAPO syndrome	*ANTXR1*

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Optic nerve disease (cont.)

Disorder	Associated gene(s)
Infantile cerebellar-retinal degeneration	*ACO2*
Microphthalmia	*ABCB6, BCOR, BMP4, GDF3, PRSS56, RARB, SHH, SOX2, STRA6*
Mitochondrial disease	*MFF*
Non-syndromic hearing loss	*ACTG1, MYH9, PNPT1, PRPS1, SIX1, SIX2, TBC1D24, TJP2*
Optic atrophy	*ACO2, OPA1, OPA3, TMEM126A*
Optic nerve hypoplasia and abnormalities of the central nervous system	*SOX2*
Phosphoribosylpyrophosphate synthetase superactivity	*PRPS1*
Warburg micro syndrome	*RAB18, RAB3GAP1, RAB3GAP2, TBC1D20*

Ptosis

3MC syndrome	*MASP1*
Baraitser-Winter syndrome	*ACTB, ACTG1*

Retinal degeneration

Alstrom syndrome	*ALMS1*
Bardet-Biedl syndrome	*ARL6, BBIP1, IFT27, LZTFL1, MKKS, MKS1*
Ciliopathy	*CC2D2A, CEP290, IFT172, SDCCAG8*
COACH syndrome	*CC2D2A, TMEM67*
Cohen syndrome	*VPS13B*
Cone-rod synaptic disorder	*CABP4*
Congenital stationary night blindness	*CACNA1F, GPR179, GRM6, LRIT3, NYX, TRPM1*
HARP syndrome	*PANK2*
Hypomagnesemia	*CLDN19, CNNM2, TRPM6*
Infantile cerebellar-retinal degeneration	*ACO2*
Joubert syndrome	*CC2D2A, CEP290, CEP41, NPHP1, NPHP4*
Knobloch syndrome	*COL18A1*
Leber congenital amaurosis	*AIPL1, CEP290, CRB1, CRX, GDF6, GUCY2D, IMPDH1, IQCB1, KCNJ13, LCA5, LRAT, NMNAT1, RPE65, RPGRIP1, TULP1*
Macular dystrophy with central cone involvement	*MFSD8*
Microcephaly and chorioretinopathy	*TUBGCP6*
Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation	*KIF11*
Norrie disease	*NDP*
Optic atrophy	*ACO2, OPA1, OPA3, TMEM126A*
Retinal degeneration	*ABCA4*
Retinitis pigmentosa	*HGSNAT, IMPDH1*
Senior-Loken syndrome	*CEP290, IQCB1, NPHP1, NPHP4, SDCCAG8, WDR19*
Usher syndrome	*USH1C*

Miscellaneous

Disorder	Associated gene(s)
Alacrima, achalasia, and mental retardation syndrome	*GMPPA*
Cerebroretinal microangiopathy with calcifications and cysts	*CTC1*
Dysplasminogenemia	*PLG*
Exudative vitreoretinopathy	*LRP5*
Hypomyelinating leukodystrophy	*AIMP1, FAM126A, GJC2, HSPD1, POLR1C, POLR3A, POLR3B, PYCR2, TUBB4A, VPS11*
Kabuki syndrome	*KDM6A*
Lacrimo-auriculo-dento-digital syndrome	*FGF10*
Lymphedema-distichiasis syndrome	*FOXC2*
Marden-Walker syndrome	*PIEZO2*
Megalocornea	*CHRDL1*
Orofacial cleft	*BMP4*
Osteopetrosis	*CLCN7, LRP5, OSTM1, TCIRG1, TNFRSF11A, TNFSF11*
Pierson syndrome	*LAMB2*
Retinal arterial macroaneurysm with supravalvular pulmonic stenosis	*IGFBP7*
Temtamy syndrome	*C12orf57*
Treacher Collins syndrome	*POLR1C, POLR1D, TCOF1*

Endocrine

Adrenal dysfunction

17-alpha-hydroxylase-deficient congenital adrenal hyperplasia	*CYP17A1*
3-beta-hydroxystero dehydrogenase-deficient congenital adrenal hyperplasia	*HSD3B2*
Adrenal hypoplasia congenita	*NR0B1*
Congenital adrenal hyperplasia	*CYP11B1*
Congenital lipoid adrenal hyperplasia	*STAR*
Corticosterone methyloxidase deficiency	*CYP11B2*
Glucocorticoid deficiency	*MC2R, MRAP, NNT*
Natural killer cell and glucocorticoid deficiency with DNA repair defect	*MCM4*
Natural killer cell and glucocorticoid deficiency, DNA repair defect	*MCM4*
Pseudohypoaldosteronism	*NR3C2, SCNN1A, SCNN1B, SCNN1G, WNK1*

Hyper-/hypo-calcemia

Familial hypocalciuric hypercalcemia	*AP2S1, GNA11*
Hypercalcemia	*CYP24A1*
Hypocalcemia	*GNA11*

Hyper/hypo-glycemia

3-hydroxyacyl-coenzyme dehydrogenase deficiency	*HADH*
Abnormal insulin secretion	*TFB1M*
Adrenocorticotropic hormone deficiency	*TBX19*

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Hyper/hypo-glycemia (cont.)

Disorder	Associated gene(s)
Bardet-Biedl syndrome	*ARL6, BBIP1, IFT27, LZTFL1, MKKS, MKS1*
Beckwith-Wiedemann syndrome	*CDKN1C*
Congenital hyperinsulinism	*UCP2*
Cytosolic phosphoenolpyruvate carboxykinase-1 deficiency	*PCK1*
Erythrocyte lactate transporter defect	*SLC16A1*
Familial hyperinsulinism	*ABCC8, GCK, HADH, KCNJ11*
Fanconi renotubular syndrome	*HNF4A*
Fructose 1,6 bisphosphatase deficiency	*FBP1*
Glucose metabolism	*ARRDC4*
Growth retardation with deafness and mental retardation due to IGF1 deficiency	*IGF1*
Hyperinsulinemic hypoglycemia	*INSR*
Hyperinsulinism-hyperammonemia syndrome	*GLUD1*
Hyperproinsulinemia	*INS*
Hypoinsulinemic hypoglycemia with hemihypertrophy	*AKT2*
Maturity-onset diabetes of the young	*BLK, HNF1A, KLF11, NEUROD1, NKX2-2*
Microcephaly, epilepsy, and diabetes syndrome	*IER3IP1*
Microcephaly, short stature, and impaired glucose metabolism	*TRMT10A*
Mitchell-Riley syndrome	*RFX6*
Monocarboxylate transporter deficiency	*SLC16A1*
Multiple epiphyseal dysplasia with early-onset diabetes mellitus	*EIF2AK3*
Neonatal diabetes mellitus	*ABCC8, GLIS3, INS, KCNJ11, ZFP57*
Nephrogenic diabetes insipidus	*AQP2*
Neurodevelopmental disorder with diabetes	*GFM2*
Pancreatic agenesis	*PDX1, PTF1A*
Renal cysts and diabetes syndrome	*HNF1B*
SLC16A1-related hyperinsulinism	*SLC16A1*
Wolfram syndrome	*WFS1*
Wolfram-like syndrome	*WFS1*

Hyper-/hypo-parathyroidism

Bamforth-Lazarus syndrome	*FOXE1*
Benign hereditary chorea	*NKX2-1*
Choreoasthetosis, hypothyroidism, and neonatal respiratory distress	*NKX2-1*
Combined pituitary hormone deficiency	*HESX1, LHX3, LHX4, POU1F1, PROP1*
Congenital cardiac defects	*FOXH1, GATA4, GATA6, HAND2, NKX2-5, NR2F2, TAB2, ZIC3*
Congenital hypothyroidism	*NKX2-5, PAX8, TSHB, TSHR*
Generalized thyrotropin-releasing hormone resistance	*TRHR*
Hyperparathyroidism	*CASR*
Hypocalciuric hypercalcemia, type I	*CASR*

Hyper-/hypo-parathyroidism (cont.)

Disorder	Associated gene(s)
Hypoparathyroidism-retardation-dysmorphism syndrome	*TBCE*
Hypoparathyroidism, sensorineural deafness, and renal disease	*GATA3*
Hypothyroidism, central, and testicular enlargement	*IGSF1*
Kenny-Caffey syndrome	*TBCE*
McCune-Albright syndrome	*GNAS*
Nonautoimmune hyperthyroidism	*TSHR*
Progressive osseous heteroplasia	*GNAS*
Pseudohypoparathyroidism	*GNAS*
Thyroid carcinoma	*TSHR*
Thyroid dyshormonogenesis	*DUOX2, DUOXA2, IYD, SECISBP2, TG, TPO*
Thyroid hormone resistance	*THRB*

Pituitary abnormalities

Combined pituitary hormone deficiency	*HESX1, LHX3, LHX4, POU1F1, PROP1*
Culler-Jones syndrome	*GLI2*
Endocrine-cerebroosteodysplasia	*ICK*
Hypogonadotropic hypogonadism	*FGF8, KISS1, KISS1R, NSMF, PROKR2, TAC3, TACR3*
Panhypopituitarism	*SOX3*
Septo-optic dysplasia	*HESX1*

Miscellaneous

Adrenal hypoplasia congenita	*NR0B1*
Antenatal corticosteroid exposure, risk factor	*CRHBP*
Antley-Bixler syndrome	*POR*
Bardet-Biedl syndrome	*ARL6, BBIP1, IFT27, LZTFL1, MKKS, MKS1*
Berardinelli-Seip congenital lipodystrophy	*AGPAT2*
Borjeson-Forssman-Lehmann syndrome	*PHF6*
Ciliopathy	*CC2D2A, CEP290, IFT172, SDCCAG8*
Complex gycerol kinase deficiency	*NR0B1*
Congenital generalized lipodystrophy	*BSCL2*
Enlarged vestibular aqueduct	*SLC26A4*
Familial lipoprotein lipase deficiency	*LPL*
Glucocorticoid resistance	*NR3C1*
Hereditary lymphedema	*FLT4, VEGFC*
Histiocytosis-lymphadenopathy plus syndrome	*SLC29A3*
Hyperchlorhidrosis	*CA12*
Hypergonadotropic hypogonadism	*LHCGR*
Hyperlipoproteinemia	*APOC2*
Hypertriglyceridemia	*GPD1*
Immunodysregulation, polyendocrinopathy, and enteropathy	*FOXP3*

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Miscellaneous (cont.)

