Ambry Genetics®

EXPANDED SECONDARY FINDINGS GENE LIST

SUPPLEMENTAL TEST FORM December 2016

GENE	DISORDER(S)	CHILDHOOD ONSET	ADULT ONSET	CANCER PREDISPOSITION	CARRIER	ACMG*
AAAS	Achalasia-addisonianism-alacrimia syndrome	X			X	
AASS	Hyperlysinemia; Saccharopinuria	X			X	
ABCA1	Tangier disease; HDL deficiency, type 2	X			X	
ABCA12	Ichthyosis, lamellar 2; Ichthyosis, harlequin	X			X	
ABCA3	Surfactant metabolism dysfunction, pulmonary, 3	X			X	
ABCA4	Stargardt disease-1; Retinitis pigmentosa-19; Cone-rod dystrophy 3; Macular degeneration, age-related, 2; Fundus flavimaculatus; Retinal dystrophy, early-onset severe	X	X		X	
ABCB11	Cholestasis, progressive familial intrahepatic; Cholestasis, benign recurrent intrahepatic	X	X		X	
ABCB4	Cholestasis, progressive familial intrahepatic; Cholestasis, familial intrahepatic, of pregnancy; Gallbladder disease	X	X		X	
ABCB7	Anemia, sideroblastic, with ataxia	X			X	
ABCC2	Dubin-Johnson syndrome	X			X	
ABCC6	Pseudoxanthoma elasticum; Pseudoxanthoma elasticum, forme fruste	X			X	
ABCC8	Familial hyperinsulinemic hypoglycemia; Hypoglycemia of infancy, leucine-sensitive; Diabetes mellitus	X			X	
ABCC8	Familial hyperinsulinemic hypoglycemia; Hypoglycemia of infancy, leucine-sensitive; Diabetes mellitus	X	X		X	
ABCC9	Cardiomyopathy, dilated, 1O		X			
ABCD1	Adrenoleukodystrophy; Adrenomyeloneuropathy,	X			X	
ABCG5	Sitosterolemia	X			X	
ABCG8	Sitosterolemia; Gallbladder disease 4	X			X	
ABHD5	Chanarin-Dorfman syndrome	X			X	
ACAD8	Isobutyryl-CoA dehydrogenase deficiency	X			X	
ACAD9	ACAD9 deficiency	X			X	
ACADM	Acyl-CoA dehydrogenase deficiency, medium chain	X			X	
ACADS	Acyl-CoA dehydrogenase deficiency, short-chain	X			X	
ACADSB	2-methylbutyrylglycinuria	X			X	
ACADVL	VLCAD deficiency	X			X	
ACAN	Spondyloepiphyseal dysplasia, Kimberley type; Spondyloepimetaphyseal dysplasia, aggrecan type; Osteochondritis dissecans, short stature, and early-onset osteoarthritis	X			X	
ACAT1	Alpha-methylacetoacetic aciduria	X			X	
ACE	Renal tubular dysgenesis	X			X	
ACOX1	Peroxisomal acyl-CoA oxidase deficiency	X			X	
ACSL4	Mental retardation, X-linked nonspecific, 63	X			X	
ACTA1	Myopathy, nemaline, 3; Myopathy, actin, congenital, with excess of thin myofilaments; Myopathy, actin, congenital, with cores; Myopathy, congenital, with fiber-type disproportion 1	X			X	

*ACMG minimum list genes

GENE	DISORDER(S)	CHILDHOOD ONSET	ADULT ONSET	CANCER PREDISPOSITION	CARRIER	ACMG*
ACTA2	Marfan syndrome; Loeys-Dietz syndrome; and Familial thoracic aortic aneurysms and dissections	X	X			X
ACTB	Dystonia, juvenile-onset	X				
ACTC1	Familial hypertrophic cardiomyopathy; Dilated cardiomyopathy	X	X			X
ACTG1	Deafness, autosomal dominant 20/26	X				
ACVR1	Fibrodysplasia ossificans progressiva	X	X			
ACVRL1	Telangiectasia, hereditary hemorrhagic, type 2	X	X			
ADA	Severe combined immunodeficiency due to ADA deficiency; Adenosine deaminase deficiency, partial	X			X	
ADAMTS10	Weill-Marchesani syndrome, recessive	X			X	
ADAMTS13	Thrombotic thrombocytopenic purpura, familial	X			X	
ADAMTS2	Ehlers-Danlos syndrome, type VIIC	X			X	
ADAMTSL2	Geleophysic dysplasia	X			X	
ADCK3	Coenzyme Q10 deficiency); Spinocerebellar ataxia, autosomal recessive 9	X			X	
ADHR	Hypophosphatemic rickets, autosomal dominant	X	X			
AGA	Aspartylglucosaminuria	X			X	
AGL	Glycogen storage disease IIIa; Glycogen storage disease IIIb; Glycogen Storage Disease Type III	X	X		X	
AGPAT2	Lipodystrophy, congenital generalized, type 1	X			X	
AGPS	Rhizomelic chondrodysplasia punctata, type 3	X			X	
AGS2	Alagille syndrome 2; Aicardi-Goutieres syndrome 2	X			X	
AGT	Renal tubular dysgenesis	X			X	
AGTR1	Hypertension, essential; Renal tubular dysgenesis	X			X	
AGTR2	Mental retardation, X-linked-88	X			X	
AGXT	Hyperoxaluria, primary, type 1	X	X		X	
AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteinehydrolase	X			X	
AHI1	Joubert syndrome-3	X			X	
AIP	Pituitary adenoma, growth hormone-secreting; Pituitary adenoma, prolactin-secreting; Pituitary adenoma, ACTH-secreting	X				
AIRE	Autoimmune polyendocrinopathy syndrome , type I, with or withoutreversible metaphyseal dysplasia	X			X	
ALAD	Porphyria, acute hepatic	X			X	
ALAS2	Anemia, sideroblastic, X-linked; Protoporphyria, erythropoietic, X-linked dominant	X			X	
ALDH3A2	Sjogren-Larsson syndrome	X			X	
ALDH4A1	Hyperprolinemia, type II	X			X	
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency	X			X	
ALDH7A1	Epilepsy, pyridoxine-dependent	X			X	
ALDOB	Fructose intolerance	X			X	
ALG12	Congenital disorder of glycosylation, type Ig	X			X	
ALG2	Congenital disorder of glycosylation, type Ii	X			X	
ALG3	Congenital disorder of glycosylation, type Id	X			X	
ALG6	Congenital disorder of glycosylation, type Ic	X			X	
ALG8	Congenital disorder of glycosylation, type Ih	X			X	

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ALG9	Congenital disorder of glycosylation, type II	X			X	
ALMS1	Alstrom syndrome	X	X		X	
ALOX12B	Ichthyosiform erythroderma, congenital, nonbullous, 1	X			X	
ALPL	Hypophosphatasia, infantile; Hypophosphatasia, childhood; Odontohypophosphatasia; Hypophosphatasia, adult	X	X		X	
ALS2	Amyotrophic lateral sclerosis, juvenile; Primary lateral sclerosis, juvenile; Spastic paralysis, infantile onset ascending	X			X	
ALX4	Parietal foramina 2; Frontonasal dysplasia 2	X			X	
AMACR	Alpha-methylacyl-CoA racemase deficiency; Bile acid synthesis defect, congenital, 4	X			X	
AMELX	Amelogenesis imperfecta, hypoplastic/hypomaturation type	X				
AMH	Persistent Mullerian duct syndrome, type I	X			X	
AMHR2	Persistent Mullerian duct syndrome, type II	X			X	
AMT	Glycine encephalopathy	X			X	
ANG	Amyotrophic lateral sclerosis 9		X			
ANH1	Anemia, sideroblastic, X-linked; Protoporphyria, erythropoietic, X-linked dominant		X		X	
ANK1	Spherocytosis, type 1	X			X	
ANK2	Long QT syndrome-4; Cardiac arrhythmia, ankyrin-B-related	X				
ANKH	Craniometaphyseal dysplasia; Chondrocalcinosis	X	X			
ANO5	Gnathodiaphyseal dysplasia; Muscular dystrophy, limb-girdle, type 2L; Miyoshi muscular dystrophy 3	x			X	
ANTXR2	Fibromatosis, juvenile hyaline; Hyalinosis, infantilesystemic	X			X	
AP1S2	Mental retardation, X-linked 59	X			X	
AP3B1	Hermansky-Pudlak syndrome 2	X			X	
APC	Familial adenomatous polyposis	X		X		X
APOA1	Hypoalphalipoproteinemia; Amyloidosis, 3 or more types		X			
APOB	Familial Hypercholesterolemia	X				X
APP	Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants; Alzheimer disease 1, familial		X			
APRT	Urolithiasis, 2,8-dihydroxyadenine	X			X	
APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia; Coenzyme Q10 deficiency	X			X	
AQP2	Diabetes insipidus, nephrogenic	X			X	
AR	Androgen insensitivity; Spinal and bulbar muscularatrophy of Kennedy; Androgen insensitivity, partial, with or without breast cancer; Hypospadias 1, X-linked	X	X		X	
ARFGEF2	Periventricular heterotopia with microcephaly	X			X	
ARG1	Argininemia	X			X	
ARHGEF6	Mental retardation, X-linked nonspecific, type 46	X			X	
ARHGEF9	Epileptic encephalopathy, early infantile, 8	X			X	
ARSA	Metachromatic leukodystrophy	X	X		X	
ARSB	Mucopolysaccharidosis type VI (Maroteaux-Lamy)	X			X	
ARSE	Chondrodysplasia punctata, X-linked recessive	X			X	

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ARX	Epileptic encephalopathy, early infantile, 1; Lissencephaly, X-linked 2; Mental retardation, X-linked 36/43/54; Proud syndrome; Partington syndrome; Hydranencephaly with abnormal genitalia	X			X	
ASAH1	Farber lipogranulomatosis	X			X	
ASL	Argininosuccinic aciduria	X			X	
ASPA	Canavan disease	X			X	
ASPM	Microcephaly, primary autosomal recessive, 5, with or without simplified gyral pattern	X			X	
ASS1	Citrullinemia	X	X		X	
ATL1	Spastic paraplegia-3A	X	X			
ATM	Ataxia-telangiectasia; Lymphoma, B-cell non-Hodgkin,somatic; Lymphoma, mantle cell; T-cell prolymphocytic leukemia, sporadic	X		X	X	
ATP1A2	Migraine, familial hemiplegic, 2; Alternating hemiplegia of childhood; Migraine, familial basilar	X	X			
ATP1A3	Dystonia-12; Alternating hemiplegia of childhood 2	X	X			
ATP2A1	Brody myopathy	X			X	
ATP2A2	Darier disease; Acrokeratosis verruciformis	X	X			
ATP2C1	Hailey-Hailey disease		X			
ATP6V0A2	Cutis laxa, autosomal recessive, type II; Wrinkly skin syndrome	X			X	
ATP7A	Menkes disease; Occipital horn syndrome; Spinal muscular atrophy, distal, X-linked 3	X	X		X	
ATP7B	Wilson disease	X	X		X	X
ATP8B1	Cholestasis, progressive familial intrahepatic 1; Cholestasis, benign recurrent intrahepatic	X	X		X	
ATR	Seckel syndrome 1	X			X	
ATRX	Alpha-thalassemia/mental retardation syndrome; Mental retardation-hypotonic facies syndrome, X-linked	X			X	
ATS	Arterial tortuosity syndrome	X			X	
AUH	3-methylglutaconic aciduria, type I	X			X	
AVP	Diabetes insipidus, neurohypophyseal	X				
AVPR2	Diabetes insipidus, nephrogenic; Nephrogenic syndrome of inappropriate antidiuresis	X			X	
B3GALTL	Peters-plus syndrome	X			X	
B4GALT1	Congenital disorder of glycosylation, type IId	X			X	
BBS1	Bardet-Biedl syndrome 1	X			X	
BBS10	Bardet-Biedl syndrome 10	X			X	
BCKDHA	Maple syrup urine disease, type Ia	X			X	
BCKDHB	Maple syrup urine disease, type Ib	X			X	
BCOR	Microphthalmia, syndromic 2	X				
BCS1L	Mitochondrial complex III deficiency; GRACILE syndrome; Leigh syndrome; Bjornstad syndrome	X			X	
BEST1	Best macular dystrophy; Maculopathy, bull's-eye; Vitelliform macular dystrophy, adult-onset; Bestrophinopathy; Vitreoretinochoroidopathy; Microcornea, rod-conedystrophy, cataract, and posterior staphyloma; Retinitis pigmentosa-50; Retinitis pigmentosa, concentric	X				
BIN1	Myopathy, centronuclear, autosomal recessive	X			X	

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BLM	Bloom syndrome	X			X	
BLOC1S6	Hermansky-pudlak syndrome 9	X			X	
BMPR1A	Polyposis, juvenile intestinal; Polyposis syndrome, hereditary mixed, 2; Juvenile polyposis syndrome, infantile form		X	X		X
BMPR2	Pulmonary hypertension, familial primary; Pulmonaryvenoocclusive disease; Pulmonary hypertension, primary, fenfluramine-associated	X				
BRAF	Cardiofaciocutaneous syndrome; Noonan syndrome 7; LEOPARD syndrome 3	X		X		
BRCA1	Hereditary breast and ovarian cancer		X			X
BRCA2	Hereditary breast and ovarian cancer		X			X
BRIP1	Breast cancer, early-onset; Fanconi anemia, complementation group J	X		X	X	
BRWD3	Mental retardation, X-linked 93	X			X	
BSCL2	Lipodystrophy, congenital generalized, type 2; Silverspastic paraplegia syndrome; Neuropathy, distal hereditary motor, type V	X			X	
BSCL2	Neuropathy, distal hereditary motor, type VA; Lipodystrophy, congenital generalized, type 2; Silver spastic paraplegia syndrome	X	X		X	
BSND	Bartter syndrome, type 4a; Sensorineural deafness with mild renal dysfunction	X			X	
BTD	Biotinidase deficiency	X			X	
BTK	Agammaglobulinemia, X-linked 1; Agammaglobulinemia and isolated hormone deficiency	X			X	
C10orf2	Progressive external ophthalmoplegia, autosomal dominant, 3; Mitochondrial DNA depletion syndrome 7 (hepatocerebral type)	X	X		X	
C3	C3 deficiency	X			X	
CA2	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis	X			X	
CACNA1A	Hemiplegic migraine, familial; Episodic ataxia, type 2; Spinocerebellar ataxia-6; Cerebellar ataxia, pure	X				
CACNA1C	Timothy syndrome; Brugada syndrome 3	X				
CACNA1F	Night blindness, congenital stationary, X-linked, type 2A; Cone-rod dystrophy, X-linked, 3; Al and Island eye disease	X			X	
CACNA1S	Malignant hyperthermia susceptibility; Hypokalemic periodic paralysis, type 1	X				X
CACNB4	Episodic ataxia, type 5	X				
CAPN3	Muscular dystrophy, limb-girdle, type 2A	X	X		X	
CASK	Mental retardation and microcephaly with pontine and cerebellarhypoplasia; FG syndrome 4	X				
CASQ2	Ventricular tachycardia, catecholaminergic polymorphic, 2	X			X	
CASR	Hypocalciuric hypercalcemia, type I; Hyperparathyroidism, neonatal; Hypocalcemia, autosomal dominant; Hypocalcemia, autosomal dominant, with Bartter syndrome	X			X	
CATSPER1	Male infertility, nonsyndromic, autosomal recessive	X			X	
CAV3	Muscular dystrophy, limb-girdle, type IC; Rippling muscle disease; Creatine phosphokinase, elevated serum; Myopathy, distal, with decreased caveolin 3; Cardiomyopathy, familial hypertrophic; Long QT syndrome-9	X			X	