Disorder	Associated gene(s)
Joubert syndrome	*CC2D2A, CEP290, CEP41, NPHP1, NPHP4, AHI1, ARL13B, C5orf42, CSPP1, INPP5E, KIF7, PDE6D, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237*
Kowarski syndrome	*GH1*
Leber congenital amaurosis	*AIPL1, CEP290, CRB1, CRX, GDF6, GUCY2D, IMPDH1, IQCB1, KCNJ13, LCA5, LRAT, NMNAT1, RPE65, RPGRIP1, TULP1*
Lymphedema-distichiasis syndrome	*FOXC2*
McCune-Albright syndrome	*GNAS*
McKusick-Kaufman syndrome	*MKKS*
Meckel syndrome	*B9D1, CEP290, EXOC4, NPHP3, TMEM67*
Obesity	*MC4R*
Pallister-Hall syndrome	*GLI3*
Pendred syndrome	*FOXI1, SLC26A4*
Prader-Willi-like syndrome	*MAGEL2*
Progressive osseous heteroplasia	*GNAS*
Proopiomelanocortin deficiency	*POMC*
Pseudohypoaldosteronism	*NR3C2, SCNN1A, SCNN1B, SCNN1G, WNK1*
Pseudohypoparathyroidism	*GNAS*
Resistance to insulin-like growth factor I	*IGF1R*
Senior-Loken syndrome	*CEP290, IQCB1, NPHP1, NPHP4, SDCCAG8, WDR19*
Silver syndrome	*BSCL2*

Central nervous system

Abnormal reflexes

Allan-Herndon-Dudley syndrome	*SLC16A2*
Amish infantile epilepsy syndrome	*ST3GAL5*
Amyotrophic lateral sclerosis	*FIG4*
Benign hereditary chorea	*NKX2-1*
BH4-deficient hyperphenylalaninemia	*GCH1, PCBD1, QDPR*
Cerebellar hypoplasia and mental retardation syndrome	*VLDLR*
Cerebral palsy	*KANK1*
Cerebral palsy, spastic quadriplegic	*GAD1*
Charcot-Marie-Tooth disease	*DNM2, EGR2, FGD4, GARS, GDAP1, IGHMBP2, MPZ, NAGLU, PMP22, PRPS1, PRX, SURF1*
Dysautonomia	*IKBKAP*
GM2-gangliosidosis	*GM2A*
Hereditary hyperekplexia	*GLRA1, GLRB*
Hereditary sensory and autonomic neuropathy	*NGF, SCN11A, SPTLC1, WNK1*
Hereditary sensory neuropathy, spastic paraplegia	*CCT5*
Hypomyelinating leukodystrophy	*AIMP1, FAM126A, GJC2, HSPD1, POLR1C, POLR3A, POLR3B, PYCR2, TUBB4A, VPS11*

Abnormal reflexes (cont.)

Disorder	Associated gene(s)
Infantile cerebellar-retinal degeneration	*ACO2*
Lethal congenital contracture syndrome	*ADCY6, CNTNAP1, DNM2, ERBB3, GLE1, MYBPC1, PIP5K1C*
Lowe syndrome	*OCRL*
Optic atrophy	*ACO2, OPA1, OPA3, TMEM126A*
Psychomotor retardation, epilepsy, and craniofacial dysmorphism	*SNIP1*
Smith-Magenis syndrome	*RAI1, SMS*
Spastic paraplegia	*AP4B1, AP4E1, AP4M1, AP4S1, C12orf65, ERLIN2, HSPD1, IBA57, KIF1A, KIF5A, PLP1, TECPR2*
Yunis-Varon Syndrome	*FIG4*

Ataxia

2-aminoadipic 2-oxoadipic aciduria	*DHTKD1*
3-methylglutaconyl-CoA hydratase deficiency	*AUH*
Alacrima, achalasia, and mental retardation syndrome	*GMPPA*
Alexander disease	*GFAP*
Allan-Herndon-Dudley syndrome	*SLC16A2*
Alpha-mannosidosis	*MAN2B1*
Angelman syndrome	*UBE3A*
Arts syndrome	*PRPS1*
Ataxia-oculomotor apraxia	*PNKP*
Ataxia-telangiectasia	*ATM*
Ataxia-telangiectasia-like disorder	*MRE11A*
Bilateral frontoparietal polymicrogyria	*ADGRG1*
Carbamoylphosphate synthetase I deficiency	*CPS1*
Cerebellar ataxia	*ATP8A2*
Cerebellar ataxia and mental retardation syndrome	*CA8*
Cerebellar ataxia, mental retardation, and dysequilibrium syndrome	*WDR81*
Cerebellar hypoplasia and mental retardation syndrome	*VLDLR*
Charcot-Marie-Tooth disease	*DNM2, EGR2, FGD4, GARS, GDAP1, IGHMBP2, MPZ, NAGLU, PMP22, PRPS1, PRX, SURF1*
Ciliopathy	*CC2D2A, CEP290, IFT172, SDCCAG8*
COACH syndrome	*CC2D2A, TMEM67*
Cockayne syndrome	*ERCC6, ERCC8*
Congenital mirror movements	*RAD51*
D-bifunctional protein deficiency	*HSD17B4*
Enlarged vestibular aqueduct	*SLC26A4*
Epileptic encephalopathy	*ALG13, ARHGEF9, ARX, CDKL5, CHD2, CLCN4, DNM1, DOCK7, EEF1A2, GABRA1, GNAO1, GRIN2B, HCN1, KCNA2, KCNB1, KCNH5, KCNQ2, KCNT1, MECP2, NECAP1, PCDH19, PIGQ, PLCB1, SCN2A, SCN8A, SIK1, SLC12A5, SLC13A5, SLC25A12, SLC25A22, SPTAN1, ST3GAL3, STXBP1, SZT2, TBC1D24, WWOX*

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Ataxia (cont.)

Disorder	Associated gene(s)
Generalized epilepsy and paroxysmal dyskinesia	*KCNMA1*
Glutathione synthetase deficiency	*GSS*
Guanidinoacetate methyltransferase deficiency	*GAMT*
Hawkinsinuria	*HPD*
Hypomyelinating leukodystrophy	*AIMP1, FAM126A, GJC2, HSPD1, POLR1C, POLR3A, POLR3B, PYCR2, TUBB4A, VPS11*
Joubert syndrome	*CC2D2A, CEP290, CEP41, NPHP1, NPHP4*
Leber congenital amaurosis	*AIPL1, CEP290, CRB1, CRX, GDF6, GUCY2D, IMPDH1, IQCB1, KCNJ13, LCA5, LRAT, NMNAT1, RPE65, RPGRIP1, TULP1*
Leukoencephalopathy	*DARS2, RNASET2*
Leukoencephalopathy with vanishing white matter	*EIF2B4*
Meckel syndrome	*B9D1, CEP290, EXOC4, NPHP3, TMEM67*
Megalencephalic leukoencephalopathy	*HEPACAM, MLC1*
Microcephaly, seizures, and developmental delay	*PNKP*
Nemaline myopathy	*ACTA1, KLHL40, KLHL41, LMOD3, NEB, TNNT1, TPM2, TPM3*
Neuronal ceroid lipofuscinosis	*CLN3, CLN5, CLN6, CLN8, CTSD, MFSD8, PPT1, TPP1*
Neuronaxonal dystrophy	*PLA2G6*
Non-syndromic hearing loss	*ACTG1, MYH9, PNPT1, PRPS1, SIX1, SIX2, TBC1D24, TJP2*
Pelizaeus-Merzbacher disease	*PLP1*
Perrault syndrome	*CLPP, HARS2, HSD17B4, LARS2*
Phosphoribosylpyrophosphate synthetase superactivity	*PRPS1*
Progressive myoclonic epilepsy	*EPM2A, KCTD7, NHLRC1, PRICKLE1*
Rett syndrome	*MECP2*
Senior-Loken syndrome	*CEP290, IQCB1, NPHP1, NPHP4, SDCCAG8, WDR19*
SESAME syndrome	*KCNJ10*
Spastic paraplegia	*AP4B1, AP4E1, AP4M1, AP4S1, C12orf65, ERLIN2, HSPD1, IBA57, KIF1A, KIF5A, PLP1, TECPR2*
Spinocerebellar ataxia	*ATP2B3, GRM1, ITPR1, SPTBN2*
Sulfite oxidase deficiency	*SUOX*
Syndromic mental retardation	*ADAT3, ATP6AP2, CLIC2, CTCF, CTNNB1, CUL4B, DLG3, DYNC1H1, DYRK1A, FOXP1, GATAD2B, GRIN1, HUWE1, KDM5C, KIAA2022, KIF1A, KPTN, MAN1B1, MBD5, NSUN2, PHF8, PURA, SETBP1, SETD5, SOX11, SYNGAP1, TRAPPC9, TUSC3, UPF3B, USP9X*
Tyrosinemia, type III	*HPD, TAT*

Brain malformations

Disorder	Associated gene(s)
3-methylglutaconyl-C hydratase deficiency	*AUH*
Amish lethal microcephaly	*SLC25A19*
Andermann syndrome	*SLC12A6*
Ataxia-oculomotor apraxia	*PNKP*
Ataxia-telangiectasia-like disorder	*MRE11A*
Band-like calcification, simplified gyration, polymicrogyria	*OCLN*
Bardet-Biedl syndrome	*ARL6, BBIP1, IFT27, LZTFL1, MKKS, MKS1*
Bilateral frontoparietal polymicrogyria	*ADGRG1*
Bohring-Opitz syndrome	*ASXL1*
Canavan disease	*ASPA*
Carbonic anhydrase VA deficiency	*CA5A*
Cardioencephalomyopathy	*SCO2*
Cerebellar ataxia	*ATP8A2*
Cerebellar ataxia and mental retardation syndrome	*CA8*
Cerebellar ataxia, mental retardation, and dysequilibrium syndrome	*WDR81*
Cerebellar hypoplasia and mental retardation syndrome	*VLDLR*
Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome	*SNAP29*
Cerebral palsy	*KANK1*
Cerebroretinal microangiopathy with calcifications and cysts	*CTC1, CTC1*
CHILD syndrome	*NSDHL*
Christianson syndrome	*SLC9A6*
Chudley-McCullough syndrome	*GPSM2*
Ciliopathy	*CC2D2A, CEP290, IFT172, SDCCAG8*
CK syndrome	*NSDHL*
COACH syndrome	*CC2D2A, TMEM67*
Combined D-2- and L-2-hydroxyglutaric aciduria	*SLC25A1*
Complex cortical dysplasia	*KIF2A, KIF5C, TUBB2A, TUBB3, TUBG1*
Congenital fibrosis of extraocular muscles	*KIF21A, TUBB3*
Congenital variant Rett syndrome	*FOXG1*
Craniosynostosis	*ERF, TCF12*
Dandy-Walker malformation with occipital cephalocele	*NID1*
Endocrine-cerebroosteodysplasia	*ICK*
Epileptic encephalopathy, global delay, and cerebellar atrophy	*CACNA2D2*
Escobar syndrome	*CHRNG*
FG syndrome	*CASK, FLNA*
Forebrain defects	*TDGF1*
Goldberg-Shprintzen megacolon syndrome	*KIAA1279*
Growth retardation, developmental delay, and facial dysmorphism	*FTO*
Hartsfield syndrome	*FGFR1*
Hennekam lymphangiectasia-lymphedema syndrome	*FAT4*

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Brain malformations (cont.)