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CBS	Homocystinuria, B6-responsive and nonresponsive types; Thrombosis, hyperhomocysteinemic	X			X	
CCM2	Cerebral cavernous malformations-2		X			
CD19	Immunodeficiency, common variable, 3	X			X	
CD247	Immunodeficiency due to defect in CD3-zeta	X			X	
CD3D	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive	X			X	
CD3E	Immunodeficiency due to defect in CD3-epsilon; Severe combined immunodeficiency, T cell-negative, B-cell/ natural killer-cell positive	X			X	
CD3G	Immunodeficiency due to defect in CD3-gamma	X			X	
CD40LG	Immunodeficiency, X-linked, with hyper-IgM	X			X	
CDAN1	Anemia, congenital dyserythropoietic, type I	X			X	
CDC73	Hyperparathyroidism-jaw tumor syndrome; Hyperparathyroidism, familial primary; Parathyroid adenoma with cystic changes; Parathyroid carcinoma		X			
CDH1	Breast cancer, lobular; Gastric cancer, familial diffuse, with or without cleft lip and/or palate		X	X		
CDH23	Usher syndrome, type 1D; Deafness, autosomal recessive12		X		X	
CDKL5	Epileptic encephalopathy, early infantile, 2; Angelman syndrome-like	X				
CDKN1B	Multiple endocrine neoplasia, type IV		X			
CDKN1C	Beckwith-Wiedemann syndrome; IMAGE syndrome	X				
CEP290	Joubert syndrome 5; Senior-Loken syndrome 6; Leber congenital amaurosis 10; Meckel syndrome type 4; Bardet-Biedl syndrome 14	X			X	
CFI	Complement factor I deficiency	X			X	
CFP	Properdin deficiency, X-linked	X	X		X	
CFTR	Cystic fibrosis; Congenital bilateral absence of vas deferens; Sweat chloride elevation without CF	X	X		X	
CHAT	Myasthenic syndrome, congenital, associated with episodic apnea	X			X	
CHD7	CHARGE syndrome; Kallmann syndrome 5; Hypogonadotropic hypogonadism	X				
CHEK2	Li-Fraumeni syndrome	X	X	X	X	
CHM	Choroideremia		X			
CHMP2B	Dementia, familial, nonspecific; Amyotrophic lateralsclerosis 17, CHMP2B-related		X			
CHN1	Duane retraction syndrome 2	X				
CHRNA1	Myasthenic syndrome, slow-channel congenital; Myasthenic syndrome, fast-channel congenital; Multiple pterygium syndrome, lethal type	X	X		X	
CHRNA2	Epilepsy, nocturnal frontal lobe, type 4	X				
CHRNA4	Epilepsy, nocturnal frontal lobe, 1	X				
CHRNB2	Epilepsy, nocturnal frontal lobe, 3	X				
CHRND	Myasthenic syndrome, slow-channel congenital; Myasthenic syndrome, fast-channel congenital; Multiple pterygium syndrome, lethal type	X			X	
CHRND	Myasthenic syndrome, slow-channel congenital; Myasthenic syndrome, fast-channel congenital; Multiple pterygium syndrome, lethal type	X			X	

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CHRNE	Myasthenic syndrome, slow-channel congenital; Myasthenic syndrome, fast-channel congenital; Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency	X			X	
CHRNG	Myasthenia gravis, neonatal transient; Escobar syndrome; Multiple pterygium syndrome, lethal type	X			X	
CHST3	Spondyloepiphyseal dysplasia with congenital joint dislocations	X			X	
CIRH1A	Cirrhosis, North American Indian childhood type	X			X	
CISD2	Wolfram syndrome 2 (DIDMOAD)	X				
CLCF1	Cold-induced sweating syndrome 1	X				
CLCN1	Myotonia congenita, recessive; Myotonia congenita, dominant; Myotonia levior, recessive	X			X	
CLCN5	Dent disease; Nephrolithiasis, type I; Hypophosphatemic rickets; Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis	X	X		X	
CLCN7	Osteopetrosis, autosomal recessive 4; Osteopetrosis, autosomal dominant 2	X			X	
CLCNKA	Bartter syndrome, type 4b, digenic	X				
CLCNKB	Bartter syndrome, type 3; Bartter syndrome, type 4	X			X	
CLDN1	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis	X			X	
CLDN14	Deafness, autosomal recessive	X			X	
CLDN19	Hypomagnesemia, renal, with ocular involvement	X			X	
CLN3	Ceroid-lipofuscinosis, neuronal-3, juvenile	X			X	
CLN5	Ceroid-lipofuscinosis, neuronal-5, variant late infantile	X			X	
CLN6	Ceroid-lipofuscinosis, neuronal-6, variant late infantile	X	X		X	
CLN8	Ceroid lipofuscinosis, neuronal 8; Ceroid lipofuscinosis,neuronal, 8, Northern epilepsy variant	X			X	
CLRN1	Usher syndrome, type 3; Usher syndrome type 3A; Usher Syndrome Type 3, Retinitis pigmentosa-61	X			X	
CMS1D	Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency; Myasthenic syndrome, slow-channel congenital	X			X	
CMS1E	Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency; Myasthenic syndrome, slow-channel congenital; Myasthenic syndrome, fast-channel congenital	X			X	
CMS2A	Myasthenic syndrome, slow-channel congenital	X			X	
CNBP	Myotonic dystrophy, type 2	X	X			
CNGA3	Achromatopsia-2	X			X	
CNGB3	Achromatopsia-3; Macular degeneration, juvenile	X			X	
COCH	Deafness, autosomal dominant	X				
COG1	Congenital disorder of glycosylation, type Iig	X			X	
COG4	Congenital disorder of glycosylation, type Iij	X			X	
COG7	Congenital disorder of glycosylation, type Iie	X			X	
COG8	Congenital disorder of glycosylation, type Iih	X			X	
COL11A1	Stickler syndrome, type II; Marshall syndrome; Fibrochondrogenesis	X	X		X	

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COL11A2	Stickler syndrome, type III; Otospondylomegaepiphyseal dysplasia; Weissenbacher-Zweymuller syndrome; Deafness, autosomal dominant 13; Deafness, autosomal recessive 53; Fibrochondrogenesis 2	X			X	
COL17A1	Epidermolysis bullosa, junctional, non-Herlitz type	X			X	
COL18A1	Knobloch syndrome, type 1	X			X	
COL1A1	Osteogenesis imperfecta, type I; OI type II; OI type III; OI type IV; Ehlers-Danlos syndrome, type I; Ehlers-Danlos syndrome, type VIIA; Caffey disease	X				
COL1A2	Ehlers-Danlos syndrome, type VIIB; Osteogenesis imperfecta, type IV; Osteogenesis imperfecta, type III; Osteogenesis imperfecta, type II; Ehlers-Danlos syndrome, cardiac valvular form	X				
COL2A1	Stickler syndrome, type I; Kniest dysplasia; Achondrogenesis-hypochondrogenesis, type II; SED congenita; SMED Strudwick type; Epiphyseal dysplasia, multiple, with myopia and deafness; Spondyloperipheral dysplasia; SED, Namaqualand type; Osteoarthritis with mild chondrodysplasia; Vitreoretinopathy with phalangeal epiphyseal dysplasia; Platyspondylic skeletal dysplasia, Torrance type; Otospondylomegaepiphyseal dysplasia; Avascular necrosis of the femoral head; Legg-Calve-Perthes disease; Stickler sydrome, type I, nonsyndromic ocular	X			X	
COL3A1	Ehlers-Danlos syndrome-vascular type	X				X
COL4A1	Porencephaly; Brain small vessel disease with hemorrhage; Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps; Brain small vessel disease with Axenfeld-Rieger anomaly	X	X			
COL4A3	Alport syndrome, autosomal recessive; Hematuria, benign familial	X			X	
COL4A4	Alport syndrome, autosomal recessive; Hematuria, familial benign	X			X	
COL4A5	Alport syndrome	X				
COL5A1	Ehlers-Danlos syndrome, type II; Ehlers-Danlos syndrome, type I	X				
COL5A2	Ehlers-Danlos syndrome, type I	X				
COL6A1	Bethlem myopathy; Ullrich congenital muscular dystrophy	X			X	
COL6A2	Bethlem myopathy; Ullrich congenital muscular dystrophy; Myosclerosis, congenital	X			X	
COL6A3	Bethlem myopathy; Ullrich congenital muscular dystrophy	X			X	
COL7A1	Epidermolysis bullosa dystrophica, AD; Epidermolysis bullosa dystrophica, AR; Epidermolysis bullosa, pretibial; EBD, Bart type; EBD, localisata variant; Transient bullous of the newborn; Epidermolysis bullosa pruriginosa; Toenail dystrophy, isolated; EBD inversa	X			X	
COL8A2	Corneal dystrophy, Fuchs endothelial, 1; Corneal dystrophy polymorphous posterior	X				
COL9A1	Epiphyseal dysplasia, multiple, 6	X				
COL9A2	Epiphyseal dysplasia, multiple, 2	X			X	
COL9A3	Epiphyseal dysplasia, multiple, 3; Epiphyseal dysplasia, multiple, with myopathy	X				
COLQ	Endplate acetylcholinesterase deficiency	X			X	
COMP	Pseudoachondroplasia; Epiphyseal dysplasia, multiple 1	X				
COQ2	Coenzyme Q10 deficiency; Multiple system Atrophy 1	X			X	
COQ9	Coenzyme Q10 deficiency	X			X	

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COX10	Encephalopathy, progressive mitochondrial, with proximal renaltubulopathy due to cytochrome c oxidase deficiency; Complex IV deficiency	X			X	
COX15	Cardiomyopathy, hypertrophic, early-onset fatal; Leigh syndrome due to cytochrome c oxidase deficiency; Complex IV deficiency	X			X	
COX6B1	Cytochrome c oxidase deficiency; Complex IV deficiency	X			X	
CP	Cerebellar ataxia; Hemosiderosis, systemic, due to aceruloplasminemia		X		X	
CPOX	Coproporphyria; Harderoporphyria	X	X			
CPS1	Carbamoylphosphate synthetase I deficiency	X			X	
CPT1A	CPT deficiency, hepatic, type IA	X			X	
CPT2	Myopathy due to CPT II deficiency; CPT deficiency ,hepatic, type II; CPT II deficiency, lethal neonatal	X			X	
CPX	Chondrodysplasia punctata, X-linked dominant ;Cleft palate with ankyloglossia	X				
CRB1	Retinitis pigmentosa-12, autosomal recessive; Leber congenital amaurosis; Pigmented paravenous chorioretinal atrophy	X			X	
CRD	Cone-rod retinal dystrophy-2; Leber congenitalamaurosis 7; Retinitis pigmentosa, late-onset dominant; Retinitis pigmentosa-3; Cone-rod dystrophy-1; Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness; Macular degeneration, X-linked atrophic	X				
CREBBP	Rubinstein-Taybi syndrome	X				
CRLF1	Cold-induced sweating syndrome; Crisponi syndrome	X			X	
CRTAP	Osteogenesis imperfecta, type IIB; Osteogenesisimperfecta, type VII	X			X	
CST3	Cerebral amyloid angiopathy; Macular degeneration, age-related, 11		X			
CSTB	Epilepsy, progressive myoclonic 1	X			X	
CTDP1	Congenital cataracts, facial dysmorphism, and neuropathy	X			X	
CTNS	Cystinosis, nephropathic; Cystinosis, ocularnonnephropathic; Cystinosis, late-onset juvenile or adolescent nephropathic; Cystinosis, atypical nephropathic	X			X	
CTSA	Galactosialidosis	X			X	
CTSD	Ceroid lipofuscinosis, neuronal	X			X	
CTSK	Pycnodysostosis	X			X	
CUL4B	Mental retardation, X-linked, syndromic 15 (Cabezas type)	X				
CUL7	3-M syndrome	X			X	
CYBA	Chronic granulomatous disease, autosomal, due to deficiency of CYBA	X			X	
CYBB	Chronic granulomatous disease, X-linked	X			X	
CYLD	Cylindromatosis, familial; Brooke-Spiegler syndrome; Trichoepithelioma, multiple familial, 1	X				
CYP11A1	Lipoid congenital adrenal hyperplasia; Adrenal insufficiency, congenital with or without 46, XY sex reversal	X			X	
CYP11B1	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylasedeficiency; Aldosteronism, glucocorticoid-remediable	X			X	

*ACMG minimum list genes

GENE	DISORDER(S)	CHILDHOOD ONSET	ADULT ONSET	CANCER PREDISPOSITION	CARRIER	ACMG*
CYP11B2	Hypoaldosteronism, congenital, due to CMO II deficiency; Hypoaldosteronism, congenital, due to CMO I deficiency; Aldosterone to renin ratioraised	X			X	
CYP17A1	17-alpha-hydroxylase/17, 20-lyase deficiency; 17, 20-lyase deficiency, isolated	X	X		X	
CYP1B1	Glaucoma 3A, primary congenital; Peters anomaly; Glaucoma, early-onset, digenic; Glaucoma, primary open angle, adult-onset; Glaucoma, primary open angle, juvenile-onset	X	X		X	
CYP21A2	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency; Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency	X			X	
CYP27A1	Cerebrotendinous xanthomatosis	X			X	
CYP27B1	Vitamin D-dependent rickets, type I	X			X	
CYP4V2	Bietti crystalline corneoretinal dystrophy		X		X	
D2HGDH	D-2-hydroxyglutaric aciduria	X			X	
DA2B	Arthrogryposis multiplex congenita, distal, type 2B; Arthyrgryposis, distal, type 2B; Arthrogryposis multiplex congenita, distal, type 1; Nemaline myopath	X				
DARS2	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation	X			X	
DBH	Dopamine beta-hydroxylase deficiency	X			X	
DBT	Maple syrup urine disease, type II	X			X	
DCLRE1C	Severe combined immunodeficiency, Athabascan type; Omenn syndrome	X			X	
DCN	Corneal dystrophy, congenital stromal	X				
DCTN1	Neuropathy, distal hereditary motor, type VIIB; Perry syndrome		X		X	
DCX	Lissencephaly, X-linked; Subcortical laminalheteropia, X-linked	X				
DDB2	Xeroderma pigmentosum, group E, DDB-negative subtype	X			X	
DDC	Aromatic L-amino acid decarboxylase deficiency	X			X	
DES	Myopathy, desmin-related, cardioskeletal; Cardiomyopathy, dilated, 1I; Scapuloperoneal syndrome, neurogenic, Kaeser type		X		X	
DFNA5	Deafness, autosomal dominant 5	X	X			
DFNB31	Deafness, autosomal recessive 31; Usher syndrome, typeIID	X			X	
DFNB59	Deafness, autosomal recessive 59	X			X	
DGUOK	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type)	X			X	
DHCR24	Desmosterolosis	X			X	
DHCR7	Smith-Lemli-Opitz syndrome	X			X	
DHH	46XY partial gonadal dysgenesis, with minifascicular neuropathy; 46XY complete gonadal dysgenesis	X			X	
DHODH	Miller syndrome	X			X	
DICER1	Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors; Pleuropulmonary blastoma; Rhabdomyosarcoma, embryonal, 2	X				
DKC1	Dyskeratosis congenita-1; Hoyeraal-Hreidarsson syndrome	X			X	
DLAT	Pyruvate dehydrogenase E2 deficiency	X			X	
DLD	Dihydrolipoamide dehydrogenase deficiency	X			X	

*ACMG minimum list genes

GENE	DISORDER(S)	CHILDHOOD ONSET	ADULT ONSET	CANCER PREDISPOSITION	CARRIER	ACMG*
DLG3	Mental retardation, X-linked-90	X			X	
DLL3	Spondylocostal dysostosis, autosomal recessive, 1	X			X	
DMD	Duchenne muscular dystrophy; Becker muscular dystrophy; Cardiomyopathy, dilated, 3B	X	X		X	
DMP1	Hypophosphatemic rickets, AR	X			X	
DMPK	Myotonic dystrophy	X	X			
DNAH5	Ciliary dyskinesia, primary, 3, with or without situs inversus	X			X	
DNAI1	Ciliary dyskinesia, primary, 1, with our without situs inversus	X			X	
DNAI1	Ciliary dyskinesia, primary, 3, with or without situs inversus; Ciliary dyskinesia, primary, 1, with our without situs inversus	X			X	
DNAJC19	3-methylglutaconic aciduria, type V	X			X	
DNAJC5	Ceroid lipofuscinosis, neuronal, 4, Parry type	X				
DNM2	Charcot-Marie-Tooth disease, dominant intermediate B; Myopathy, centronuclear; Charcot-Marie-Tooth disease, axonal, type 2M	X	X		X	
DNMT1	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, Neuropathy, hereditary sensory, type IE		X			
DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome	X			X	
DOCK6	Adams-Oliver syndrome 2	X			X	
DOCK8	Mental retardation, autosomal dominant 2; Hyper-IgE recurrentinfection syndrome, autosomal recessive	X			X	
DOK7	Myasthenia, limb-girdle, familial; Fetal akinesiadeformation sequence	X			X	
DOLK	Congenital disorder of glycosylation, type Im	X			X	
DPAGT1	Congenital disorder of glycosylation, type Ij	X			X	
DPM1	Congenital disorder of glycosylation, type Ie	X			X	
DPYD	Dihydropyrimidine dehydrogenase deficiency; 5-fluorouracil toxicity	X			X	
DRD5	Dystonia, primarycervical	X	X			
DSC2	Arrhythmogenic right venticular cardiomyopathy	X	X		X	X
DSG2	Arrhythmogenic right venticular cardiomyopathy	X	X			X
DSG4	Hypotrichosis, localized, autosomal recessive	X			X	
DSP	Arrhythmogenic right venticular cardiomyopathy	X	X		X	X
DSPP	Dentinogenesis imperfecta, Shields type II; Deafness, autosomal dominant 36, with dentinogenesis; Dentinogenesis imperfecta, Shields type III; Dentin dysplasia, type II	X				
DUOX2	Thryoid dyshormonogenesis 6	X			X	
DYNC1H1	Charcot-Marie-Tooth disease, axonal, type 20; Mental retardation, autosomal dominant 13; Spinal muscular atrophy, lower extremity-predominant, AD	X				
DYNC2H1	Asphyxiating thoracic dystrophy 3; Short rib-polydactylysyndrome, type III; Short rib-polydactyly syndrome, type II, digenic	X			X	
DYSF	Muscular dystrophy, limb-girdle, type 2B; Miyoshimyopathy; Myopathy, distal, with anterior tibial onset	X	X		X	
EBP	Chondrodysplasia punctata, X-linked dominant	X				

*ACMG minimum list genes

GENE	DISORDER(S)	CHILDHOOD ONSET	ADULT ONSET	CANCER PREDISPOSITION	CARRIER	ACMG*
EDA	Ectodermal dysplasia, anhidrotic, X-linked; Tooth agenesis, selective, X-linked 1	X				
EDAR	Ectodermal dysplasia, hypohidrotic, autosomal dominant; Ectodermal dysplasia, hypohidrotic, autosomal recessive	X			X	
EDARADD	Ectodermal dysplasia, anhidrotic, autosomal recessive; Ectodermal dysplasia, anhidrotic, autosomal dominant	X			X	
EDN3	Waardenburg syndrome, type 4B; Central hypoventilation syndrome, congenital	X			X	
EDNRB	ABCD syndrome; Waardenburg syndrome, type 4A	X			X	
EFEMP2	Cutis laxa, autosomal recessive, type I	X			X	
EFNB1	Craniofrontonasal dysplasia	X				
EGLN1	Erythrocytosis, familial, 3	X				
EGR2	Neuropathy, congenital hypomyelinating, 1; Charcot-Marie-Tooth disease, type 1D; Dejerine-Sottas neuropathy	X			X	
EHK	Epidermolytic hyperkeratosis; Ichthyosis, cyclic, with epidermolytic hyperkeratosis; Ichthyosis histrix, Curth-Macklin Palmoplantar keratoderma, nonepidermolytic; Plamoplantar keratoderma, epidermolytic; Keratosis palmoplantaris striata II; Epidermolytic hyperkeratosis; Ichthyosis, cyclic, with epidermolytic hyperkeratosis; Ichthyosis with confetti	X				
EHMT1	Kleefstra syndrome	X				
EIF2AK3	Wolcott-Rallison syndrome	X			X	
EIF2B1	Leukoencephalopathy with vanishing white matter	X			X	
EIF2B2	Leukoencephalopathy with vanishing white matter; Ovarioleukodystrophy	X			X	
EIF2B3	Leukoencephalopathy with vanishing white matter	X			X	
EIF2B4	Leukoencephaly with vanishing white matter; Ovarioleukodystrophy	X			X	
EIF2B5	Leukoencephalopathy with vanishing white matter; Ovarioleukodystrophy	X			X	
ELANE	Hematopoiesis, cyclic; Neutropenia, severe congenital, autosomal dominant 1	X				
ELN	Supravalvar aortic stenosis; Cutis laxa, AD	X				
ELOVL4	Stargardt disease 3; Macular dystrophy, autosomal dominant, chromosome 6-linked	X			X	
EMD	Emery-Dreifuss muscular dystrophy	X			X	
ENAM	Amelogenesis imperfecta, type IB; Amelogenesis imperfecta, type IC	X			X	
ENG	Telangiectasia, hereditary hemorrhagic, type 1	X				
ENPP1	Ossification of posterior longitudinal ligament of spine; Arterial calcification, generalized, of infancy; Hypophosphatemic rickets, autosomal recessive, 2	X	X		X	
EOGT	Adams-Oliver syndrome 4	X			X	
EP300	Colorectal cancer; Rubinstein-Taybi syndrome 2	X	X	X		
EPAS1	Erythrocytosis, familial, 4		X			
EPCAM	Diarrhea 5, with tufting enteropathy, congenital; Colorectal cancer, hereditary nonpolyposis, type I		X	X	X	
EPM2A	Epilepsy, progressive myoclonic 2A (Lafora)	X			X	

*ACMG minimum list genes

GENE	DISORDER(S)	CHILDHOOD ONSET	ADULT ONSET	CANCER PREDISPOSITION	CARRIER	ACMG*
EPPK	Epidermolytic hyperkeratosis; Ichthyosis, cyclic, with epidermolytic hyperkeratosis; Ichthyosis histrix, Curth-Macklin Palmoplantar keratoderma, nonepidermolytic; Plamoplantarkeratoderma, epidermolytic; Keratosis palmoplantaris striata III; Epidermolytic palmoplantar keratoderma	X				
ERBB3	Lethal congenital contractural syndrome 2	X			X	
ERCC2	Xeroderma pigmentosum, group D; Trichothiodystrophy; Cerebrooculofacioskeletal syndrome 2	X			X	
ERCC3	Xeroderma pigmentosum, group B; Trichothiodystrophy	X			X	
ERCC4	Xeroderma pigmentosum, group F; XFE progeroid syndrome	X			X	
ERCC5	Xeroderma pigmentosum, group G; Cerebrooculofacioskeletalsyndrome 3	X			X	
ERCC6	Cockayne syndrome, type B; Cerebrooculofacioskeletal syndrome 1; De Sanctis-Cacchione syndrome; UV-sensitive syndrome	X	X		X	
ERCC8	Cockayne syndrome, type A	X			X	
ESCO2	Roberts syndrome; SC phocomelia syndrome	X			X	
ESPN	Deafness, autosomal recessive 36; Deafness, neurosensory, without vestibular involvement, autosomal dominant	X	X		X	
ETFA	Glutaricaciduria, type IIA	X			X	
ETFB	Glutaricaciduria, type IIB	X			X	
ETFDH	Glutaricaciduria, type IIC	X	X		X	
ETHE1	Ethylmalonic encephalopathy	X			X	
EVC	Ellis-van Creveld syndrome; Weyers acrodental dysostosis	X			X	
EVC2	Ellis-van Creveld syndrome; Weyers acrodental dysostosis	X			X	
EXT1	Exostoses, multiple, type 1	X				
EXT2	Exostoses, multiple, type 2	X				
EYA1	Branchiootorenal syndrome 1 with or without cataract; Branchiootic syndrome 1; Anterior segment anomalies with or without cataract; Otofaciocervical syndrome	X				
EYA4	Deafness, autosomal dominant 10; Cardiomyopathy, dilated, 1J	X	X			
EZH2	Weaver syndrome	X				
F11	Factor XI deficiency, autosomal recessive; Factor XI deficiency, autosomal dominant	X	X		X	
F12	Factor XII deficiency; Angioedema, hereditary, type III	X	X		X	
F2	Hypoprothrombinemia; Dysprothrombinemia; Thrombophilia due to thrombin defect	X	X		X	
F5	Factor V deficiency; Thrombophilia due to activated protein C resistance	X	X		X	
F8	Hemophilia A	X			X	
F9	Hemophilia B; Thrombophilia, X-linked, due to factor IX defect	X	X		X	
FA2H	Spastic paraplegia 35, autosomal recessive	X			X	
FAH	Tyrosinemia, type I	X			X	
FAM126A	Leukodystrophy, hypomyelinating, 5	X			X	
FAM134B	Neuropathy, hereditary sensory and autonomic, type IIB	X			X	
FAM20C	Raine syndrome	X			X	

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GENE	DISORDER(S)	CHILDHOOD ONSET	ADULT ONSET	CANCER PREDISPOSITION	CARRIER	ACMG*
FANCA	Fanconi anemia, complementation group A	X			X	
FANCC	Fanconi anemia, complementation group C	X			X	
FANCG	Fanconi anemia, complementation group G	X			X	
FAS	Autoimmune lymphoproliferative syndrome IA	X			X	
FASTKD2	Mitochondrial complex IV deficiency	X			X	
FBLN5	Cutis laxa, autosomal recessive, type IA; Cutis laxa, autosomal dominant 2; Macular degeneration, age-related, 3	X	X		X	
FBN1	Marfan syndrome; Loeys-Dietz syndrome; and Familial thoracic aortic aneurysms and dissections	X				X
FBN2	Contractural arachnodactyly, congenital	X				
FECH	Protoporphyria, erythropoietic, autosomal recessive	X	X		X	
FERMT3	Leukocyte adhesion deficiency, type III	X			X	
FGA	Amyloidosis, hereditary renal; Afibrinogenemia, congenital; Dysfibrinogenemia, alpha type, causing bleeding diathesis; Dysfibrinogenemia, alpha type, causing recurrent thrombosis	X	X		X	
FGB	Dysfibrinogenemia, beta type; Afibrinogenemia, congenital; Thrombophilia, dysfibrinogenemic	X			X	
FGD1	Aarskog-Scott syndrome; Mental retardation, X-linked syndromic 16	X			X	
FGD4	Charcot-Marie-Tooth disease, type 4H	X			X	
FGF10	Aplasia of lacrimal and salivary glands; LADD syndrome	X				
FGF23	Hypophosphatemic rickets, autosomal dominant; Osteomalacia, tumor-induced; Tumoral calcinosis, hyperphosphatemic, familial	X	X		X	
FGF3	Deafness, congenital with inner ear agenesis, microtia, and microdontia	X			X	
FGFR1	Hartsfield syndrome; Pfeiffer syndrome; Jackson-Weiss syndrome; Kallmann syndrome 2; Osteoglophonic dysplasia; Trigonocephaly 1	X				
FGFR2	Crouzon syndrome; Jackson-Weiss syndrome; Beare-Stevenson cutis gyrata syndrome; Pfeiffer syndrome; Apert syndrome; Saethre-Chotzen syndrome; Craniosynostosis, nonspecific; Craniofacial-skeletal-dermatologic dysplasia; Antley-Bixler syndrome; Scaphocephaly and Axenfeld-Rieger anomaly; LADD syndrome; Scaphocephaly, maxillary retrusion, and mental retardation; Bent bone dysplasia syndrome	X				
FGFR3	Achondroplasia; Hypochondroplasia; Thanatophoric dysplasia, type I; Thanatophoric dysplasia, type II; Crouzon syndrome with acanthosis nigricans; Muenke syndrome; LADD syndrome; CATSHL syndrome; Nevus, keratinocytic, nonepidermolytic	X				
FGG	Dysfibrinogenemia, gamma type; Hypofibrinogenemia, gamma type; Thrombophilia, dysfibrinogenemic	X	X		X	
FH	Fumarase deficiency; Leiomyomatosis and renal cell cancer	X	X	X	X	
FIG4	Charcot-Marie-Tooth disease, type 4J; Amyotrophic lateralsclerosis 11; Yunis-Varon syndrome	X	X		X	
FKBP10	Osteogenesis imperfecta, type XI; Bruck syndrome 1	X			X	
FKRP	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5; Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5	X	X		X	