Disorder	Associated gene(s)
Holoprosencephaly	*CDON, DISP1, GLI2, SHH, SIX3, TGIF1, ZIC2*
Hydrocephalus	*L1CAM*
Hypomyelinating leukodystrophy	*AIMP1, FAM126A, GJC2, HSPD1, POLR1C, POLR3A, POLR3B, PYCR2, TUBB4A, VPS11*
Hypomyelination with brainstem, spinal cord involvement, and leg spasticity	*DARS*
Intellectual disability, microcephaly, and pontine cerebellar hypoplasia	*CASK*
Joubert syndrome	*CC2D2A, CEP290, CEP41, NPHP1, NPHP4, AHI1, ARL13B, C5orf42, CSPP1, INPP5E, KIF7, PDE6D, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237*
Leber congenital amaurosis	*AIPL1, CEP290, CRB1, CRX, GDF6, GUCY2D, IMPDH1, IQCB1, KCNJ13, LCA5, LRAT, NMNAT1, RPE65, RPGRIP1, TULP1*
Leukoencephalopathy with vanishing white matter	*EIF2B4*
Lissencephaly	*DCX, LAMB1, NDE1, PAFAH1B1, RELN, TUBA1A*
Meckel syndrome	*B9D1, CEP290, EXOC4, NPHP3, TMEM67*
Megalencephaly-capillary syndrome	*PIK3CA*
Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome	*AKT3, CCND2, PIK3R2*
Mevalonic aciduria	*MVK*
Microcephaly, epilepsy, and diabetes syndrome	*IER3IP1*
Microcephaly, progressive, with seizures and cerebral and cerebellar atrophy	*QARS*
Microcephaly, seizures, and developmental delay	*PNKP*
Microcephaly, short stature, and polymicrogyria with seizures	*RTTN*
Multiple pterygium syndrome	*CHRNG*
Muscular dystrophy-dystroglycanopathy	*DAG1, FKRP, FKTN, GMPPB, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, TMEM5, B3GALNT2, B4GAT1*
Neonatal diabetes with cerebellar agenesis	*PTF1A*
Nephronophthisis	*DCDC2, INVS, NPHP1, NPHP3, NPHP4, WDR19*
Neurodevelopmental disorder with diabetes	*GFM2*
Non-syndromic hydrocephalus	*CCDC88C, MPDZ*
Pancreatic agenesis	*PDX1, PTF1A*
Periventricular heterotopia	*ARFGEF2*
Polymicrogyria	*TUBA8, TUBB2B*
Pontocerebellar hypoplasia	*AMPD2, CHMP1A, CLP1, EXOSC3, RARS2, SEPSECS, TSEN2, TSEN34, TSEN54, VPS53, VRK1*
Progressive microcephaly with seizures and brain atrophy	*MED17*

Brain malformations (cont.)

Disorder	Associated gene(s)
Proliferative vasculopathy and hydraencephaly-hydrocephaly syndrome	*FLVCR2*
Ritscher-Schinzel syndrome	*CCDC22, KIAA0196*
Schuss-Hoeijmakers syndrome	*PACS1*
Senior-Loken syndrome	*CEP290, IQCB1, NPHP1, NPHP4, SDCCAG8, WDR19*
Shprintzen-Goldberg syndrome	*SKI*
Spastic paraplegia	*AP4B1, AP4E1, AP4M1, AP4S1, C12orf65, ERLIN2, HSPD1, IBA57, KIF1A, KIF5A, PLP1, TECPR2*
Syndromic mental retardation	*ADAT3, ATP6AP2, CLIC2, CTCF, CTNNB1, CUL4B, DLG3, DYNC1H1, DYRK1A, FOXP1, GATAD2B, GRIN1, HUWE1, KDM5C, KIAA2022, KIF1A, KPTN, MAN1B1, MBD5, NSUN2, PHF8, PURA, SETBP1, SETD5, SOX11, SYNGAP1, TRAPPC9, TUSC3, UPF3B, USP9X*
Temtamy syndrome	*C12orf57*
Thiamine metabolism dysfunction syndrome	*SLC19A3, SLC25A19*
Van Maldergem syndrome	*FAT4*
Vici syndrome	*EPG5*

Hypotonia

17-beta-hydroxysteroid dehydrogenase X deficiency	*HSD17B10*
2-aminoadipic 2-oxoadipic aciduria	*DHTKD1*
3-hydroxyisobutryl-CoA hydrolase deficiency	*HIBCH, HMGCL*
Adenosine kinase deficiency	*ADK*
Aicardi-Goutieres syndrome	*ADAR, IFIH1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1*
Alacrima, achalasia, and mental retardation syndrome	*GMPPA*
Allan-Herndon-Dudley syndrome	*SLC16A2*
Alpha-mannosidosis	*MAN2B1*
Aminoacylase deficiency	*ACY1*
Amish infantile epilepsy syndrome	*ST3GAL5*
Amish lethal microcephaly	*SLC25A19*
Andermann syndrome	*SLC12A6*
Angelman Syndrome	*UBE3A*
Arts syndrome	*PRPS1*
Ataxia-oculomotor apraxia	*PNKP*
Bainbridge-Ropers syndrome	*ASXL3*
BH4-deficient hyperphenylalaninemia	*GCH1, PCBD1, QDPR*
Canavan disease	*ASPA*
Cerebellar hypoplasia and mental retardation syndrome	*VLDLR*
Cerebral creatine deficiency syndrome	*SLC6A8*
Cerebral palsy	*KANK1*
Cerebral palsy, spastic quadriplegic	*GAD1*
Charcot-Marie-Tooth disease	*DNM2, EGR2, FGD4, GARS, GDAP1, IGHMBP2, MPZ, NAGLU, PMP22, PRPS1, PRX, SURF1*

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Hypotonia (cont.)

Disorder	Associated gene(s)
Christianson syndrome	*SLC9A6*
Ciliopathy	*CC2D2A, CEP290, IFT172, SDCCAG8*
COACH syndrome	*CC2D2A, TMEM67*
Cockayne syndrome	*ERCC6, ERCC6, ERCC8*
Coffin-Lowry syndrome	*RPS6KA3*
Cognitive impairment with or without cerebellar ataxia	*SCN8A*
Combined D-2- and L-2-hydroxyglutaric aciduria	*SLC25A1*
Complex cortical dysplasia	*KIF2A, KIF5C, TUBB2A, TUBB3, TUBG1*
Congenital contractures, hypotonia, and developmental delay	*NALCN*
Congenital fibrosis of extraocular muscles	*KIF21A, TUBB3*
Congenital muscular dystrophy	*CHKB, ITGA7, LAMA2, LMNA, POMT1, POMT2*
Congenital variant Rett syndrome	*FOXG1*
D-2-hydroxyglutaric aciduria	*D2HGDH*
D-glyceria aciduria	*GLYCTK*
Desanto-Shinawi syndrome	*WAC*
Dihydropyrimidine dehydrogenase deficiency	*DPYD*
DOOR syndrome	*TBC1D24*
Dopa-responsive dystonia	*SPR*
Dopamine beta-hydroxylase deficiency	*DBH*
Dysautonomia	*IKBKAP*
Enlarged vestibular aqueduct	*SLC26A4*
Epileptic encephalopathy	*ALG13, ARHGEF9, ARX, CDKL5, CHD2, CLCN4, DNM1, DOCK7, EEF1A2, GABRA1, GNAO1, GRIN2B, HCN1, KCNA2, KCNB1, KCNH5, KCNQ2, KCNT1, MECP2, NECAP1, PCDH19, PIGQ, PLCB1, SCN2A, SCN8A, SIK1, SLC12A5, SLC13A5, SLC25A12, SLC25A22, SPTAN1, ST3GAL3, STXBP1, SZT2, TBC1D24, WWOX*
Ethylmalonic encephalopathy	*ETHE1*
Familial myoclonic epilepsy	*TBC1D24*
FG syndrome	*CASK, FLNA*
GM2-gangliosidosis	*GM2A*
Guanidinoacetate methyltransferase deficiency	*GAMT*
Helsmoortel-van der Aa syndrome	*ADNP*
Homocystinuria	*CBS, MTHFR*
Hypokalemic periodic paralysis	*CACNA1S*
Hypomagnesemia	*CLDN19, CNNM2, TRPM6*
Hypomyelinating leukodystrophy	*AIMP1, FAM126A, GJC2, HSPD1, POLR1C, POLR3A, POLR3B, PYCR2, TUBB4A, VPS11*
Hypotonia-cystinuria syndrome	*PREPL*
Infantile cerebellar-retinal degeneration	*ACO2*

Hypotonia (cont.)

Disorder	Associated gene(s)
Infantile hypotonia, psychomotor retardation, and characteristic facies	*NALCN*
Intellectual disability, microcephaly, and pontine cerebellar hypoplasia	*CASK*
Joubert syndrome	*CC2D2A, CEP290, CEP41, NPHP1, NPHP4*
Kaufman oculocerebrofacial syndrome	*UBE3B*
Kleefstra syndrome	*EHMT1*
Koolen-De Vries syndrome	*KANSL1*
L-2-hydroxyglutaric aciduria	*L2HGDH*
L-arginine:glyci amidinotransferase deficiency	*GATM*
Leber congenital amaurosis	*AIPL1, CEP290, CRB1, CRX, GDF6, GUCY2D, IMPDH1, IQCB1, KCNJ13, LCA5, LRAT, NMNAT1, RPE65, RPGRIP1, TULP1*
Lowe syndrome	*OCRL*
Meckel syndrome	*B9D1, CEP290, EXOC4, NPHP3, TMEM67*
Menkes disease	*ATP7A*
Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations	*MEF2C*
Microcephaly, epilepsy, and diabetes syndrome	*IER3IP1*
Microcephaly, progressive, with seizures and cerebral and cerebellar atrophy	*QARS*
Microcephaly, seizures, and developmental delay	*PNKP*
Mowat-Wilson syndrome	*ZEB2*
Multiple congenital anomalies-hypotonia-seizure syndrome	*PIGA, PIGN, PIGT*
Muscular dystrophy-dystroglycanopathy	*DAG1, FKRP, FKTN, GMPPB, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, TMEM5, B3GALNT2, B4GAT1*
Myoclonic dystonia	*SGCE*
Nephronophthisis	*DCDC2, INVS, NPHP1, NPHP3, NPHP4, WDR19*
Non-ketotic hyperglycinemia	*AMT, GCSH, GLDC*
Non-syndromic hearing loss	*ACTG1, MYH9, PNPT1, PRPS1, SIX1, SIX2, TBC1D24, TJP2*
Opitz-Kaveggia syndrome	*MED12*
Optic atrophy	*ACO2, OPA1, OPA3, TMEM126A*
Phelan-McDermid syndrome	*SHANK3*
Phosphoribosylpyrophosphate synthetase superactivity	*PRPS1*
Prader-Willi syndrome	*SNRPN*
Prader-Willi-like syndrome	*MAGEL2*
Progressive neonatal encephalopathy	*ADSL*
Pseudo-neonatal adrenoleukodystrophy	*ACOX1*
Psychomotor retardation, epilepsy, and craniofacial dysmorphism	*SNIP1*
Pyridoxine-dependent epilepsy	*ALDH7A1*
Schuss-Hoeijmakers syndrome	*PACS1*

Hypotonia (cont.)