*ACMG minimum list genes

GENE	DISORDER(S)	CHILDHOOD ONSET	ADULT ONSET	CANCER PREDISPOSITION	CARRIER	ACMG*
FKTN	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4; Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4; Cardiomyopathy,dilated, 1X; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4	X			X	
FLCN	Birt-Hogg-Dube syndrome; Pneumothorax, primary spontaneous	X	X			
FLNA	Heterotopia, periventricular; Otopalatodigital syndrome,type I; Otopalatodigital syndrome, type II; Intestinal pseudoobstruction, neuronal; Melnick-Needles syndrome; Frontometaphyseal dysplasia; Heterotopia, periventricular, ED variant; FG syndrome 2; Cardiac valvular dysplasia, X-linked; Terminal osseous dysplasia; Congenital short bowel syndrome	X				
FLNB	Spondylocarpotarsal synostosis syndrome; Larsen syndrome; Atelostogenesis, type I; Atelosteogenesis, type III; Boomerang dysplasia	X			X	
FLT4	Lymphedema, hereditary I	X				
FMR1	Fragile X syndrome; Fragile X tremor/ataxia syndrome; Fragile X Syndrome; Premature ovarian failure 1	X			X	
FOLR1	Neurodegeneration due to cerebral folate transport deficiency	X			X	
FOXC1	Iridogoniodysgenesis, type 1; Rieger or Axenfeld anomalies; Axenfeld-Rieger syndrome, type 3; Iris hypoplasia and glaucoma	X				
FOXC2	Lymphedema-distichiasis syndrome; Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus	X				
FOXF1	Alveolar capillary dysplasia with misalignment of pulmonary veins	X			X	
FOXG1	Rett syndrome, congenital variant	X				
FOXL2	Blepharophimosis, epicanthus inversus, and ptosis, type 1; Blepharophimosis, epicanthus inversus, and ptosis, type 2 Premature ovarian failure 3	X				
FOXN1	T-cell immunodeficiency, congenital alopecia, and nail dystrophy	X				
FOXP3	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked	X			X	
FRAS1	Fraser syndrome	X			X	
FREM1	Bifid nose with or without anorectal and renal anomalies; Manitoba oculotrichoanal syndrome; Trigonocephaly 2	X			X	
FREM2	Fraser syndrome	X				
FRMD7	Nystagmus 1, congenital, X-linked	X				
FTCD	Glutamate formiminotransferase deficiency	X			X	
FTL	Hyperferritinemia-cataract syndrome; Neurodegeneration with brain iron accumulation 3; L-ferritin deficiency, dominant and recessive	X	X		X	
FTSJ1	Mental retardation, X-linked-9	X			X	
FUCA1	Fucosidosis	X			X	
FUS	Amyotrophic lateral sclerosis 6, autosomal recessive, with or without frontotemporal dementia; Tremor, hereditary essential, 4	X	X		X	
FXN	Friedreich ataxia; Friedreich ataxia with retained reflexes	X	X		X	
FZD4	Exudative vitreoretinopathy; Retinopathy of prematurity	X				

*ACMG minimum list genes

GENE	DISORDER(S)	CHILDHOOD ONSET	ADULT ONSET	CANCER PREDISPOSITION	CARRIER	ACMG*
G6PC	Glycogen Storage Disease Type Ia	X			X	
G6PC3	Neutropenia, severe congenital, autosomal recessive 4; Darsun syndrome	X			X	
G6PD	Favism; Hemolytic anemia due to G6PD deficiency	X			X	
GAA	Glycogen storage disease II	X	X		X	
GALC	Krabbe disease	X	X		X	
GALE	Galactose epimerase deficiency (Galactosemia type III)	X			X	
GALK1	Galactokinase deficiency with cataracts (Galactosemia type II)	X			X	
GALNS	Mucopolysaccharidosis IVA	X			X	
GALNT3	Tumoral calcinosis, hyperphosphatemic, familial	X	X		X	
GALT	Galactosemia	X			X	
GAMT	Cerebral creatine deficiency syndrome 2	X			X	
GAN	Giant axonal neuropathy-1	X			X	
GARS	Charcot-Marie-Tooth disease, type 2D; Neuropathy, distal hereditary motor, type VA	X	X			
GATA1	Anemia, X-linked, with/without neutropenia and/or platelet abnormalities; Thrombocytopenia, X-linked, with or without dyserythropoietic anemia; Leukemia, megakaryoblastic, with or without Down syndrome; Thrombocytopenia with beta-thalassemia, X-linked	X			X	
GATM	Cerebral creatine deficiency syndrome 3	X			X	
GBA	Gaucher disease, type I; Gaucher disease, type II; Gaucher disease, type III; Gaucher disease, type IIIC; Gaucher disease, perinatal lethal	X	X		X	
GBE1	Glycogen storage disease IV; Polyglucosan body disease, adult form	X	X		X	
GCDH	Glutaricaciduria, type I	X	X		X	
GCH1	Dystonia, DOPA-responsive, with or without hyperphenylalainemia; Hyperpehnylalaninemia, BH4-deficient, B	X	X		X	
GCSH	Glycine encephalopathy	X			X	
GDAP1	Charcot-Marie-Tooth disease, type 4A; Charcot-Marie-Tooth disease, axonal, with vocal cord paresis; Charcot-Marie-Tooth disease, axonal, type 2K; Charcot-Marie-Tooth disease, recessive intermediate, A	X	X		X	
GDF3	Klippel-Feil syndrome 3, autosomal dominant; Microphthalmia with coloboma 6; Microphthalmia, isolated 7	X				
GDF6	Klippel-Feil syndrome 1, autosomal dominant; Microphthalmia, isolated 4; Leber congenital amaurosis 17; Microphthalmia with coloboma 6, digenic	X				
GDI1	Mental retardation, X-linked nonspecific	X			X	
GFAP	Alexander disease	X	X			
GFER	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay	X			X	
GFM1	Combined oxidative phosphorylation deficiency 1	X			X	
GH1	Growth hormone deficiency, isolated, type IA; Growthhormone deficiency, isolated, type IB; Growth hormone deficiency, isolated, type II; Kowarski syndrome	X			X	
GHRHR	Growth hormone deficiency, isolated, type IB	X			X	

*ACMG minimum list genes

GENE	DISORDER(S)	CHILDHOOD ONSET	ADULT ONSET	CANCER PREDISPOSITION	CARRIER	ACMG*
GJA1	Oculodentodigital dysplasia; Craniometaphyseal dysplasia, autosomal recessive; Syndactyly, type III; Hypoplastic left heart syndrome; Atrioventricular septal defect; Oculodentodigital dysplasia, autosomal recessive	X			X	
GJB1	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1	X				
GJB2	Deafness, autosomal recessive 1A; Deafness, autosomal dominant 3A; Vohwinkel syndrome; Keratoderma, palmoplantar, with deafness; Keratitis-ichthyosis-deafness syndrome; Hystrix-like ichthyosis with deafness; Bart-Pumphrey syndrome	X			X	
GJB3	Erythrokeratodermia variabilis et progressiva; Deafness,autosomal dominant 2B; Deafness, autosomal recessive; Deafness, autosomal dominant, with peripheral neuropathy; Deafness, digenic,GJB2/GJB3	X			X	
GJB4	Erythrokeratodermia variabilis with erythema gyratum repens	X				
GJB6	Deafness, autosomal dominant 3B; Ectodermal dysplasia 2, Clouston type; Deafness, autosomal recessive 1B; Deafness, digenic GJB2/GJB6	X			X	
GJC2	Leukodystrophy, hypomyelinating, 2; Spastic paraplegia, 44; Lymphedema, hereditary, IC	X			X	
GLA	Familial hypertrophic cardiomyopathy; Dilated cardiomyopathy; Fabry disease	X	X			X
GLB1	GM1-gangliosidosis, type I; GM1-gangliosidosis, type II; GM1-gangliosidosis, type III; Mucopolysaccharidosis type IVB (Morquio)	X	X		X	
GLDC	Glycine encephalopathy	X			X	
GLE1	Lethal congenital contracture syndrome 1; Arthrogryposis, lethal, with anterior horn cell disease	X			X	
GLI3	Greig cephalopolysyndactyly syndrome; Pallister-Hallsyndrome; Polydactyly, preaxial, type IV; Polydactyly, postaxial, types A1 and B	X				
GLRA1	Hyperekplexia, hereditary 1, autosomal dominant or recessive	X			X	
GM2A	GM2-gangliosidosis, AB variant	X			X	
GNAS	Pseudohypoparathyroidism; McCune-Albright syndrome; Osseous heteroplasia, progressive; Prolonged bleeding time, brachydactyly and mental retardation; Pseudopseudohypoparathyroidism	X				
GNAT1	Night blindness, congenital stationary, autosomal dominant 3	X				
GNAT2	Achromatopsia-4	X			X	
GNE	Sialuria; Inclusion body myopathy, autosomal recessive; Nonaka myopathy	X	X		X	
GNMT	Glycine N-methyltransferase deficiency	X			X	
GNPAT	Chondrodysplasia punctata, rhizomelic, type 2	X			X	
GNPTAB	Mucolipidosis III alpha/beta; Mucolipidosis II alpha/beta	X			X	
GNPTG	Mucolipidosis III gamma	X			X	
GNRHR	Hypogonadotropic hypogonadism 7 with or without anosmia; Fertile eunuch syndrome	X	X		X	
GNS	Mucopolysaccharidosis type IIID	X			X	
GPC3	Simpson-Golabi-Behmel syndrome, type 1	X			X	
GPI	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency	X			X	

*ACMG minimum list genes

GENE	DISORDER(S)	CHILDHOOD ONSET	ADULT ONSET	CANCER PREDISPOSITION	CARRIER	ACMG*
GPR143	Ocular albinism, type I, Nettleship-Falls type; Nystagmus 6, congenital, X-linked	X			X	
GPR56	Polymicrogyria, bilateral frontoparietal	X			X	
GPR98	Febrile seizures, familial, 4; Usher syndrome, type IIC	X			X	
GRHPR	Hyperoxaluria, primary, type II	X	X		X	
GRIK2	Mental retardation, autosomal recessive, 6	X			X	
GRM6	Night blindness, congenital stationary, type 1B	X			X	
GRN	Frontotemporal lobar degeneration with ubiquitin-positive inclusions; Aphasia, primary progressive; Ceroid lipofuscinosis, neuronal		X		X	
GSN	Amyloidosis, Finnish type		X			
GSS	Hemolytic anemia due to glutathione synthetase deficiency; Glutathione synthetase deficiency	X			X	
GTF2H5	Trichothiodystrophy, complementation group A	X			X	
GUCY2D	Leber congenital amaurosis 1; Cone-rod dystrophy 6	X			X	
GUSB	Mucopolysaccharidosis VII	X			X	
GYS1	Glycogen storage disease 0, muscle	X			X	
GYS2	Glycogen storage disease, type 0	X			X	
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency; Hyperinsulinemic hypoglycemia, familial, 4	X			X	
HADHA	LCHAD deficiency; Trifunctional protein deficiency; HELLP syndrome, maternal, of pregnancy; Fatty liver, acute, of pregnancy	X			X	
HADHB	Trifunctional protein deficiency	X			X	
HAMP	Hemochromatosis, type 2B	X	X		X	
HAX1	Neutropenia, severe congenital, autosomal recessive 3	X			X	
HBA1	Thalassemias, alpha-; Methemoglobinemias, alpha-; Erythremias, alpha-; Heinz body anemias, alpha-; Hemoglobin H disease, nondeletional	X			X	
HBA2	Thalassemia, alpha-; Hemoglobin H disease, nondeletional; Heinz body anemia; Erythrocytosis; Hypochromic microcytic anemia	X			X	
HBB	Delta-beta thalassemia; Sickle cell anemia; Thalassemias, beta-; Erythremias, beta-; Methemoglobinemias, beta-; Heinz body anemias, beta-; Thalassemia-beta, dominant inclusion-body; Hereditary persistence of fetal hemoglobin	X			X	
HCCS	Microphthalmia, syndromic 7	X				
HCN4	Sick sinus syndrome 2; Brugada syndrome 8	X	X			
HES7	Spondylocostal dysostosis 4, autosomal recessive	X			X	
HESX1	Septooptic dysplasia; Pituitary hormone deficiency,combined, 5; Growth hormone deficiency with pituitary anomalies	X			X	
HEXA	Tay-Sachs disease; GM2-gangliosidosis, several forms	X	X		X	
HEXB	Sandhoff disease, infantile, juvenile, and adult forms	X	X		X	
HFE	Hemochromatosis		X		X	
HFE2	Hemochromatosis, type 2A	X	X		X	
HGD	Alkaptonuria	X	X		X	
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo C)	X			X	
HIBCH	3-hydroxyisobutryl-CoA hydrolase deficiency	X			X	
HLCS	Holocarboxylase synthetase deficiency	X			X	