Disorder	Associated gene(s)
Senior-Loken syndrome	*CEP290, IQCB1, NPHP1, NPHP4, SDCCAG8, WDR19*
SESAME syndrome	*KCNJ10*
Sialic acid storage disorder	*SLC17A5*
Singleton-Merten syndrome	*IFIH1*
Smith-Lemli-Opitz syndrome	*DHCR7*
Smith-Magenis syndrome	*RAI1, SMS*
Spastic paraplegia	*AP4B1, AP4E1, AP4M1, AP4S1, C12orf65, ERLIN2, HSPD1, IBA57, KIF1A, KIF5A, PLP1, TECPR2*
Syndromic mental retardation	*ADAT3, ATP6AP2, CLIC2, CTCF, CTNNB1, CUL4B, DLG3, DYNC1H1, DYRK1A, FOXP1, GATAD2B, GRIN1, HUWE1, KDM5C, KIAA2022, KIF1A, KPTN, MAN1B1, MBD5, NSUN2, PHF8, PURA, SETBP1, SETD5, SOX11, SYNGAP1, TRAPPC9, TUSC3, UPF3B, USP9X*
Thiamine metabolism dysfunction syndrome	*SLC19A3, SLC25A19*
Warsaw breakage syndrome	*DDX11*
Xia-Gibbs syndrome	*AHDC1*

Macrocephaly

Adenosine kinase deficiency	*ADK*
Alpha-mannosidosis	*MAN2B1*
Bannayan-Riley-Ruvalcaba syndrome	*PTEN*
Canavan disease	*ASPA*
Cantu syndrome	*ABCC9, KCNJ8*
Cowden syndrome	*PTEN, SDHB*
D-2-hydroxyglutaric aciduria	*D2HGDH*
Legius syndrome	*SPRED1*
Megalencephalic leukoencephalopathy	*HEPACAM, MLC1*
Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome	*AKT3, CCND2, PIK3R2*
Noonan-like syndrome with loose anagen hair	*SHOC2*
Osteopathia striata with cranial sclerosis	*AMER1*
Proteus syndrome	*PTEN*
PTEN-related cardiovascular disease	*PTEN*
Ritscher-Schinzel syndrome	*CCDC22, KIAA0196*
Simpson-Golabi-Behmel syndrome	*GPC3*
Smith-Kingsmore syndrome	*MTOR*

Microcephaly

Achalasia-addisonianism-alacrimia syndrome	*AAAS*
Aicardi-Goutieres syndrome	*ADAR, IFIH1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1*
Allan-Herndon-Dudley syndrome	*SLC16A2*
Alpha-thalassemia intellectual disability syndrome	*ATRX*
Amish lethal microcephaly	*SLC25A19*

Microcephaly (cont.)

Disorder	Associated gene(s)
Angelman syndrome	*UBE3A*
Asparagine synthetase deficiency	*ASNS*
Ataxia-oculomotor apraxia	*PNKP*
Band-like calcification, simplified gyration, and polymicrogyria	*OCLN*
BH4-deficient hyperphenylalaninemia	*GCH1, PCBD1, QDPR*
Borjeson-Forssman-Lehmann syndrome	*PHF6*
Cerebral creatine deficiency syndrome	*SLC6A8*
Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome	*SNAP29*
Cerebrocostomandibular syndrome	*SNRPB*
Cerebrooculofacioskeletal syndrome	*ERCC1, ERCC5*
CHILD syndrome	*NSDHL*
Christianson syndrome	*SLC9A6*
CK syndrome	*NSDHL*
Cockayne syndrome	*ERCC6, ERCC6, ERCC8*
Coffin-Lowry syndrome	*RPS6KA3*
Cohen syndrome	*VPS13B*
Complex cortical dysplasia	*KIF2A, KIF5C, TUBB2A, TUBB3, TUBG1*
Congenital contractures, hypotonia, and developmental delay	*NALCN*
Congenital disorder of glycosylation	*ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, B4GALT1, COG1, COG4, COG5, COG6, COG7, COG8, DDOST, DOLK, DPAGT1, DPM1, DPM2, DPM3, MGAT2, MOGS, MPDU1, MPI, NGLY1, NHEJ1, PGM1, PGM3, PMM2, RFT1, RPN2, SLC35A1, SLC35A2, SLC35C1, SRD5A3, SSR4, STT3A, STT3B, TMEM165*
Congenital fibrosis of extraocular muscles	*KIF21A, TUBB3*
Congenital muscular dystrophy	*CHKB, ITGA7, LAMA2, LMNA, POMT1, POMT2*
Congenital variant Rett syndrome	*FOXG1*
D-glyceria aciduria	*GLYCTK*
Dihydropyrimidine dehydrogenase deficiency	*DPYD*
Dihydropyrimidinuria	*DPYS*
DOOR syndrome	*TBC1D24*
Dopa-responsive dystonia	*SPR*
Epilepsy, hearing loss, and mental retardation syndrome	*SPATA5*
Epileptic encephalopathy	*ALG13, ARHGEF9, ARX, CDKL5, CHD2, CLCN4, DNM1, DOCK7, EEF1A2, GABRA1, GNAO1, GRIN2B, HCN1, KCNA2, KCNB1, KCNH5, KCNQ2, KCNT1, MECP2, NECAP1, PCDH19, PIGQ, PLCB1, SCN2A, SCN8A, SIK1, SLC12A5, SLC13A5, SLC25A12, SLC25A22, SPTAN1, ST3GAL3, STXBP1, SZT2, TBC1D24, WWOX*
Familial cutaneous telangiectasia and cancer syndrome	*ATR*
Familial myoclonic epilepsy	*TBC1D24*
Feingold syndrome	*MYCN*

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Microcephaly (cont.)

Disorder	Associated gene(s)
FG syndrome	*CASK, FLNA*
Galloway-Mowat syndrome	*WDR73*
Genitopatellar syndrome	*KAT6B*
Goldberg-Shprintzen megacolon syndrome	*KIAA1279*
Growth retardation, developmental delay, facial dysmorphism	*FTO*
Holoprosencephaly	*CDON, DISP1, GLI2, SHH, SIX3, TGIF1, ZIC2*
Homocystinuria	*CBS, MTHFR*
Hypoparathyroidism-retardation-dysmorphism syndrome	*TBCE*
Infantile hypotonia, psychomotor retardation, characteristic facies	*NALCN*
Intellectual disability, microcephaly, and pontine cerebellar hypoplasia	*CASK*
Johanson-Blizzard syndrome	*UBR1*
Juvenile polyposis	*SMAD4*
Kaufman oculocerebrofacial syndrome	*UBE3B*
Kenny-Caffey syndrome	*TBCE*
Leukoencephalopathy	*DARS2, RNASET2*
Meier-Gorlin syndrome	*CDC6, CDT1, ORC1, ORC4, ORC6*
Mevalonic aciduria	*MVK*
Microcephaly and chorioretinopathy	*TUBGCP6*
Microcephaly and variable mental retardation	*DPP6*
Microcephaly-capillary malformation syndrome	*STAMBP*
Microcephaly, epilepsy, and diabetes syndrome	*IER3IP1*
Microcephaly, progressive, with seizures and cerebral and cerebellar atrophy	*QARS*
Microcephaly, seizures, and developmental delay	*PNKP*
Microcephaly, short stature, and polymicrogyria with seizures	*RTTN*
Mosaic variegated aneuploidy syndrome	*BUB1B*
Mowat-Wilson syndrome	*ZEB2*
Myhre syndrome	*SMAD4*
Natural killer cell and glucocorticoid deficiency with DNA repair defect	*MCM4*
Natural killer cell and glucocorticoid deficiency, DNA repair defect	*MCM4*
Neonatal diabetes with cerebellar agenesis	*PTF1A*
Neu-Laxova syndrome	*PHGDH, PSAT1*
Nijmegen breakage syndrome	*NBN*
Non-syndromic hearing loss	*ACTG1, MYH9, PNPT1, PRPS1, SIX1, SIX2, TBC1D24, TJP2*
Pancreatic agenesis	*PDX1, PTF1A*
Phosphoglycerate dehydrogenase deficiency	*PHGDH*
Phosphoserine aminotransferase deficiency	*PSAT1*
Pitt-Hopkins syndrome	*NRXN1, TCF4*
Pontocerebellar hypoplasia	*AMPD2, CHMP1A, CLP1, EXOSC3, RARS2, SEPSECS, TSEN2, TSEN34, TSEN54, VPS53, VRK1*

Microcephaly (cont.)

Disorder	Associated gene(s)
Primary microcephaly	*ASPM, CASC5, CDK5RAP2, CDK6, CENPJ, CEP135, CEP152, MCPH1, STIL, WDR62, ZNF335*
Progressive microcephaly with seizures and brain atrophy	*MED17*
Renpenning syndrome	*PQBP1*
Rett syndrome	*MECP2*
Rigidity and multifocal seizure syndrome	*BRAT1*
Rubenstein-Taybi syndrome	*CREBBP*
Seckel syndrome	*ATR, CENPJ, CEP63, NIN, RBBP8*
Smith-Lemli-Opitz syndrome	*DHCR7*
Spastic paraplegia	*AP4B1, AP4E1, AP4M1, AP4S1, C12orf65, ERLIN2, HSPD1, IBA57, KIF1A, KIF5A, PLP1, TECPR2*
Syndromic mental retardation	*ADAT3, ATP6AP2, CLIC2, CTCF, CTNNB1, CUL4B, DLG3, DYNC1H1, DYRK1A, FOXP1, GATAD2B, GRIN1, HUWE1, KDM5C, KIAA2022, KIF1A, KPTN, MAN1B1, MBD5, NSUN2, PHF8, PURA, SETBP1, SETD5, SOX11, SYNGAP1, TRAPPC9, TUSC3, UPF3B, USP9X*
Thiamine metabolism dysfunction syndrome	*SLC19A3, SLC25A19*
Warburg micro syndrome	*RAB18, RAB3GAP1, RAB3GAP2, TBC1D20*
Warsaw breakage syndrome	*DDX11*
Xeroderma pigmentosum	*ERCC2, ERCC3, ERCC4, ERCC5, XPA, XPC*

Neurodegenerative

17-beta-hydroxyster dehydrogenase X deficiency	*HSD17B10*
3-hydroxyisobutryl-C hydrolase deficiency	*HIBCH, HMGCL*
Acute necrotizing encephalopathy	*RANBP2*
Aicardi-Goutieres syndrome	*ADAR, IFIH1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1*
Alexander disease	*GFAP*
Alternating hemiplegia of childhood	*ATP1A3*
Amyotrophic lateral sclerosis	*FIG4*
Andermann syndrome	*SLC12A6*
Asparagine synthetase deficiency	*ASNS*
Ataxia-telangiectasia-like disorder	*MRE11A*
Cerebellar hypoplasia and mental retardation syndrome	*VLDLR*
Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome	*SNAP29*
Cerebrooculofacioskeletal syndrome	*ERCC1, ERCC5*
Charcot-Marie-Tooth disease	*DNM2, EGR2, FGD4, GARS, GDAP1, IGHMBP2, MPZ, NAGLU, PMP22, PRPS1, PRX, SURF1*
Cockayne syndrome	*ERCC6, ERCC6, ERCC8*
Congenital cataracts, hearing loss, and neurodegeneration	*CCS, SLC33A1*

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Neurodegenerative (cont.)

Disorder	Associated gene(s)
Congenital variant Rett syndrome	*FOXG1*
Epileptic encephalopathy	*ALG13, ARHGEF9, ARX, CDKL5, CHD2, CLCN4, DNM1, DOCK7, EEF1A2, GABRA1, GNAO1, GRIN2B, HCN1, KCNA2, KCNB1, KCNH5, KCNQ2, KCNT1, MECP2, NECAP1, PCDH19, PIGQ, PLCB1, SCN2A, SCN8A, SIK1, SLC12A5, SLC13A5, SLC25A12, SLC25A22, SPTAN1, ST3GAL3, STXBP1, SZT2, TBC1D24, WWOX*
Familial encephalopathy with neuroserpin inclusion bodies	*SERPINI1*
FG syndrome	*CASK, FLNA*
GM2-gangliosidosis	*GM2A*
HARP syndrome	*PANK2*
Infantile cerebellar-retinal degeneration	*ACO2*
Infantile parkinsonism-dystonia	*SLC6A3*
Intellectual disability, microcephaly, and pontine cerebellar hypoplasia	*CASK*
Kosaki overgrowth syndrome	*PDGFRB*
L-arginine:glyci amidinotransferase deficiency	*GATM*
Leukoencephalopathy	*DARS2, RNASET2*
Leukoencephalopathy with vanishing white matter	*EIF2B4*
Mandibulofacial dysostosis	*EFTUD2*
Marinesco-Sjögren syndrome	*SIL1*
Muscular dystrophy-dystroglycanopathy	*DAG1, FKRP, FKTN, GMPPB, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, TMEM5, B3GALNT2, B4GAT1*
Nijmegen breakage syndrome	*NBN*
Optic atrophy	*ACO2, OPA1, OPA3, TMEM126A*
Pontocerebellar hypoplasia	*AMPD2, CHMP1A, CLP1, EXOSC3, RARS2, SEPSECS, TSEN2, TSEN34, TSEN54, VPS53, VRK1*
Progressive microcephaly with seizures and brain atrophy	*MED17*
Progressive myoclonic epilepsy	*EPM2A, KCTD7, NHLRC1, PRICKLE1*
Ritscher-Schinzel syndrome	*CCDC22, KIAA0196*
Sialic acid storage disorder	*SLC17A5*
Singleton-Merten syndrome	*IFIH1*
Spastic ataxia	*MARS2, MTPAP*
Spastic paraplegia	*AP4B1, AP4E1, AP4M1, AP4S1, C12orf65, ERLIN2, HSPD1, IBA57, KIF1A, KIF5A, PLP1, TECPR2*
Spinocerebellar ataxia	*ATP2B3, GRM1, ITPR1, SPTBN2*
Syndromic mental retardation	*ADAT3, ATP6AP2, CLIC2, CTCF, CTNNB1, CUL4B, DLG3, DYNC1H1, DYRK1A, FOXP1, GATAD2B, GRIN1, HUWE1, KDM5C, KIAA2022, KIF1A, KPTN, MAN1B1, MBD5, NSUN2, PHF8, PURA, SETBP1, SETD5, SOX11, SYNGAP1, TRAPPC9, TUSC3, UPF3B, USP9X*

Neurodegenerative (cont.)

Disorder	Associated gene(s)
Vici syndrome	*EPG5*
Xeroderma pigmentosum	*ERCC2, ERCC3, ERCC4, ERCC5, XPA, XPC*
Yunis-Varon Syndrome	*FIG4*

Neuropathy

Andermann syndrome	*SLC12A6*
Arts syndrome	*PRPS1*
Ataxia-oculomotor apraxia	*PNKP*
Ataxia-telangiectasia	*ATM*
Cerebral palsy	*KANK1*
Cerebral palsy, spastic quadriplegic	*GAD1*
Charcot-Marie-Tooth disease	*DNM2, EGR2, FGD4, GARS, GDAP1, IGHMBP2, MPZ, NAGLU, PMP22, PRPS1, PRX, SURF1*
Cockayne syndrome	*ERCC6, ERCC6, ERCC8*
D-bifunctional protein deficiency	*HSD17B4*
Dejerine-Sottas syndrome	*PRX*
Familial episodic pain syndrome	*SCN11A, TRPA1*
Familial hemiplegic migraine	*ATP1A2*
Hereditary sensory and autonomic neuropathy	*NGF, SCN11A, SPTLC1, WNK1*
Hereditary sensory neuropathy, spastic paraplegia	*CCT5*
Lesch-Nyhan syndrome	*HPRT1*
Lethal congenital contracture syndrome	*ADCY6, CNTNAP1, DNM2, ERBB3, GLE1, MYBPC1, PIP5K1C*
Marinesco-Sjögren syndrome	*SIL1*
Menkes disease	*ATP7A*
Microcephaly, seizures, and developmental delay	*PNKP*
Mucopolysaccharidosis, type III	*HGSNAT, NAGLU, SGSH*
Myoclonic dystonia	*SGCE*
Neurodegeneration with brain iron accululation	*WDR45*
Neuronaxonal dystrophy	*PLA2G6*
Non-syndromic hearing loss	*ACTG1, MYH9, PNPT1, PRPS1, SIX1, SIX2, TBC1D24, TJP2*
Paroxysmal nonkinesigenic dyskinesia	*PNKD*
Pelizaeus-Merzbacher disease	*PLP1*
Perrault syndrome	*CLPP, HARS2, HSD17B4, LARS2*
Phosphoribosylpyrophosphate synthetase superactivity	*PRPS1*
Segawa syndrome	*TH*
Severe orofacial dyspraxia resulting in largely incomprehensible speech	*FOXP2*
Spastic paraplegia	*AP4B1, AP4E1, AP4M1, AP4S1, C12orf65, ERLIN2, HSPD1, IBA57, KIF1A, KIF5A, PLP1, TECPR2*

Seizures

17-beta-hydroxyster dehydrogenase X deficiency	*HSD17B10*
2-aminoadipic 2-oxoadip aciduria	*DHTKD1*

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Seizures (cont.)

Disorder	Associated gene(s)
3-hydroxyisobutryl-C hydrolase deficiency	*HIBCH, HMGCL*
Acute necrotizing encephalopathy	*RANBP2*
Adenosine kinase deficiency	*ADK*
AICA-ribosiduria due to ATIC deficiency	*ATIC*
Alexander disease	*GFAP*
Alpha-thalassemia intellectual disability syndrome	*ATRX*
Aminoacylase deficiency	*ACY1*
Amish infantile epilepsy syndrome	*ST3GAL5*
Angelman syndrome	*UBE3A*
Ataxia-oculomotor apraxia	*PNKP*
Ayme-Gripp syndrome	*MAF*
Band-like calcification, simplified gyration, polymicrogyria	*OCLN*
Benign familial epilepsy	*KCNQ2, KCNQ3, PRRT2, SCN2A*
BH4-deficient hyperphenylalaninemia	*GCH1, PCBD1, QDPR*
Bilateral frontoparietal polymicrogyria	*ADGRG1*
Borjeson-Forssman-Lehmann syndrome	*PHF6*
Brachydactyly-mental retardation syndrome	*HDAC4*
Brugada syndrome	*SCN1B, SCN5A*
Canavan disease	*ASPA*
Carbamoylphosphate synthetase I deficiency	*CPS1*
Cerebral creatine deficiency syndrome	*SLC6A8*
Cerebral folate transport deficiency	*FOLR1*
CHILD syndrome	*NSDHL*
Christianson syndrome	*SLC9A6*
Ciliopathy	*CC2D2A, CEP290, IFT172, SDCCAG8*
CK syndrome	*NSDHL*
Cognitive impairment with or without cerebellar ataxia	*SCN8A*
Combined D-2- and L-2-hydroxyglutaric aciduria	*SLC25A1*
Complex cortical dysplasia	*KIF2A, KIF5C, TUBB2A, TUBB3, TUBG1*
Congenital fibrosis of extraocular muscles	*KIF21A, TUBB3*
Congenital muscular dystrophy	*CHKB, ITGA7, LAMA2, LMNA, POMT1, POMT2*
Congenital variant Rett syndrome	*FOXG1*
Cornelia de Lange syndrome	*HDAC8, NIPBL, SMC1A, SMC3*
D-glyceria aciduria	*GLYCTK*
Dihydropyrimidine dehydrogenase deficiency	*DPYD*
DOOR syndrome	*TBC1D24*
Dopa-responsive dystonia	*SPR*
Enlarged vestibular aqueduct	*SLC26A4*
Epilepsy, hearing loss, and mental retardation syndrome	*SPATA5*

Seizures (cont.)