*ACMG minimum list genes

GENE	DISORDER(S)	CHILDHOOD ONSET	ADULT ONSET	CANCER PREDISPOSITION	CARRIER	ACMG*
HMBS	Porphyria, acute intermittent; Porphyria, acute intermittent, nonerythroid variant		X			
HMGCL	HMG-CoA lyase deficiency	X			X	
HNF1B	Renal cysts and diabetes syndrome; Diabetes mellitus,noninsulin-dependent	X	X			
HOXA13	Hand-foot-uterus syndrome; Guttmacher syndrome	X				
HPD	Tyrosinemia, type III; Hawkinsinuria	X			X	
HPRT1	Lesch-Nyhan syndrome; HPRT-related gout	X			X	
HPS1	Hermansky-Pudlak syndrome 1	X			X	
HPS3	Hermansky-Pudlak syndrome 3	X			X	
HPSE2	Urofacial syndrome 1	X			X	
HRAS	Congenital myopathy with excess of muscle spindles; Costello syndrome; Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic	X				
HSD11B2	Apparent mineralocorticoid excess	X			X	
HSD17B10	17-beta-hydroxysteroid dehydrogenase X deficiency; Mental retardation, X-linked syndromic 10; Mental retardation, X-linked 17/31, microduplication	X			X	
HSD17B3	Pseudohermaphroditism, male, with gynecomastia (17-beta hydroxysteroid dehydrogenase 3 deficiency)	X			X	
HSD17B4	D-bifunctional protein deficiency; Perrault syndrome 1	X			X	
HSD3B2	3-beta-hydroxysteroid dehydrogenase, type II, deficiency	X			X	
HSPB1	Neuropathy, distal hereditary motor, type IIB; Charcot-Marie-Tooth disease, axonal, type 2F	X	X		X	
HSPB8	Neuropathy, distal hereditary motor, type IIA; Charcot-Marie-Tooth disease, axonal, type 2L	X	X			
HSPG2	Schwartz-Jampel syndrome, type 1; Dyssegmental dysplasia, Silverman-Handmaker type	X			X	
HTRA1	CARASIL syndrome	X	X		X	
HUWE1	Mental retardation, X-linked syndromic, Turner type	X				
ICOS	Immunodeficiency, common variable, 1		X		X	
IDH2	D-2-hydrosyglutaric aciduria 2	X				
IDS	Mucopolysaccharidosis II	X			X	
IDUA	Mucopolysaccharidosis Ih; Mucopolysaccharidosis Is; Mucopolysaccharidosis Ih/s	X			X	
IFNGR1	Mycobacterial infection, atypical, familial disseminated; BCG infection, generalized familial	X			X	
IFNGR2	Mycobacterial infection, atypical, familial disseminated	X			X	
IFT122	Cranioectodermal dysplasia	X			X	
IFT80	Asphyxiating thoracic dystrophy 2	X			X	
IGHD1B	Growth hormone deficiency, isolated, type IA; Growth hormone deficiency, isolated, type IB; Growth hormone deficiency, isolated, type II; Kowarski syndrome; Growth hormone deficiency, isolated, type IB	X			X	
IGHMBP2	Neuronopathy, distal hereditary motor, type VI	X			X	
IKBKAP	Dysautonomia, familial	X			X	
IKBKG	Incontinentia pigmenti, type II; Ectodermal dysplasia,hypohidrotic, with immune deficiency; Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency; Immunodeficiency,isolated; Invasive pneumococcal disease, recurrent isolated, 2	X			X	

*ACMG minimum list genes

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IL12B	BCG and salmonella infection, disseminated	X			X	
IL12RB1	Mycobacterial and salmonella infections, susceptibility to	X			X	
IL1RAPL1	Mental retardation, X-linked, 21/34	X			X	
IL1RN	Interleukin 1 receptor antagonist deficiency	X			X	
IL2RG	Severe combined immunodeficiency, X-linked; Combined immunodeficiency, X-linked, moderate	X			X	
IL7R	Severe combined immunodeficiency, T-cell negative, B-cell/naturalkiller cell-positive type	X			X	
INS	Hyperproinsulinemia, familial, with or without diabetes; Maturity-onset diabetes of the young, type 10; Diabetes mellitus, permanent neonatal; Diabetes mellitus, type 1; Diabetes mellitus, insulin-dependent, 2	X				
INSR	Leprechaunism; Rabson-Mendenhall syndrome; Diabetes mellitus, insulin-resistant, with acanthosis nigricans; Hyperinsulinemic hypoglycemia, familial, 5	X			X	
INVS	Nephronophthisis 2, infantile	X			X	
IQCB1	Senior-Loken syndrome 5	X			X	
IRAK4	IRAK4 deficiency; Invasive pneumococcal disease, recurrent isolated, 1	X			X	
IRF6	van der Woude syndrome; Popliteal pterygium syndrome; Orofacial cleft 6	X				
ISCU	Myopathy with lactic acidosis, hereditary	X			X	
ITGA6	Epidermolysis bullosa, junctional, with pyloric stenosis	X			X	
ITGB2	Leukocyte adhesion deficiency	X			X	
ITGB4	Epidermolysis bullosa, junctional, with pyloric atresia; Epidermolysis bullosa, junctional, non-Herlitz type; Epidermolysis bullosa of hands and feet	X			X	
ITM2B	Dementia, familial British; Dementia, familial Danish	X				
IVD	Isovaleric acidemia	X			X	
JAG1	Alagille syndrome; Tetralogy of Fallot; Deafness, congenital heart defects, and posterior embryotoxon	X				
JAK2	Thrombocythemia-3		X			
JAK3	SCID, autosomal recessive, T-negative/B-positive type	X			X	
KAL1	Kallmann syndrome	X			X	
KCNA1	Episodic ataxia/myokymia syndrome	X				
KCNE1	Jervell and Lange-Nielsen syndrome 2; Long QT syndrome-5	X			X	
KCNE2	Long QT syndrome-6; Atrial fibrillation, familial, 4	X	X			
KCNE3	Brugada syndrome 6		X			
KCNH2	Romano-Ward Long QT syndromes types 1, 2 and 3; Brugada syndrome		X			X
KCNJ1	Bartter syndrome, type 2	X				
KCNJ11	Hyperinsulinemic hypoglycemia, familial, 2; Diabetes, permanent neonatal; Diabetes mellitus, permanent neonatal, with neurologic features; Diabetes mellitus, transient neonatal, 3	X			X	
KCNJ2	Long QT syndrome-7; Short QT syndrome-3	X				
KCNQ1	Romano-Ward Long QT syndromes types 1, 2 and 3; Brugada syndrome; Jervell and Lange-Nielsen syndrome	X			X	X
KCNQ2	Seizures, benign neonatal, 1; Myokymia; Epileptic encephalopathy, early infantile, 7	X				

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KCNQ3	Epilepsy, benign neonatal, type 2	X				
KCNQ4	Deafness, autosomal dominant 2A	X				
KDM5C	Mental retardation, X-linked, syndromic, JARID1C-related	X			X	
KIAA0196	Spastic paraplegia-8; Ritscher-Schinzel syndrome	X	X		X	
KIF1B	Charcot-Marie-Tooth disease, type 2A1; Pheochromocytoma	X	X			
KIF21A	Fibrosis of extraocular muscles, congenital, 1; Fibrosis of extraocular muscles, congenital, 3B	X				
KIT	Piebaldism; Mast cell disease; Gastrointestinal stromal tumor, familial; Leukemia, acute myeloid	X	X	X		
KMT2D	Kabuki syndrome	X				
KRIT1	Cerebral cavernous malformations-1; Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations; Cavernous malformations of CNS and retina	X	X			
KRT1	Epidermolytic hyperkeratosis; Ichthyosis, cyclic, with epidermolytic hyperkeratosis; Ichthyosis histrix, Curth-Macklin type; Palmoplantar keratoderma, nonepidermolytic; Plamoplantar keratoderma, epidermolytic; Keratosis palmoplantaris striata III	X				
KRT10	Epidermolytic hyperkeratosis; Ichthyosis, cyclic, with epidermolytic hyperkeratosis; Ichthyosis with confetti	X			X	
KRT12	Meesmann corneal dystrophy	X				
KRT14	Epidermolysis bullosa simplex, Dowling-Meara type; Epidermolysis bullosa simplex, Koebner type; Epidermolysis bullosa simplex, recessive 1; Naegeli-Franceschetti-Jadassohn syndrome; Dermatopathia pigmentosa reticularis; Epidermolysis bullosa simplex, Weber-Cockayne type	X	X		X	
KRT16	Pachyonychia congenita, Jadassohn-Lewandowsky type; Palmoplantar keratoderma, nonepidermolytic,focal	X				
KRT17	Pachyonychia congenita, Jackson-Lawler type; Steatocystoma multiplex	X				
KRT3	Meesmann corneal dystrophy	X				
KRT5	Epidermolysis bullosa simplex, Dowling-Meara type; Epidermolysis bullosa simplex, Koebner type; Epidermolysis bullosa simplex, Weber-Cockayne type; Epidermolysis bullosa simplex with mottled pigmentation; Dowling-Degos disease 1; Epidermolysis bullosa simplex with migratory circinate erythema; Epidermolysis bullosa simplex, recessive 1	X	X		X	
KRT6A	Pachyonychia congenita, Jadassohn-Lewandowsky type	X				
KRT6B	Pachyonychia congenita, Jackson-Lawler type	X				
KRT6B	Pachyonychia congenita, Jackson-Lawler type; Steatocystoma multiplex; Pachyonychia congenita, Jackson-Lawler type	X	X			
KRT81	Monilethrix	X				
KRT83	Monilethrix	X				
KRT86	Monilethrix	X				
L1CAM	Hydrocephalus due to aqueductal stenosis; MASA syndrome; CRASH syndrome; Hydrocephalus with congenital idiopathic intestinal pseudoobstruction; Corpus callosum, partial agenesis of; Hydrocephalus with Hirschsprung disease	X			X	
L2HGDH	L-2-hydroxyglutaric aciduria	X			X	

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GENE	DISORDER(S)	CHILDHOOD ONSET	ADULT ONSET	CANCER PREDISPOSITION	CARRIER	ACMG*
LAMA2	Muscular dystrophy, congenital merosin-deficient; Muscular dystrophy, congenital, due to partial LAMA2 deficiency	X	X		X	
LAMA3	Epidermolysis bullosa, junctional, Herlitz type; Epidermolysis bullosa, generalized atrophic benign; Laryngoonychocutaneous syndrome	X			X	
LAMB2	Nephrotic syndrome, type 5, with or without ocular abnormalities; Pierson syndrome	X			X	
LAMB3	Epidermolysis bullosa, junctional, Herlitz type; Epidermolysis bullosa, junctional, non-Herlitz type	X			X	
LAMC2	Epidermolysis bullosa, junctional, Herlitz type; Epidermolysis bullosa, junctional, non-Herlitz type	X			X	
LAMP2	Danon disease	X	X			
LARGE	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6	X			X	
LBR	Pelger-Huet anomaly; Greenberg skeletal dysplasia; Reynolds syndrome	X			X	
LCAT	Norum disease; Fish-eye disease	X	X		X	
LDB3	Myopathy, myofibrillar, 4; Cardiomyopathy, dilated 1C; Left ventricular noncompaction 3, with or without dilated cardiomyopathy	X	X			
LDHA	Glycogen storage disease XI	X			X	
LDLR	Familial Hypercholesterolemia	X			X	X
LDLRAP1	Hypercholesterolemia, familial, autosomal recessive	X			X	
LEMD3	Osteopoikilosis; Buschke-Ollendorff syndrome; Melorheostosis with osteopoikilosis	X				
LEPR	Obesity, morbid, due to leptin receptor deficiency	X			X	
LEPRE1	Osteogenesis imperfecta, type VIII	X			X	
LFNG	Spondylocostal dysostosis, autosomal recessive 3	X			X	
LGI1	Epilepsy, familial temporal lobe, 1	X	X			
LHCGR	Precocious puberty, male; Leydig cell hypoplasia with pseudohermaphroditism; Leydig cell hypoplasia with hypergonadotropic hypogonadism; Luteinizing hormone resistance, female	X			X	
LHFPL5	Deafness, autosomal recessive 67	X			X	
LHX3	Pituitary hormone deficiency, combined, 3	X			X	
LIFR	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome	X			X	
LIG4	LIG4 syndrome; Severe combined immunodeficiency with sensitivity to ionizing radiation	X			X	
LIPA	Wolman disease; Cholesteryl ester storage disease	X			X	
LIPH	Hypotrichosis, localized, autosomal recessive 2; Woolly hair, autosomal recessive 2 with or without hypotrichosis	X			X	
LITAF	Charcot-Marie-Tooth disease, type 1C	X	X			
LMNA	Familial hypertrophic cardiomyopathy; Dilated cardiomyopathy; Emery-Dreifuss muscular dystrophy 2	X	X		X	X
LMX1B	Nail-patella syndrome	X				
LOR	Vohwinkel syndrome with ichthyosis	X				
LPAR6	Hypotrichosis, localized, autosomal recessive, 3; Woolly hair, autosomal recessive 1	X			X	

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GENE	DISORDER(S)	CHILDHOOD ONSET	ADULT ONSET	CANCER PREDISPOSITION	CARRIER	ACMG*
LPIN2	Majeed syndrome	X			X	
LPL	Lipoprotein lipase deficiency; Combined hyperlipidemia, familial	X			X	
LRP2	Donnai-Barrow syndrome	X			X	
LRP5	Osteoporosis-pseudoglioma syndrome; Osteopetrosis, AD type I; Hyperostosis, endosteal; van Buchem disease, type 2; Osteosclerosis; Exudative vitreoretinopathy 4	X	X		X	
LRPPRC	Leigh syndrome, French-Canadian type	X			X	
LRRK2	Parkinson disease-8		X			
LYST	Chediak-Higashi syndrome	X			X	
M33	46XY gonadal dysgenesis, complete, CBS2-related; Mental retardation, autosomal recessive 7	X			X	
MAN2B1	Mannosidosis, alpha-, types I and II	X			X	
MANBA	Mannosidosis, beta	X				
MAP2K1	Cardiofaciocutaneous syndrome	X				
MAP2K2	Cardiofaciocutaneous syndrome	X				
MAPT	Dementia, frontotemporal, with or without parkinsonism; Pick disease; Supranuclear palsy, progressive; Supranuclear palsy, progressive atypical; Tauopathy and respiratory failure		X			
MATN3	Epiphyseal dysplasia, multiple, 5; Spondyloepimetaphyseal dysplasia	X				
MATR3	Myopathy, distal 2		X			
MBTPS2	Ichthyosis follicularis, atrichia, and photophobia syndrome	X			X	
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency	X			X	
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency	X			X	
MCEE	Methylmalonyl-CoA epimerase deficiency	X			X	
MCOLN1	Mucolipidosis IV	X			X	
MECP2	Rett syndrome; Mental retardation, X-linked, syndromic 13; Rett syndrome, preserved speech variant; Encephalopathy, neonatal severe; Angelman syndrome; Mental retardation, X-linked, Lubs type	X			X	
MED12	Opitz-Kaveggia syndrome; Lujan-Fryns syndrome	X			X	
MEFV	Familial Mediterranean fever, AR; Familial Mediterranean fever, AD	X			X	
MEN1	Multiple endocrine neoplasia, type 1	X	X	X		X
MESP2	Spondylocostal dysostosis, autosomal recessive 2	X			X	
MFN2	Charcot-Marie-Tooth disease, type 2A2; Hereditary motor and sensory neuropathy VI	X	X		X	
MFSD8	Ceroid lipofuscinosis, neuronal, 7	X			X	
MGAT2	Congenital disorder of glycosylation, type Iia	X			X	
MID1	Opitz G syndrome, type I	X			X	
MITF	Waardenburg syndrome, type 2A; Waardenburg syndrome/ocular albinism, digenic; Tietz albinism-deafness syndrome	X				
MKKS	McKusick-Kaufman syndrome; Bardet-Biedl syndrome 6	X			X	
MKS1	Meckel syndrome, type 1; Bardet-Biedl syndrome 13	X			X	
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts	X			X	