Disorder	Associated gene(s)
Epileptic encephalopathy	*ALG13, ARHGEF9, ARX, CDKL5, CHD2, CLCN4, DNM1, DOCK7, EEF1A2, GABRA1, GNAO1, GRIN2B, HCN1, KCNA2, KCNB1, KCNH5, KCNQ2, KCNT1, MECP2, NECAP1, PCDH19, PIGQ, PLCB1, SCN2A, SCN8A, SIK1, SLC12A5, SLC13A5, SLC25A12, SLC25A22, SPTAN1, ST3GAL3, STXBP1, SZT2, TBC1D24, WWOX*
Epileptic encephalopathy, global delay, and cerebellar atrophy	*CACNA2D2*
Ethylmalonic encephalopathy	*ETHE1*
Familial cutaneous telangiectasia and cancer syndrome	*ATR*
Familial encephalopathy with neuroserpin inclusion bodies	*SERPINI1*
Familial epilepsy	*CHRNA7, DEPDC5, EFHC1, GABRB3, GABRD, GABRG2, KCND2, LGI1, SCN1A, SCN9A, STX1B, TNK2*
Familial myoclonic epilepsy	*TBC1D24*
Familial paroxysmal kinesigenic dyskinesia	*PRRT2*
Focal epilepsy, speech disorder, with or without mental retardation	*GRIN2A*
Generalized epilepsy and paroxysmal dyskinesia	*KCNMA1*
Glass syndrome	*SATB2*
Glucocorticoid deficiency	*MC2R, MRAP, NNT*
Glutathione synthetase deficiency	*GSS*
Glycosylphosphatidylinositol deficiency	*PIGM*
GM2-gangliosidosis	*GM2A*
Growth retardation, developmental delay, facial dysmorphism	*FTO*
Guanidinoacetate methyltransferase deficiency	*GAMT*
Hawkinsinuria	*HPD*
Holoprosencephaly	*CDON, DISP1, GLI2, SHH, SIX3, TGIF1, ZIC2*
Homocystinuria	*CBS, MTHFR*
Hyperphosphatasia with mental retardation syndrome	*PGAP3, PIGV*
Hypomagnesemia	*CLDN19, CNNM2, TRPM6*
Hypoparathyroidism-retardation-dysmorphism syndrome	*TBCE*
Hypophosphatasia mental retardation syndrome	*PIGO*
Infantile cerebellar-retinal degeneration	*ACO2*
Joubert syndrome	*CC2D2A, CEP290, CEP41, NPHP1, NPHP4*
Kaufman oculocerebrofacial syndrome	*UBE3B*
Kenny-Caffey syndrome	*TBCE*
Kleefstra syndrome	*EHMT1*
Kohlschutter-Tonz syndrome	*ROGDI*
L-2-hydroxyglutaric aciduria	*L2HGDH*

Seizures (cont.)

Disorder	Associated gene(s)
Leber congenital amaurosis	*AIPL1, CEP290, CRB1, CRX, GDF6, GUCY2D, IMPDH1, IQCB1, KCNJ13, LCA5, LRAT, NMNAT1, RPE65, RPGRIP1, TULP1*
Leukoencephalopathy	*DARS2, RNASET2*
Leukoencephalopathy with vanishing white matter	*EIF2B4*
Lissencephaly	*DCX, LAMB1, NDE1, PAFAH1B1, RELN, TUBA1A*
Lowe syndrome	*OCRL*
Macular dystrophy with central cone involvement	*MFSD8*
Meckel syndrome	*B9D1, CEP290, EXOC4, NPHP3, TMEM67*
Megalencephalic leukoencephalopathy	*HEPACAM, MLC1*
Megaloblastic anemia	*CUBN, DHFR*
Mental retardation with cerebellar hypoplasia and distinctive facial appearance	*OPHN1*
Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations	*MEF2C*
Microcephaly, epilepsy, and diabetes syndrome	*IER3IP1*
Microcephaly, progressive, with seizures and cerebral and cerebellar atrophy	*QARS*
Microcephaly, seizures, and developmental delay	*PNKP*
Microcephaly, short stature, and polymicrogyria with seizures	*RTTN*
Multiple congenital anomalies-hypotonia-seizure syndrome	*PIGA, PIGN, PIGT*
Myoclonic-atonic epilepsy	*SLC6A1*
Neonatal diabetes with cerebellar agenesis	*PTF1A*
Nephrogenic diabetes insipidus	*AQP2*
Neuronal ceroid lipofuscinosis	*CLN3, CLN5, CLN6, CLN8, CTSD, MFSD8, PPT1, TPP1*
Nocturnal frontal lobe epilepsy	*CHRNA4*
Non-ketotic hyperglycinemia	*AMT, GCSH, GLDC*
Non-syndromic hearing loss	*ACTG1, MYH9, PNPT1, PRPS1, SIX1, SIX2, TBC1D24, TJP2*
Nonspecific cardiac conduction defect	*SCN1B*
Optic atrophy	*ACO2, OPA1, OPA3, TMEM126A*
Pancreatic agenesis	*PDX1, PTF1A*
Parkinsonism with spasticity	*ATP6AP2*
Periventricular heterotopia	*ARFGEF2*
Periventricular nodular heterotopia	*ERMARD*
Pitt-Hopkins syndrome	*NRXN1, TCF4*
Pitt-Hopkins-like syndrome	*CNTNAP2*
Polymicrogyria	*TUBA8, TUBB2B*
Pontocerebellar hypoplasia	*AMPD2, CHMP1A, CLP1, EXOSC3, RARS2, SEPSECS, TSEN2, TSEN34, TSEN54, VPS53, VRK1*
Primary aldosteronism, seizures, and neurologic abnormalities	*CACNA1D*
Primary microcephaly	*ASPM, CASC5, CDK5RAP2, CDK6, CENPJ, CEP135, CEP152, MCPH1, STIL, WDR62, ZNF335*

Seizures (cont.)

Disorder	Associated gene(s)
Progressive myoclonic epilepsy	*EPM2A, KCTD7, NHLRC1, PRICKLE1*
Progressive neonatal encephalopathy	*ADSL*
Pseudo-neonatal adrenoleukodystrophy	*ACOX1*
Psychomotor retardation, epilepsy, and craniofacial dysmorphism	*SNIP1*
Pyridoxine-dependent epilepsy	*ALDH7A1*
Recurrent infections with encephalopathy, hepatic dysfunction, and cardiovasuclar malformations	*FADD*
Rett syndrome	*MECP2*
Rigidity and multifocal seizure syndrome	*BRAT1*
Rolandic epilepsy, mental retardation, and speech dyspraxia	*SRPX2*
Schizencephaly	*EML1, EMX2*
Seckel syndrome	*ATR, CENPJ, CEP63, NIN, RBBP8*
Senior-Loken syndrome	*CEP290, IQCB1, NPHP1, NPHP4, SDCCAG8, WDR19*
SESAME syndrome	*KCNJ10*
Sinoatrial node dysfunction and deafness	*CACNA1D*
Smith-Kingsmore syndrome	*MTOR*
Spastic paraplegia	*AP4B1, AP4E1, AP4M1, AP4S1, C12orf65, ERLIN2, HSPD1, IBA57, KIF1A, KIF5A, PLP1, TECPR2*
Succinic semialdehyde dehydrogenase deficiency	*ALDH5A1*
Sulfite oxidase deficiency	*SUOX*
Syndromic mental retardation	*ADAT3, ATP6AP2, CLIC2, CTCF, CTNNB1, CUL4B, DLG3, DYNC1H1, DYRK1A, FOXP1, GATAD2B, GRIN1, HUWE1, KDM5C, KIAA2022, KIF1A, KPTN, MAN1B1, MBD5, NSUN2, PHF8, PURA, SETBP1, SETD5, SOX11, SYNGAP1, TRAPPC9, TUSC3, UPF3B, USP9X*
Thiamine metabolism dysfunction syndrome	*SLC19A3, SLC25A19*
Tuberous sclerosis	*TSC1, TSC2*
Tyrosinemia, type III	*HPD, TAT*
Velocardiofacial syndrome	*TBX1*

Ear

Hearing loss

3MC syndrome	*MASP1, MASP1*
Alpha-mannosidosis	*MAN2B1*
Alstrom syndrome	*ALMS1*
Bartter syndrome	*BSND, CLCNKB, SLC12A1*
Biotinidase deficiency	*BTD*
Brachiootic syndrome	*SIX1*
Branchiootorenal spectrum disorders	*EYA1, SIX5*
Brown-Vialetto-Van Laere syndrome	*SLC52A2, SLC52A3*
Central hypoventilation syndrome	*ASCL1, EDN3, GDNF, PHOX2B, RET*
CHARGE syndrome	*CHD7, SEMA3E*

Hearing loss (cont.)

Disorder	Associated gene(s)
Chudley-McCullough syndrome	*GPSM2*
Coffin-Lowry syndrome	*RPS6KA3*
Combined mitochondrial complex deficiency	*GFER*
Combined oxidative phosphorylation deficiency	*AARS2, AIFM1, ATP5A1, C12orf65, EARS2, ELAC2, FARS2, GFM1, GTPBP3, LYRM4, MARS2, MRPL3, MRPL44, MRPS16, MRPS22, MTFMT, MTO1, NARS2, PNPT1, RMND1, SFXN4, SURF1, TARS2, TSFM, TUFM, VARS2*
Combined pituitary hormone deficiency	*LHX3*
Congenital cataracts, hearing loss, and neurodegeneration	*CCS, SLC33A1*
Congenital deafness with labyrinthine aplasia microtia and microdontia	*FGF3*
Craniofacial-deafness-hand syndrome	*PAX3*
D-bifunctional protein deficiency	*HSD17B4*
Donnai-Barrow syndrome	*LRP2*
Duane-radial ray syndrome	*SALL4*
Enlarged vestibular aqueduct	*SLC26A4*
Epidermolysis bullosa	*CD151, COL17A1, COL7A1, DSP, DST, EXPH5, ITGA6, ITGB4, KRT14, KRT5, LAMA3, LAMB3, LAMC2, PLEC*
Epilepsy, hearing loss, and mental retardation syndrome	*SPATA5*
Epstein syndrome	*MYH9*
Fechtner syndrome	*MYH9*
FG syndrome	*CASK, FLNA*
Fibrochondrogenesis	*COL11A1*
Frontometaphyseal dysplasia	*FLNA*
Growth retardation with deafness and mental retardation due to IGF1 deficiency	*IGF1*
Hypoparathyroidism, sensorineural deafness, and renal disease	*GATA3*
Jervell and Lange-Nielsen syndrome	*KCNE1, KCNQ1*
Keratitis-ichthyosis-deafness syndrome	*GJB2*
Leigh syndrome, mitochondrial complex III deficiency	*BCS1L*
LEOPARD syndrome	*BRAF, RAF1*
Macrothrombocytopenia and progressive sensorineural deafness	*MYH9*
Marshall syndrome	*COL11A1*
May-Hegglin anomaly	*MYH9*
Nephropathy with pretibial epidermolysis bullosa and deafness	*CD151*
Non-syndromic hearing loss	*ACTG1, MYH9, PNPT1, PRPS1, SIX1, SIX2, TBC1D24, TJP2*
Noonan syndrome	*A2ML1, BRAF, KRAS, LZTR1, NRAS, PTPN11, RAF1, RASA2, RIT1, RRAS, SOS1, SOS2*
Otospondylomegaepiphyseal dysplasia	*COL11A2*
Pendred syndrome	*FOXI1, SLC26A4*
Perrault syndrome	*CLPP, HARS2, HSD17B4, LARS2*
Piebaldism	*SNAI2*
Primary aldosteronism, seizures, and neurologic abnormalities	*CACNA1D*

Hearing loss (cont.)

Disorder	Associated gene(s)
Romano-Ward syndrome	*KCNE1*
Sebastian syndrome	*MYH9*
Sensorineural deafness with mild renal dysfunction	*BSND*
SESAME syndrome	*KCNJ10*
Sinoatrial node dysfunction and deafness	*CACNA1D*
Split-hand/foot malformation	*DLX5, FBXW4*
Split-hand/foot malformation with sensorineural hearing loss	*DLX5*
Stickler syndrome	*COL11A1, COL2A1, COL9A1*
Temtamy preaxial brachydactyly syndrome	*CHSY1*
Thiamine-responsive megaloblastic anemia syndrome	*SLC19A2*
Tietz albinism-deafness syndrome	*MITF*
Townes-Brocks syndrome	*SALL1*
Waardenburg syndrome	*EDN3, EDNRB, MITF, SNAI2, SOX10*
Weissenbacher-Zweymuller syndrome	*COL11A2*
Wolfram syndrome	*WFS1*
Wolfram-like syndrome	*WFS1*

Structural abnormalities

Auriculocondylar syndrome	*GNAI3, PLCB4*
Brachiootic syndrome	*SIX1*
Branchiootorenal spectrum disorders	*EYA1, SIX5*
Congenital deafness with labyrinthine aplasia microtia and microdontia	*FGF3*
Distal renal tubular acidosis with progressive nerve deafness	*ATP6V1B1*
Enlarged vestibular aqueduct	*SLC26A4*
Noonan syndrome	*A2ML1, BRAF, KRAS, LZTR1, NRAS, PTPN11, RAF1, RASA2, RIT1, RRAS, SOS1, SOS2*
Pendred syndrome	*FOXI1, SLC26A4*
Townes-Brocks syndrome	*SALL1*

Cardiac

Aneurysm, other

Arrhythmogenic right ventricular dysplasia	*PKP2, TMEM43*
Bannayan-Riley-Ruvalcaba syndrome	*PTEN*
Coronary artery disease	*MEF2A*
Cowden syndrome	*PTEN, SDHB*
Generalized arterial calcification of infancy	*ENPP1*
Loeys-Dietz syndrome	*SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2*
Marfan syndrome	*FBN1*
Proteus syndrome	*PTEN*
PTEN-related cardiovascular disease	*PTEN*
Thoracic aortic aneurysms and aortic dissections	*MYH11*
Tuberous sclerosis	*TSC1, TSC2*

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Cardiomyopathy

Disorder	Associated gene(s)
3-methylglutaconic aciduria	*CLPB, DNAJC19, OPA3*
Adult polyglucosan body disease	*GBE1*
Alstrom syndrome	*ALMS1*
Arrhythmogenic right ventricular dysplasia	*PKP2, TMEM43*
Atrial septal defect	*CITED2, MYH6, TBX20*
Barth syndrome	*TAZ*
Beta-ketothiolase deficiency	*ACAT1*
Cardiac conduction defect, dilated cardiomyopathy	*TNNI3K*
Cardioencephalomyopathy	*SCO2*
Cardiofaciocutaneous syndrome	*BRAF, KRAS, MAP2K1*
Cardiomyopathy	*ACTC1, ACTN2, ANKRD1*
Carnitine palmitoyltransferase deficiency, type II	*CPT2*
Cataracts	*AGK, CRYAA, CRYBA1, CRYGC, CRYGD, EPHA2, NHS*
Central core disease	*RYR1*
Congenital muscular dystrophy	*CHKB, ITGA7, LAMA2, LMNA, POMT1, POMT2*
Congenital neuromuscular disease with uniform type 1 fiber	*RYR1*
Dilated cardiomyopathy	*MYH6, SCN5A*
Distal cardiomyopathy	*MYH7*
Glutaric acidemia, type II	*ETFA, ETFB, ETFDH*
Glycogen storage disease	*AGL, ALDOA, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LDHA, PFKM, PHKA2, PHKG2, PRKAG2, PYGL, SLC37A4*
Hypertrophic cardiomyopathy	*MYH6, MYH7*
King-Denborough syndrome	*RYR1*
Left ventricular non-compaction cardiomyopathy	*ACTC1, DTNA, MIB1, MYH7B*
LEOPARD syndrome	*BRAF, RAF1*
Liang distal myopathy	*MYH7*
Limb-girdle muscular dystrophy	*DAG1, DYSF, PLEC, TCAP, TRIM32*
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	*HADHA*
Lymphedema-distichiasis syndrome	*FOXC2*
Malignant hyperthermia susceptibility	*RYR1*
Minicore myopathy	*RYR1*
Miyoshi muscular dystrophy	*DYSF*
Myofibrillar myopathy	*CRYAB*
Myopathy, lactic acidosis, and sideroblastic anemia	*YARS2, YARS2*
Myosin storage myopathy	*MYH7*
Noonan syndrome	*A2ML1, BRAF, KRAS, LZTR1, NRAS, PTPN11, RAF1, RASA2, RIT1, RRAS, SOS1, SOS2*
Noonan-like syndrome with loose anagen hair	*SHOC2*
Peroxisome biogenesis disorder	*PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7*
Propionic acidemia	*PCCA, PCCB*
Refsum disease	*PHYH*
Rhizomelic chondrodysplasia	*PEX7*

Cardiomyopathy (cont.)

Disorder	Associated gene(s)
Sengers syndrome	*AGK*
Systemic primary carnitine deficiency	*SLC22A5*
Trifunctional protein deficiency	*HADHA, HADHB*
Very long chain acyl-coenzyme A dehydrogenase deficiency	*ACADVL*
Vici syndrome	*EPG5*
Wolff-Parkinson-White syndrome	*PRKAG2*
Wolfram syndrome	*WFS1*
Wolfram-like syndrome	*WFS1*

Conduction defects

Acro-renal-ocular syndrome	*SALL4*
Andersen syndrome	*KCNJ2*
Arrhythmogenic right ventricular dysplasia	*PKP2, TMEM43*
Atrial septal defect	*CITED2, MYH6, TBX20*
Beta-ketothiolase deficiency	*ACAT1*
Brugada syndrome	*SCN1B, SCN5A*
Cardiac conduction defect	*CALM3*
Cardiac conduction defect, dilated cardiomyopathy	*TNNI3K*
Cardiofaciocutaneous syndrome	*BRAF, KRAS, MAP2K1*
Congenital muscular dystrophy	*CHKB, ITGA7, LAMA2, LMNA, POMT1, POMT2*
Dilated cardiomyopathy	*MYH6, SCN5A*
Duane-radial ray syndrome	*SALL4*
Familial atrial fibrillation	*SCN4B, SCN5A*
Familial epilepsy	*CHRNA7, DEPDC5, EFHC1, GABRB3, GABRD, GABRG2, KCND2, LGI1, SCN1A, SCN9A, STX1B, TNK2*
Generalized congenital lipodystrophy	*PTRF*
Glycogen storage disease	*AGL, ALDOA, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LDHA, PFKM, PHKA2, PHKG2, PRKAG2, PYGL, SLC37A4*
Holt-Oram syndrome	*SALL4, TBX5*
Hypertrophic cardiomyopathy	*MYH6, MYH7*
Jervell and Lange-Nielsen syndrome	*KCNE1, KCNQ1*
Long QT syndrome	*KCNE2, KCNH2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1*
Muscular dystrophy-dystroglycanopathy	*DAG1, FKRP, FKTN, GMPPB, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, TMEM5, B3GALNT2, B4GAT1*
Nonprogressive heart block	*SCN5A*
Noonan syndrome	*A2ML1, BRAF, KRAS, LZTR1, NRAS, PTPN11, RAF1, RASA2, RIT1, RRAS, SOS1, SOS2*
Peroxisome biogenesis disorder	*PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7*
Refsum disease	*PHYH*
Rhizomelic chondrodysplasia	*PEX7*
Right atrial isomerism	*GDF1*
Romano-Ward syndrome	*KCNE1*

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Conduction defects (cont.)

Disorder	Associated gene(s)
Sick sinus syndrome	*HCN4, SCN5A*
Smith-Lemli-Opitz syndrome	*DHCR7*
Tetralogy of Fallot	*GDF1, ZFPM2*
Transposition of the great arteries, dextro-looped	*GDF1, MED13L*
Ventricular septal defect	*CITED2, IRX4*
Wolff-Parkinson-White syndrome	*PRKAG2*
Wolfram syndrome	*WFS1*
Wolfram-like syndrome	*WFS1*

Structural defects

Aarskog-Scott syndrome	*FGD1*
Acro-renal-ocular syndrome	*SALL4*
Adams-Oliver syndrome	*ARHGAP31, DOCK6, EOGT, NOTCH1*
Alagille syndrome	*JAG1, NOTCH2*
Antley-Bixler syndrome	*POR*
Aortic valve disease	*NOTCH1, SMAD6*
Atrial septal defect	*CITED2, MYH6, TBX20*
Atrioventricular septal defect	*CRELD1, GJA1*
Beckwith-Wiedemann syndrome	*CDKN1C*
Cantu syndrome	*ABCC9, KCNJ8*
Cardiofaciocutaneous syndrome	*BRAF, KRAS, MAP2K1*
Carpenter syndrome	*MEGF8, RAB23*
Char syndrome	*TFAP2B*
CHARGE syndrome	*CHD7, SEMA3E*
CHIME syndrome	*PIGL*
Congenital cardiac defects	*FOXH1, GATA4, GATA6, HAND2, NKX2-5, NR2F2, TAB2, ZIC3*
Congenital heart defects, hamartomas, and polysyndactyly	*WDPCP*
Congenital left heart obstructive defects	*MCTP2*
Congenital severe neutropenia	*G6PC3*
Conotruncal heart malformations	*NKX2-6*
Cornelia de Lange syndrome	*HDAC8, NIPBL, SMC1A, SMC3*
Dilated cardiomyopathy	*MYH6, SCN5A*
Duane-radial ray syndrome	*SALL4*
FG syndrome	*CASK, FLNA*
Frontometaphyseal dysplasia	*FLNA*
Glycogen storage disease	*AGL, ALDOA, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LDHA, PFKM, PHKA2, PHKG2, PRKAG2, PYGL, SLC37A4*
Heterotaxy syndrome	*ACVR2B, CRELD1, NODAL, ZIC3*
Holt-Oram syndrome	*SALL4, TBX5*
Hypertrophic cardiomyopathy	*MYH6, MYH7*
Hypoplastic left heart syndrome	*GJA1*
Left-right axis malformations	*LEFTY2*
LEOPARD syndrome	*BRAF, RAF1*
Mucopolysaccharidosis type IV	*GLB1*
Multiple congenital anomalies	*MEIS2*
Noonan syndrome	*A2ML1, BRAF, KRAS, LZTR1, NRAS, PTPN11, RAF1, RASA2, RIT1, RRAS, SOS1, SOS2*

Structural defects (cont.)