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GENE	DISORDER(S)	CHILDHOOD ONSET	ADULT ONSET	CANCER PREDISPOSITION	CARRIER	ACMG*
MLH1	Hereditary nonpolyposis colorectal cancer (Lynch Syndrome)		X	X		X
MLPH	Griscelli syndrome, type 3	X			X	
MLYCD	Malonyl-CoA decarboxylase deficiency	X			X	
MMAA	Methylmalonic aciduria, vitamin B12-responsive	X			X	
MMAB	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type	X			X	
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type	X			X	
MMADHC	Homocystinuria, cblD type, variant 1; Methylmalonicaciduria, cblD type, variant 2; Methylmalonic aciduria and homocystinuria, cblD type	X			X	
MMP2	Torg-Winchester syndrome	X			X	
MMP20	Amelogenesis imperfecta, type IIA2	X			X	
MOCS1	Molybdenum cofactor deficiency, type A	X			X	
MOCS2	Molybdenum cofactor deficiency, type B	X			X	
MOGS	Congenital disorder of glycosylation, type IIb	X			X	
MPDU1	Congenital disorder of glycosylation, type If	X			X	
MPI	Congenital disorder of glycosylation, type Ib	X			X	
MPL	Thrombocytopenia, congenital amegakaryocytic; Thrombocythemia, essential	X			X	
MPV17	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type)	X			X	
MPZ	Charcot-Marie-Tooth disease, type 1B; Dejerine-Sottas syndrome; Neuropathy, congenital hypomyelinating; Charcot-Marie-Tooth disease, type 2J; Roussy-Levy syndrome; Charcot-Marie-Tooth disease, type 2I; Charcot-Marie-Tooth disease, dominant intermediate 3	X	X		X	
MRPS16	Combined oxidative phosphorylation deficiency 2	X			X	
MRPS22	Combined oxidative phosphorylation deficiency 5	X			X	
MSH2	Hereditary nonpolyposis colorectal cancer (Lynch Syndrome)		X	X		X
MSH6	Hereditary nonpolyposis colorectal cancer (Lynch Syndrome)		X	X		X
MSX1	Tooth agenesis, selective, 1, with or without orofacial cleft; Witkop syndrome; Orofacial cleft 5	X				
MSX2	Craniosynostosis, type 2; Parietal foramina 1; Parietal foramina with cleidocranial dysplasia	X				
MTHFR	Homocystinuria due to MTHFR deficiency	X	X		X	
MTM1	Myotubular myopathy, X-linked	X			X	
MTMR2	Charcot-Marie-Tooth disease, type 4B1		X		X	
MTR	Methylcobalamin deficiency, cblG type	X			X	
MTRR	Homocystinuria-megaloblastic anemia, cbl E type	X			X	
MTTP	Abetalipoproteinemia	X	X		X	
MUT	Methylmalonic aciduria, mut(0) type	X			X	
MUTYH	MYH-Associated polyposis; Adenomas, multiple colorectal, FAP type 2; Colorectal adenomatous polyposis, autosomal recessive with pilomatricomas		X	X	X	X
MVK	Mevalonic aciduria; Hyper-IgD syndrome; Porokeratosis 3, disseminated superficial actinic	X			X	

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GENE	DISORDER(S)	CHILDHOOD ONSET	ADULT ONSET	CANCER PREDISPOSITION	CARRIER	ACMG*
MYBPC3	Familal hypertrophic cardiomyopathy; Dilated cardiomyopathy	X	X			X
MYCN	Feingold syndrome; Microcephaly and digital abnormalities with normal intelligence	X				
MYD88	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency	X			X	
MYH3	Arthrogryposis, distal, type 2A; Arthrogryposis, distal, type 2B	X				
MYH6	Cardiomyopathy, familial hypertrophic, 14; Atrial septal defect 3; Cardiomyopathy, dilated, 1EE	X	X			
MYH7	Familial hypertrophic cardiomyopathy; Dilated cardiomyopathy	X	X			X
MYH9	May-Hegglin anomaly; Fechtner syndrome; Sebastian syndrome; Deafness, autosomal dominant 17; Epstein syndrome; Macrothrombocytopenia and progressive sensorineural deafness	X				
MYH11	Marfan syndrome; Loeys-Dietz syndrome; and Familial thoracic aortic aneurysms and dissections		X			X
MYL2	Familial hypertrophic cardiomyopathy; Dilated cardiomyopathy					X
MYL3	Familial hypertrophic cardiomyopathy; Dilated cardiomyopathy	X	X		X	X
MYLK	Marfan syndrome; Loeys-Dietz syndrome; Familial thoracic aortic aneurysms and dissections	X	X			
MYO15A	Deafness, autosomal recessive 3	X			X	
MYO1A	Deafness, autosomal dominant 48	X				
MYO5A	Griscelli syndrome, type 1	X			X	
MYO6	Deafness, autosomal dominant 22; Deafness, autosomal recessive 37; Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy	X			X	
MYO7A	Usher syndrome, type 1B; Deafness, autosomal recessive 2, neurosensory; Deafness, autosomal dominant 11, neurosensory	X			X	
MYOC	Glaucoma 1A, primary open angle, juvenile-onset; Glaucoma 1A, primary open angle, recessive; Glaucoma, early-onset, digenic		X			
MYOT	Muscular dystrophy, limb-girdle, type 1A; Myotilinopathy; Myopathy, spheroid body		X			
NAGA	Schindler disease, type I; Kanzaki disease; Schindler disease, type III	X	X		X	
NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo B)	X			X	
NAGS	N-acetylglutamate synthase deficiency	X			X	
NBN	Nijmegen breakage syndrome; Leukemia, acute lymphoblastic	X		X	X	
NCF1	Chronic granulomatous disease due to deficiency of NCF-1	X			X	
NCF2	Chronic granulomatous disease due to deficiency of NCF-2	X			X	
NDP	Norrie disease; Exudative vitreoretinopathy, X-linked	X			X	
NDRG1	Charcot-Marie-Tooth disease, type 4D	X			X	
NDUFA1	Mitochondrial complex I deficiency	X	X			
NDUFAF2	Mitochondrial complex I deficiency; Leigh syndrome	X			X	
NDUFAF4	Mitochondrial complex I deficiency	X			X	
NDUFS3	Leigh syndrome; Complex I deficiency	X			X	

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NDUFS4	Leigh syndrome; Mitochondrial complex I deficiency	X			X	
NDUFS6	Complex I, mitochondrial respiratory chain, deficiency of	X			X	
NDUFS7	Leigh syndrome; Complex I deficiency	X			X	
NDUFS8	Leigh syndrome; Complex I deficiency	X			X	
NDUFV1	Mitochondrial complex I deficiency	X			X	
NEB	Nemaline myopathy 2, autosomal recessive	X	X		X	
NEFL	Charcot-Marie-Tooth disease, type 2E; Charcot-Marie-Tooth disease, type 1F	X	X			
NEM1	Myopathy, nemaline, 3; Myopathy, actin, congenital, with excess of thin myofilaments; Myopathy, actin, congenital, with cores; Myopathy, congenital, with fiber-type disproportion 1; Nemaline myopathy 1, autosomal dominant	X	X		X	
NEU1	Sialidosis, type I; Sialidosis, type II	X			X	
NEUROG3	Diarrhea 4, malabsorptive, congenital	X			X	
NF1	Neurofibromatosis, type 1; Leukemia, juvenile myelomonocytic; Melanoma, desmoplastic neurotrophic; Neurofibromatosis, familial spinal; Neurofibromatosis-Noonan syndrome; Watson syndrome	X		X	X	
NF2	Neurofibromatosis, type 2	X	X	X	X	X
NFKBIA	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency	X			X	
NGF	Neuropathy, hereditary sensory and autonomic, type V	X			X	
NHEJ1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	X			X	
NHLRC1	Epilepsy, progressive myoclonic 2B (Lafora)	X			X	
NHS	Nance-Horan syndrome; Cataract, congenital, X-linked	X			X	
NIPBL	Cornelia de Lange syndrome 1	X				
NKH	Glycine encephalopathy	X			X	
NLGN4X	Mental retardation, X-linked	X			X	
NLRP12	Familial cold autoinflammatory syndrome 2	X				
NLRP3	Cold-induced autoinflammatory syndrome, familial; Muckle-Wells syndrome; CINCA syndrome	X				
NOG	Symphalangism, proximal; Synostoses syndrome, multiple, 1; Tarsal-carpal coalition syndrome; Stapes ankylosis with broad thumb and toes; Brachydactyly, type B2	X				
NOTCH2	Alagille syndrome 2	X				
NOTCH3	Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy		X			
NPC1	Niemann-Pick disease type C1; Niemann-Pick disease, type D	X	X		X	
NPC2	Niemann-pick disease, type C2	X	X		X	
NPHP1	Nephronophthisis 1, juvenile; Senior-Loken syndrome-1; Joubert syndrome 4	X			X	
NPHP3	Nephronophthisis 3; Renal-hepatic-pancreatic dysplasia; Meckel syndrome 7	X			X	
NPHP4	Nephronophthisis 4; Senior-Loken syndrome 4	X			X	
NPHS1	Nephrotic syndrome, type 1	X			X	
NPHS2	Nephrotic syndrome, type 2	X			X	
NR0B1	Adrenal hypoplasia, congenital, with hypogonadotropic hypogonadism; Dosage-sensitive sex reversal	X	X		X	

*ACMG minimum list genes

GENE	DISORDER(S)	CHILDHOOD ONSET	ADULT ONSET	CANCER PREDISPOSITION	CARRIER	ACMG*
NR3C2	Pseudohypoaldosteronism type I, autosomal dominant; Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy	X				
NR5A1	46XY gonadal dysgenesis, complete or partial, with or without adrenal failure; Premature ovarian failure 7; Adrenocortical insufficiency	X	X			
NRAS	Colorectal cancer; Thyroid carcinoma, follicular; Autoimmune lymphoproliferative syndrome type IV; Noonan syndrome 6	X		X		
NSD1	Sotos syndrome; Weaver syndrome; Beckwith-Wiedemann syndrome	X				
NSDHL	CHILD syndrome; CK syndrome	X				
NSUN2	Mental retardation, autosomal recessive 5	X			X	
NTRK1	Insensitivity to pain, congenital, with anhidrosis; Medullary thyroid carcinoma, familial	X			X	
NUP62	Striatonigral degeneration, infantile	X			X	
NYX	Night blindness, congenital stationary, type 1	X			X	
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia	X			X	
OCA2	Albinism, oculocutaneous, type II; Albinism, brown oculocutaneous	X			X	
OCRL	Lowe syndrome; Dent disease 2	X			X	
OFD1	Oral-facial-digital syndrome 1; Simpson-Golabi-Behmel syndrome, type 2; Joubert syndrome 10	X			X	
OPA1	Optic atrophy 1; Optic atrophy and deafness	X	X			
OPA3	3-methylglutaconic aciduria, type III; Optic atrophy and cataract	X			X	
OPHN1	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance	X			X	
ORAI1	Immune dysfunction with T-cell inactivation due to calcium entry defect 1	X			X	
OSTM1	Osteopetrosis, autosomal recessive 5	X			X	
OTC	Ornithine transcarbamylase deficiency	X	X		X	X
OTOF	Deafness, autosomal recessive 9; Auditory neuropathy, autosomal recessive, 1	X			X	
OTX2	Microphthalmia, syndromic 5	X				
OXCT1	Succinyl CoA:3-oxoacid CoA transferase deficiency	X			X	
PABPN1	Oculopharyngeal muscular dystrophy		X		X	
PAFAH1B1	Lissencephaly-1; Subcortical laminar heterotopia	X				
PAH	Phenylketonuria	X			X	
PAK3	Mental retardation, X-linked 30	X			X	
PALB2	Fanconi anemia, complementation group N	X	X	X	X	
PANK2	Neurodegeneration with brain iron accumulation 1 (Hallervorden-Spatz); HARP syndrome	X	X		X	
PARK2	Parkinson disease, juvenile, type 2		X		X	
PARK7	Parkinson disease 7, autosomal recessive early-onset; Amyotrophic lateral sclerosis-Parkinsonism/dementia complex 2		X		X	
PAX2	Optic nerve coloboma with renal disease; Renal hypoplasia, isolated	X				

*ACMG minimum list genes

GENE	DISORDER(S)	CHILDHOOD ONSET	ADULT ONSET	CANCER PREDISPOSITION	CARRIER	ACMG*
PAX3	Waardenburg syndrome, type 1; Waardenburg syndrome, type 3; Craniofacial-deafness-hand syndrome; Rhabdomyosarcoma 2, alveolar	X			X	
PAX6	Aniridia; Peters anomaly; Cataract with late-onset corneal dystrophy; Keratitis; Foveal hyperplasia; Morning glory disc anomaly; Optic nerve hypoplasia; Coloboma, ocular; Coloboma of optic nerve; Gillespie syndrome	X				
PAX8	Hypothyroidism, congenital, due to thyroid dysgenesis orhypoplasia	X			X	
PC	Pyruvate carboxylase deficiency	X			X	
PCBD1	Hyperphenylalaninemia, BH4-deficient, D	X				
PCCA	Propionic acidemia	X			X	
PCCB	Propionic acidemia	X			X	
PCDH15	Usher syndrome, type 1F; Deafness, autosomal recessive 23; Usher syndrome, type 1D/F digenic	X			X	
PCDH19	Epileptic encephalopathy, early infantile, 9	X				
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II	X			X	
PCSK9	Familial hypercholesterolemia	X	X			X
PDCD10	Cerebral cavernous malformations 3	X	X			
PDE6B	Night blindness, congenital stationary, autosomal dominant 2; Retinitis pigmentosa-40	X			X	
PDHA1	Pyruvate dehydrogenase deficiency; Leigh syndrome,X-linked	X			X	
PDHB	Pyruvate dehydrogenase E1-beta deficiency	X			X	
PDHX	Lacticacidemia due to PDX1 deficiency	X			X	
PDSS1	Coenzyme Q10 deficiency	X			X	
PDSS2	Coenzyme Q10 deficiency	X			X	
PDX1	Pancreatic agenesis; MODY, type IV; Lactic acidemia due to PDX1 deficiency	X			X	
PEPD	Prolidase deficiency	X			X	
PEX1	Zellweger syndrome-1; Adrenoleukodystrophy, neonatal; Refsum disease, infantile	X			X	
PEX10	Zellweger syndrome; Adrenoleukodystrophy, neonatal	X			X	
PEX12	Peroxisome biogenesis disorder 3B	X			X	
PEX13	Zellweger syndrome; Adrenoleukodystrophy, neonatal	X			X	
PEX26	Adrenoleukodystrophy, neonatal; Refsum disease, infantile form; Zellweger syndrome	X			X	
PEX5	Adrenoleukodystrophy, neonatal; Zellweger syndrome	X			X	
PEX7	Rhizomelic chondrodysplasia punctata, type 1; Refsum disease	X	X		X	
PGAM2	Glycogen storage disease X	X			X	
PGK1	Phosphoglycerate kinase 1 deficiency	X	X		X	
PHEX	Hypophosphatemic rickets, X-linked dominant	X			X	
PHK	Glycogen storage disease, type IXa1; Glycogen storage disease, type IXa2	X			X	
PHKA1	Muscle glycogenosis	X			X	
PHKA2	Glycogen storage disease, type IXa1; Glycogen storage disease, type IXa2	X			X	
PHKB	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive	X			X	

*ACMG minimum list genes

GENE	DISORDER(S)	CHILDHOOD ONSET	ADULT ONSET	CANCER PREDISPOSITION	CARRIER	ACMG*
PHKG2	Glycogen storage disease IXc; Cirrhosis due to liver phosphorylase kinase deficiency	X			X	
PHOX2A	Fibrosis of extraocular muscles, congenital, 2	X			X	
PHOX2B	Central hypoventilation syndrome, congenital; Hirschsprung disease, short-segment	X	X			
PHYH	Refsum disease	X	X		X	
PINK1	Parkinson disease 6, early onset	X	X		X	
PITX2	Axenfeld-Rieger syndrome, type 1; Iridogoniodysgenesis, type 2; Ring dermoid of cornea; Peters anomaly	X				
PKD1	Polycystic kidney disease, adult type I		X			
PKD2	Polycystic kidney disease 2		X			
PKHD1	Polycystic kidney and hepatic disease; Autosomal recessive polycystic kidney disease	X				
PKLR	Pyruvate kinase deficiency; Adenosine triphosphate, elevated, of erythrocytes	X			X	
PKP2	Arrhythmogenic right venticular cardiomyopathy	X	X			X
PLA2G6	Infantile neuroaxonal dystrophy 1; Neurodegeneration with brain iron accumulation 2B; Karak syndrome; Parkinson disease 14	X			X	
PLCE1	Nephrotic syndrome, type 3	X			X	
PLEC	Muscular dystrophy with epidermolysis bullosa simplex; Epidermolysis bullosa simplex, Ogna type; Epidermolysis bullosa simplex with pyloric atresia; Muscular dystrophy, limb-girdle, type 2Q	X			X	
PLEKHG5	Charcot-Marie-Tooth disease, recessive intermediate C; Spinal muscular atrophy, distal, autosomal recessive, 4	X	X		X	
PLG	Plasminogen Tochigi disease; Thrombophilia, dysplasminogenemic; Plasminogen deficiency, types I and II; Conjunctivitis, ligneous	X	X		X	
PLOD1	Ehlers-Danlos syndrome, type VI; Nevo syndrome	X			X	
PLP1	Pelizaeus-Merzbacher disease; Spastic paraplegia-2	X			X	
PMM2	Congenital disorder of glycosylation, type Ia	X			X	
PMP22	Charcot-Marie-Tooth disease, type 1A; Dejerine-Sottasdisease; Neuropathy, recurrent, with pressure palsies; Charcot-Marie-Tooth disease, type 1E; Roussy-Levy syndrome	X	X			
PMS2	Hereditary nonpolyposis colorectal cancer (Lynch Syndrome)	X	X	X	X	X
PNKD	Paroxysmal nonkinesigenic dyskinesia	X				
PNP	Immunodeficiency due to purine nucleoside phosphorylase deficiency	X			X	
PNPLA2	Neutral lipid storage disease with myopathy		X		X	
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency	X			X	
POLG	Progressive external ophthalmoplegia, autosomal recessive; Progressive external ophthalmoplegia, autosomal dominant; Mitochondrial DNA depletion syndrome 4B (MNGIE type); Mitochondrial DNA depletion syndrome 4A (Alpers type); Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE)	X	X		X	
POLH	Xeroderma pigmentosum, variant type	X			X	
POLR1C	Treacher Collins syndrome 3	X			X	
POLR1D	Treacher Collins syndrome 2	X				

*ACMG minimum list genes

GENE	DISORDER(S)	CHILDHOOD ONSET	ADULT ONSET	CANCER PREDISPOSITION	CARRIER	ACMG*
POMGNT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3	X			X	
POMT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1	X			X	
POMT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eyeanomalies), type A, 2; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2; Musculardystrophy-dystroglycanopathy (limb-girdle), type C, 2	X			X	
POR	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis; Disordered steroidogenesis due to cytochrome P450 oxidoreductase	X			X	
PORCN	Focal dermal hypoplasia	X				
POU1F1	Pituitary hormone deficiency, combined, 1	X			X	
POU3F4	Deafness, X-linked 2	X			X	
PPM2C	Pyruvate dehydrogenase phosphatase deficiency	X			X	
PPOX	Porphyria variegata		X			
PPT1	Ceroid lipofuscinosis, neuronal-1, infantile; Ceroidlipofuscinosis, neuronal, variant juvenile type, with granular osmiophilic deposits	X	X		X	
PQBP1	Renpenning syndrome; Golabi-Ito-Hall syndrome	X			X	
PRF1	Hemophagocytic lymphohistiocytosis, familial, 2; Lymphoma, non-Hodgkin	X	X		X	
PRICKLE1	Epilepsy, progressive myoclonic 1B	X			X	
PRKAG2	Familial hypertrophic cardiomyopathy; Dilated cardiomyopathy; Wolff-Parkinson-White syndrome	X	X			X
PRKAR1A	Carney complex, type 1; Myxoma, intracardiac; Thyroid carcinoma, papillary; Pigmented adrenocortical disease, primary, 1	X	X			
PRNP	Creutzfeldt-Jakob disease; Gerstmann-Straussler disease; Insomnia, fatal familial; Prion disease with protracted course; Huntington disease-like 1		X			
PROC	Thrombophilia due to protein C deficiency, autosomal dominant; Thrombophilia due to protein C deficiency, autosomal recessive	X	X		X	
PRODH	Hyperprolinemia, type I	X			X	
PROK2	Kallmann syndrome 4; Hypogonadism, hypogonadotropic	X	X		X	
PROKR2	Kallmann syndrome 3	X	X		X	
PROP1	Pituitary hormone deficiency, combined, 2	X			X	
PROS1	Thrombophilia due to protein S deficiency	X	X		X	
PRPH2	Retinitis pigmentosa-7; Retinitis punctata albescens; Macular dystrophy, patterned; Macular dystrophy, vitelliform; Foveomacular dystrophy, adult-onset, with choroidal neovascularization; Macular dystrophy; Retinitis pigmentosa, digenic; Choriodal dystrophy, central areolar 2		X			
PRPS1	Gout, PRPS-related; Phosphoribosylpyrophosphate synthetase superactivity; Charcot-Marie-Tooth disease, X-linked recessive, 5; Arts syndrome; Deafness, X-linked 1	X	X		X	
PRSS1	Trypsinogen deficiency; Pancreatitis, hereditary	X	X			

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GENE	DISORDER(S)	CHILDHOOD ONSET	ADULT ONSET	CANCER PREDISPOSITION	CARRIER	ACMG*
PRSS12	Mental retardation, autosomal recessive 1	X			X	
PRX	Dejerine-Sottas neuropathy, autosomal recessive; Charcot-Marie-Tooth disease, type 4F	X	X		X	
PSAP	Metachromatic leukodystrophy due to SAP-b deficiency; Gaucher disease, atypical; Combined SAP deficiency; Krabbe disease, atypical	X	X		X	
PSEN1	Alzheimer disease, type 3; Alzheimer disease, type 3, with spastic paraparesis and unusual plaques; Alzheimer disease, type 3, with spastic paraparesis and apraxia; Dementia, frontotemporal; Pick disease; Cardiomyopathy, dilated, 1U; Acne inversa, familial, 3		X			
PSEN2	Alzheimer disease-4; Cardiomyopathy, dilated, 1V		X			
PTCH1	Basal cell nevus syndrome; Holoprosencephaly-7	X				
PTEN	PTEN hamartoma tumor syndrome	X	X	X		X
PTH1R	Metaphyseal chondrodysplasia, Murk Jansen type; Chondrodysplasia, Blomstrand type; Eiken syndrome; Failure of tooth eruption, primary	X			X	
PTPN11	Noonan syndrome 1; LEOPARD syndrome 1; Leukemia, juvenile myelomonocytic; Metachondromatosis	X				
PTS	Hyperphenylalaninemia, BH4-deficient, A	X			X	
PYGL	Glycogen storage disease VI	X				
PYGM	McArdle disease; Glycogen Storage Disease Type V	X				
QDPR	Hyperphenylalaninemia, BH4-deficient, C	X			X	
RAB23	Carpenter syndrome	X			X	
RAB27A	Griscelli syndrome, type 2	X			X	
RAB39B	Mental retardation, X-linked-72	X			X	
RAB3GAP1	Warburg micro syndrome 1	X			X	
RAB3GAP2	Martsolf syndrome	X			X	
RAB7A	Charcot-Marie-Tooth disease, type 2B	X	X			
RAD50	Nijmegen breakage syndrome-like disorder	X		X	X	
RAF1	Noonan syndrome 5; LEOPARD syndrome 2	X				
RAG1	Severe combined immunodeficiency, B cell-negative; Omenn syndrome; Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity; Combined cellular and humoral immune defects with granulomas	X			X	
RAG2	Severe combined immunodeficiency, B cell-negative; Omenn syndrome; Combined cellular and humoral immune defects with granulomas	X			X	
RAI1	Smith-Magenis syndrome	X				
RAPSN	Myasthenic syndrome, congenital, associated with acetylcholinereceptor deficiency; Myasthenic syndrome, congenital, associated with facial dysmorphism and acetylcholine receptor deficiency; Fetal akinesia deformation sequence	X			X	
RARS2	Pontocerebellar hypoplasia, type 6	X			X	
RASA1	Parkes Weber syndrome; Capillary malformation-arteriovenous malformation	X				
RB1	Retinoblastoma	X		X		X
RECQL4	Rothmund-Thomson syndrome; RAPADILINO syndrome; Baller-Gerold syndrome	X			X	
RELN	Lissencephaly syndrome, Norman-Roberts type	X			X	

*ACMG minimum list genes

GENE	DISORDER(S)	CHILDHOOD ONSET	ADULT ONSET	CANCER PREDISPOSITION	CARRIER	ACMG*
REN	Renal tubular dysgenesis; Hyperuricemic nephropathy, familial juvenile 2	X	X		X	
RET	Multiple endocrine neoplasia, type 2; Familial medullary thyroid cancer (FMTC)	X	X	X		X
RFT1	Congenital disorder of glycosylation, type In	X			X	
RHO	Retinitis pigmentosa 4, autosomal dominant or recessive; Night blindness, congenital stationery, autosomal dominant 1; Retinitis punctata albescens	X			X	
RMRP	Cartilage-hair hypoplasia; Metaphyseal dysplasia without hypotrichosis; Anauxetic dysplasia	X			X	
RNASEH2A	Aicardi-Goutieres syndrome 4	X			X	
RNASEH2B	Aicardi-Goutieres syndrome 2	X			X	
RNASEH2C	Aicardi-Goutieres syndrome 3	X			X	
ROBO3	Gaze palsy, horizontal, with progressive scoliosis	X			X	
ROR2	Brachydactyly, type B1; Robinow syndrome, autosomal recessive	X			X	
RP2	Retinitis pigmentosa-2	X			X	
RP7	Retinitis pigmentosa-7; Retinitis punctata albescens; Macular dystrophy, patterned; Macular dystrophy, vitelliform; Foveomacular dystrophy, adult-onset, with choroidal neovascularization; Macular dystrophy; Retinitis pigmentosa, digenic; Choriodal dystrophy, central areolar 2; Retinitis pigmentosa-7, digenic	X	X			
RPE65	Leber congenital amaurosis 2; Retinitis pigmentosa-20	X			X	
RPGR	Retinitis pigmentosa-3; Cone-rod dystrophy-1; Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness; Macular degeneration, X-linked atrophic	X			X	
RPGRIP1L	Joubert syndrome 7; Meckel syndrome, type 5; COACH syndrome	X				
RPL11	Diamond-Blackfan anemia 7	X				
RPL35A	Diamond-Blackfan anemia 5	X				
RPL5	Diamond-Blackfan anemia 6	X				
RPS10	Diamond-Blackfan anemia 9	X				
RPS17	Diamond-Blackfan anemia 4	X				
RPS19	Diamond-Blackfan anemia 1	X				
RPS24	Diamond-blackfan anemia	X				
RPS26	Diamond-Blackfan anemia 10	X				
RPS6KA3	Coffin-Lowry syndrome; Mental retardation, X-linked nonspecific, type 19	X				
RPS7	Diamond-Blackfan anemia 8	X				
RRM2B	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy); Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 5; Mitochondrial DNA depletion syndrome 8B (MNGIE type); mtDNA depletion, encephalomyopathic form	X	X		X	
RS1	Retinoschisis; X-Linked Juvenile Retinoschisis	X			X	
RUNX2	Cleidocranial dysplasia; Dental anomalies, isolated	X				
RYR1	Malignant hyperthermia susceptibility; Central coredisease; Minicore myopathy with external ophthalmoplegia; Congenital neuromuscular disease	X	X		X	X
RYR2	Catecholaminergic polymorphic ventricular tachycardia	X				X