Disorder	Associated gene(s)
Noonan-like syndrome with loose anagen hair	*SHOC2*
Oculodentodigital dysplasia	*GJA1*
Pancreatic agenesis and congenital heart defects	*GATA6*
Persistent truncus arteriosus	*NKX2-6*
Right atrial isomerism	*GDF1*
Ritscher-Schinzel syndrome	*CCDC22, KIAA0196*
Schinzel-Giedion midface retraction syndrome	*SETBP1*
Short-rib thoracic dysplasia	*DYNC2H1, IFT140, IFT80, NEK1, TTC21B, WDR19, WDR35*
Sotos syndrome	*NFIX, NSD1*
Syndactyly	*GJA1*
TARP syndrome	*RBM10*
Tetralogy of Fallot	*GDF1, ZFPM2*
Townes-Brocks syndrome	*SALL1*
Transaldolase deficiency	*TALDO1*
Transposition of the great arteries, dextro-looped	*GDF1, MED13L*
VACTERL association	*ZIC3*
Velocardiofacial syndrome	*TBX1*
Ventricular septal defect	*CITED2, IRX4*
Wolff-Parkinson-White syndrome	*PRKAG2*
Wolfram syndrome	*WFS1*
Wolfram-like syndrome	*WFS1*

Valve disease

Adams-Oliver syndrome	*ARHGAP31, DOCK6, EOGT, NOTCH1*
Aortic valve disease	*NOTCH1, SMAD6*
Atrioventricular septal defect	*CRELD1, GJA1*
Cardiofaciocutaneous syndrome	*BRAF, KRAS, MAP2K1*
Gaucher disease	*GBA*
Heterotaxy syndrome	*ACVR2B, CRELD1, NODAL, ZIC3*
Hypoplastic left heart syndrome	*GJA1*
Left-right axis malformations	*LEFTY2*
Loeys-Dietz syndrome	*SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2*
Neurofibromatosis, type 1	*NF1*
Noonan syndrome	*A2ML1, BRAF, KRAS, LZTR1, NRAS, PTPN11, RAF1, RASA2, RIT1, RRAS, SOS1, SOS2*
Noonan-like syndrome with loose anagen hair	*SHOC2*
Oculodentodigital dysplasia	*GJA1*
Tuberous sclerosis	*TSC1, TSC2*

Vascular abnormalities

Adams-Oliver syndrome	*ARHGAP31, DOCK6, EOGT, NOTCH1*
CHARGE syndrome	*CHD7, SEMA3E*
Ventricular septal defect	*CITED2, IRX4*
Atrial septal defect	*CITED2, MYH6, TBX20*
Fabry disease	*GLA*
Dysautonomia	*IKBKAP*

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Vascular abnormalities (cont.)

Disorder	Associated gene(s)
Alagille syndrome	*JAG1, NOTCH2*
Hypertrophic cardiomyopathy	*MYH6, MYH7*
Dilated cardiomyopathy	*MYH6, SCN5A*
Neurofibromatosis, type 1	*NF1*
Essential hypertension	*NOS3, PDE5A*
Aortic valve disease	*NOTCH1, SMAD6*
Coffin-Lowry syndrome	*RPS6KA3*
Arterial tortuosity syndrome	*SLC2A10*
Microcephaly-capillary malformation syndrome	*STAMBP*

Cancer

Cardiac/pulmonary

Leukemia, Sertoli-Leydig cell tumor, pleuropulmonary blastoma	*DICER1, PBX1, RUNX1*
Rubenstein-Taybi syndrome	*CREBBP*
Tuberous sclerosis	*TSC1, TSC2*

Gastrointestinal/throat

Common variable immunodeficiency	*CD19, CD81, ICOS, IL21, MS4A1, NFKB1, TNFRSF13C*
Cowden syndrome	*PTEN, SDHB*
Familial cutaneous telangiectasia and cancer syndrome	*ATR*
Gastric stromal sarcoma	*SDHB*
Intestinal carcinoid tumors	*SDHD*
Mitochondrial complex II deficiency	*SDHAF1, SDHD*
Paraganglioma	*SDHB, SDHD*
Paraganglioma with or without deafness	*SDHD*
Pheochromocytoma	*RET, SDHB, SDHD*
Renal cell carcinoma	*SDHB*
Seckel syndrome	*ATR, CENPJ, CEP63, NIN, RBBP8*
Sotos syndrome	*NFIX, NSD1*
Genitourinary	
Beckwith-Wiedemann syndrome	*CDKN1C*
Cowden syndrome	*PTEN, SDHB*
Gastric stromal sarcoma	*SDHB*
Glycogen storage disease	*AGL, ALDOA, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LDHA, PFKM, PHKA2, PHKG2, PRKAG2, PYGL, SLC37A4*
Paraganglioma	*SDHB, SDHD*
Perlman Syndrome	*DIS3L2*
Pheochromocytoma	*RET, SDHB, SDHD*
Renal cell carcinoma	*SDHB*
Simpson-Golabi-Behmel syndrome	*GPC3*
Tuberous sclerosis	*TSC1, TSC2*
Wilms tumor-aniridia-genital anomalies-retardation syndrome	*WT1*

Hematologic

Disorder	Associated gene(s)
Ataxia-telangiectasia	*ATM*
Cardiofaciocutaneous syndrome	*BRAF, KRAS, MAP2K1*
Common variable immunodeficiency	*CD19, CD81, ICOS, IL21, MS4A1, NFKB1, TNFRSF13C*
Dyskeratosis congenita	*DKC1, RTEL1, TERT, TINF2*
Fanconi anemia	*BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, RAD51C, SLX4, UBE2T, XRCC2*
Hyper IgE syndrome	*STAT3*
Infantile-onset multisystem autoimmune disease	*STAT3, ZAP70*
LEOPARD syndrome	*BRAF, RAF1*
Leukemia, acute	*DICER1, PBX1, RUNX1*
Multinodular goiter, Sertoli-Leydig cell tumors	*DICER1*
Noonan syndrome	*A2ML1, BRAF, KRAS, LZTR1, NRAS, PTPN11, RAF1, RASA2, RIT1, RRAS, SOS1, SOS2*
Pleuropulmonary blastoma, neuroendocrine tumors	*DICER1*
Rubenstein-Taybi syndrome	*CREBBP*
Sotos syndrome	*NFIX, NSD1*

Neuroendocrine/CNS

Carney complex	*MYH8, PRKAR1A*
Central hypoventilation syndrome	*ASCL1, EDN3, GDNF, PHOX2B, RET*
Costello syndrome	*HRAS*
Cowden syndrome	*PTEN, SDHB*
Gastric stromal sarcoma	*SDHB*
Hirschsprung disease	*RET*
Leukemia, acute	*DICER1, PBX1, RUNX1*
Medullary thyroid carcinoma	*RET*
Multinodular goiter, Sertoli-Leydig cell tumors	*DICER1*
Multiple endocrine neoplasia, type 2	*RET*
Neuroblastic tumor susceptibility	*ALK*
Neurofibromatosis, type 1	*NF1*
Paraganglioma	*SDHB, SDHD*
Pheochromocytoma	*RET, SDHB, SDHD*
Pleuropulmonary blastoma, neuroendocrine tumors	*DICER1*
Renal agenesis	*RET*
Renal cell carcinoma	*SDHB*
Rubenstein-Taybi syndrome	*CREBBP*
Serum retinoic acid, neuroblastoma	*ALDH1A2*
Tuberous sclerosis	*TSC1, TSC2*

Non-malignant tumors

Bannayan-Riley-Ruvalcaba syndrome	*PTEN*
Basal cell nevus syndrome	*PTCH1, PTCH2*
Costello syndrome	*HRAS*
Cowden syndrome	*PTEN, SDHB*

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Non-malignant tumors (cont.)

Disorder	Associated gene(s)
Leukemia, acute	*DICER1, PBX1, RUNX1*
Multinodular goiter, Sertoli-Leydig cell tumors	*DICER1*
Neurofibromatosis, type 1	*NF1*
Pleuropulmonary blastoma, neuroendocrine tumors	*DICER1*
Proteus syndrome	*PTEN*
PTEN-related cardiovascular disease	*PTEN*
Other malignancies	
Bannayan-Riley-Ruvalcaba syndrome	*PTEN*
Bloom syndrome	*BLM*
Cardiofaciocutaneous syndrome	*BRAF, KRAS, MAP2K1*
Cowden syndrome	*PTEN, SDHB*
Fanconi anemia	*BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, RAD51C, SLX4, UBE2T, XRCC2*
LEOPARD syndrome	*BRAF, RAF1*
Nijmegen breakage syndrome	*NBN*
Noonan syndrome	*A2ML1, BRAF, KRAS, LZTR1, NRAS, PTPN11, RAF1, RASA2, RIT1, RRAS, SOS1, SOS2*
Proteus syndrome	*PTEN*
PTEN-related cardiovascular disease	*PTEN*

Skin/bone

Basal cell nevus syndrome	*PTCH1, PTCH2*
Cardiofaciocutaneous syndrome	*BRAF, KRAS, MAP2K1*
LEOPARD syndrome	*BRAF, RAF1*
Noonan syndrome	*A2ML1, BRAF, KRAS, LZTR1, NRAS, PTPN11, RAF1, RASA2, RIT1, RRAS, SOS1, SOS2*
Rothmund-Thomson syndrome	*RECQL4*
Xeroderma pigmentosum	*ERCC2, ERCC3, ERCC4, ERCC5, XPA, XPC*

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