*ACMG minimum list genes

GENE	DISORDER(S)	CHILDHOOD ONSET	ADULT ONSET	CANCER PREDISPOSITION	CARRIER	ACMG*
SACS	Spastic ataxia, Charlevoix-Saguenay type	X			X	
SALL1	Townes-Brocks syndrome; Townes-Brocks branchiootorenal-like syndrome	X				
SALL4	Duane-radial ray syndrome; IVIC syndrome	X				
SAMHD1	Aicardi-Goutieres syndrome 5	X			X	
SAR1B	Chylomicron retention disease	X			X	
SBDS	Shwachman-Bodian-Diamond syndrome	X			X	
SBF2	Charcot-Marie-Tooth disease, type 4B2	X			X	
SC5DL	Lathosterolosis	X			X	
SCCMS	Myasthenic syndrome, slow-channel congenital; Myasthenic syndrome, fast-channel congenital; Multiple pterygium syndrome, lethal type; Myasthenic syndrome, slow-channel congenital; Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency; Myasthenic syndrome, slow-channel congenital; Myasthenic syndrome, fast-channel congenital; Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency	X	X		X	
SCKL4	Microcephaly, primary autosomal recessive, 6; Seckel syndrome 4; Microcephalic osteodysplastic primordial dwarfism, type II	X			X	
SCN1A	Epilepsy, generalized, with febrile seizures plus, type 2; Dravet syndrome; Migraine, familial hemiplegic, 3; Febrile convulsions, familial, 3A	X				
SCN4A	Hyperkalemic periodic paralysis, type 2; Paramyotonia congenita; Myotonia congenita, atypical, acetazolamide-responsive; Cramps, familial, potassium-aggravated; Myasthenic syndrome	X	X			
SCN5A	Romano-Ward Long QT syndromes types 1, 2 and 3; Brugada syndrome	X	X			X
SCN5A	Long QT syndrome-3; Brugada syndrome 1; Heartblock, progressive, type IA; Heart block, nonprogressive; Ventricular fibrillation, familial, 1; Sick sinus syndrome 1; Cardiomyopathy, dilated, 1E	X	X			
SCN9A	Erythermalgia, primary; Insensitivity to pain, channelopathy-associated; Paroxysmal extreme pain disorder; Febrile convulsions, familial, 3B	X	X		X	
SCNN1A	Pseudohypoaldosteronism, type I; Bronchiectasis with or without elevated sweat chloride 2	X			X	
SCNN1B	Liddle syndrome; Pseudohypoaldosteronism, type I; Bronchiectasis with or without elevated sweat chloride 1	X	X		X	
SCNN1G	Liddle syndrome; Pseudohypoaldosteronism, type I; Bronchiectasis with or without elevated sweat chloride 3	X	X		X	
SCO1	Hepatic failure, early onset, and neurologic disorder; Complex IV deficiency	X			X	
SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome coxidase deficiency; Complex IV deficiency	X			X	
SDHA	Leigh syndrome; Mitochondrial respiratory chain complex II deficiency; Cardiomyopathy, dilated, 1GG	X	X		X	
SDHAF2	Hereditary paraganglioma-pheochromocytoma syndrome	X		X		X
SDHB	Hereditary paraganglioma-pheochromocytoma syndrome	X		X		X
SDHC	Hereditary paraganglioma-pheochromocytoma syndrome	X		X		X
SDHD	Hereditary paraganglioma-pheochromocytoma syndrome	X		X		X
SEC23B	Anemia, dyserythropoietic congenital, type II	X	X		X	

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GENE	DISORDER(S)	CHILDHOOD ONSET	ADULT ONSET	CANCER PREDISPOSITION	CARRIER	ACMG*
SEPN1	Muscular dystrophy, rigid spine, 1	X			X	
SERPINA1	Emphysema due to AAT deficiency; Emphysema-cirrhosis, due to AAT deficiency; Hemorrhagic diathesis due to 'antithrombin' Pittsburgh; Alpha-1 antitrypsin deficiency	X	X		X	
SERPINA7	Thyroxine-binding globulin deficiency	X			X	
SERPINC1	Antithrombin III deficiency	X	X			
SERPING1	Angioedema, hereditary, types I and II; Complement component 4, partial deficiency of	X				
SERPINI1	Encephalopathy, familial, with neuroserpin inclusion bodies	X	X			
SETX	Ataxia-ocular apraxia-2; Amyotrophic lateral sclerosis 4, juvenile	X				
SFTPB	Surfactant metabolism dysfunction, pulmonary, 1	X			X	
SFTPC	Surfactant metabolism dysfunction, pulmonary, 2	X	X			
SGCA	Muscular dystrophy, limb-girdle, type 2D; Adhalinopathy, primary; Limb-girdle muscular dystrophy type 2D	X	X		X	
SGCB	Muscular dystrophy, limb-girdle, type 2E; Limb-girdle muscular dystrophy type 2E	X	X		X	
SGCD	Muscular dystrophy, limb-girdle, type 2F; Cardiomyopathy, dilated, 1L	X	X		X	
SGCE	Dystonia-11, myoclonic	X				
SGCG	Muscular dystrophy, limb-girdle, type 2C	X			X	
SGSH	Mucopolysaccharidisis type IIIA (Sanfilippo A)	X			X	
SH2D1A	Lymphoproliferative syndrome, X-linked	X	X		X	
SH3BP2	Cherubism	X				
SH3TC2	Charcot-Marie-Tooth disease, type 4C; Mononeuropathy of the median nerve, mild	X			X	
SHANK3	Autism, chromosome 22q13.3 deletion syndrome-related	X				
SHH	Holoprosencephaly-3; Solitary median maxillary central incisor; Coloboma, ocular; Microphthalmia with coloboma 5	X				
SHOX	Short stature, idiopathic familial; Leri-Weill dyschondrosteosis; Langer mesomelic dysplasia	X			X	
SHROOM4	Stocco dos Santos X-linked mental retardation syndrome	X			X	
SI	Sucrase-isomaltase deficiency, congenital	X			X	
SIL1	Marinesco-Sjogren syndrome; Marinesco-Sjogren syndrome	X			X	
SIX1	Brachiootic syndrome 3; Deafness, autosomal dominant 23	X				
SIX3	Holoprosencephaly-2	X				
SIX5	Branchiootorenal syndrome 2	X				
SLC12A1	Bartter syndrome, type 1	X			X	
SLC12A3	Gitelman syndrome	X			X	
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy; Andermann syndrome	X			X	
SLC16A2	Allan-Herndon-Dudley syndrome	X			X	
SLC17A5	Salla disease; Sialic acid storage disorder, infantile	X			X	
SLC19A2	Thiamine-responsive megaloblastic anemia syndrome	X			X	
SLC19A3	Basal ganglia disease, biotin-responsive; Encephalopathy, thiamine-responsive	X			X	

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GENE	DISORDER(S)	CHILDHOOD ONSET	ADULT ONSET	CANCER PREDISPOSITION	CARRIER	ACMG*
SLC1A3	Episodic ataxia, type 6	X	X			
SLC22A5	Carnitine deficiency, systemic primary	X			X	
SLC25A13	Citrullinemia, adult-onset type II; Citrullinemia,type II, neonatal-onset	X	X		X	
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	X	X		X	
SLC25A19	Microcephaly, Amish type	X			X	
SLC25A20	Carnitine-acylcarnitine translocase deficiency	X			X	
SLC25A22	Epileptic encephalopathy, early infantile, 3	X			X	
SLC25A4	Progressive external ophthalmoplegia with mitochondrial DNA deletions 3; Cardiomyopathy, familial hypertrophic		X			
SLC26A2	Diastrophic dysplasia; Atelosteogenesis II; Achondrogenesis Ib; Epiphyseal dysplasia, multiple, 4; Diastrophic dysplasia, broad bone-platyspondylic variant; De laChapelle dysplasia	X			X	
SLC26A4	Pendred syndrome; Enlarged vestibular aqueduct	X			X	
SLC2A1	GLUT1 deficiency syndrome 1; GLUT1 deficiency syndrome 2	X	X		X	
SLC2A10	Arterial tortuosity syndrome	X			X	
SLC33A1	Spastic paraplegia-42; Congenital cataracts, hearing loss, and neurodegeneration	X	X		X	
SLC35A1	Congenital disorder of glycosylation, type IIf	X			X	
SLC35C1	Congenital disorder of glycosylation, type IIc	X			X	
SLC35D1	Schneckenbecken dysplasia	X			X	
SLC37A4	Glycogen storage disease Ib; Glycogen storage disease Ic	X			X	
SLC3A1	Cystinuria	X	X		X	
SLC40A1	Hemochromatosis, type 4		X			
SLC45A2	Oculocutaneous albinism, type IV	X			X	
SLC46A1	Folate malabsorption, hereditary	X			X	
SLC4A11	Corneal endothelial dystrophy 2; Corneal endothelial dystrophy and perceptive deafness; Corneal dystrophy, Fuchs endothelial, 4	X			X	
SLC5A1	Glucose/galactose malabsorption	X			X	
SLC5A5	Thyroid dyshormonogenesis 1	X			X	
SLC6A8	Creatine deficiency syndrome, X-linked	X			X	
SLC7A7	Lysinuric protein intolerance	X			X	
SLC7A9	Cystinuria	X			X	
SLC9A6	Mental retardation, X-linked syndromic, Christianson type	X				
SLITRK1	Tourette syndrome; Trichotillomania	X	X			
SMAD3	Marfan syndrome; Loeys-Dietz syndrome; and Familial thoracic aortic aneurysms and dissections	X	X			X
SMAD4	Pancreatic cancer; Polyposis, juvenile intestinal; Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome	X	X	X		X
SMARCA4	Rhabdoid tumor predisposition syndrome 2, Mental retardation, autosomal dominant 16	X				
SMARCAL1	Schimke immunoosseous dysplasia	X			X	
SMARCAL1	Schimke immunoosseous dysplasia	X			X	
SMARCB1	Rhabdoid tumors; Rhabdoid predisposition syndrome 1, Mental retardation, autosomal dominant 15	X				

*ACMG minimum list genes

GENE	DISORDER(S)	CHILDHOOD ONSET	ADULT ONSET	CANCER PREDISPOSITION	CARRIER	ACMG*
SMC1A	Cornelia de Lange syndrome 2	X			X	
SMC3	Cornelia de Lange syndrome 3	X				
SMN1	Spinal muscular atrophy-1; Spinal muscular atrophy-2; Spinal muscular atrophy-3; Spinal muscular atrophy-4	X	X		X	
SMPD1	Niemann-Pick disease, type A; Niemann-Pick disease, type B	X			X	
SMS	Smith-Magenis syndrome	X				
SNAI2	Waardenburg syndrome, type 2D; Piebaldism	X			X	
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantarkeratoderma syndrome	X			X	
SNCA	Parkinson disease 4; Dementia, Lewy body; Parkinson disease 1		X			
SOD1	Amyotrophic lateral sclerosis, due to SOD1 deficiency		X		X	
SOS1	Fibromatosis, gingival; Noonan syndrome 4	X				
SOST	Sclerosteosis; Van Buchem disease	X			X	
SOX10	Waardenburg syndrome, type 4C; Waardenburg syndrome, type 2E, with or without neurologic involvement; PCWH syndrome	X				
SOX2	Microphthalmia, syndromic 3; Optic nerve hypoplasia and abnormalities of the central nervous system	X				
SOX3	Mental retardation, X-linked, with isolated growth hormone deficiency; Panhypopituitarism, X-linked	X			X	
SOX9	Campomelic dysplasia with autosomal sex reversal; Acampomelic campomelic dysplasia; Campomelic dysplasia	X				
SP110	Hepatic venoocclusive disease with immunodeficiency	X	X		X	
SPAST	Spastic paraplegia-4	X	X			
SPG11	Spastic paraplegia-11	X	X		X	
SPG20	Troyer syndrome	X			X	
SPG7	Spastic paraplegia-7		X		X	
SPH2	Elliptocytosis-3; Spherocytosis, type 2; Anemia, neonatal hemolytic, fatal and near-fatal; Spherocytosis, type 1	X				
SPR	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency	X			X	
SPRED1	Legius syndrome	X				
SQSTM1	Paget disease of bone		X			
SRD5A2	Pseudovaginal perineoscrotal hypospadias	X			X	
SRD5A3	Congenital disorder of glycosylation, type Iq; Autosomal mental retardation CDG 1Q	X			X	
SRY	46XY sex reversal 1; 46XX sex reversal 1	X				
ST3GAL3	Epileptic encephalopathy, early infantile, 15; Mental retardation, autosomal recessive 12	X			X	
ST3GAL5	Amish infantile epilepsy syndrome	X			X	
STAT1	Mycobacterial infection, atypical, familial disseminated; STAT1 deficiency, complete	X	X		X	
STAT3	Hyper-IgE recurrent infection syndrome	X				
STIM1	Immune dysfunction, with T-cell inactivation due to calcium entry defect 2	X	X		X	
STK11	Peutz-Jeghers Syndrome	X		X		X
STRA6	Microphthalmia, syndromic 9 (Matthew-Wood syndrome)	X			X	

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STRC	Deafness, autosomal recessive 16	X			X	
STX11	Hemophagocytic lymphohistiocytosis, familial, 4	X			X	
STXBP2	Hemophagocytic lymphohistiocytosis, familial, 5	X			X	
SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with methylmalonic aciduria); mtDNA depletion, encephalomyopathic form	X			X	
SUCLG1	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic typewith methylmalonic aciduria)	X			X	
SUOX	Sulfite oxidase deficiency	X			X	
SURF1	Leigh syndrome, due to COX deficiency	X			X	
SYNE1	Spinocerebellar ataxia, autosomal recessive 8; Emery-Dreifuss muscular dystrophy 4	X	X		X	
SYP	Mental retardation, X-linked, with or without epilepsy	X				
TAF1	Dystonia-Parkinsonism, X-linked		X		X	
TARDBP	Amyotrophic lateral sclerosis 10, with or without FTD; Frontotemporal lobar degeneration, TARDBP-related		X			
TAT	Tyrosinemia, type II	X			X	
TAZ	Endocardial fibroelastosis-2; Barth syndrome; Cardiomyopathy, dilated, 3A; Left ventricular noncompaction, X-linked	X			X	
TBCE	Kenny-Caffey syndrome-1; Hypoparathyroidism-retardation-dysmorphism syndrome	X			X	
TBP	Spinocerebellar ataxia 17		X			
TBX1	Conotruncal anomaly face syndrome; DiGeorge syndrome; Velocardiofacial syndrome	X			X	
TBX5	Holt-Oram syndrome	X				
TCF4	Pitt-Hopkins syndrome	X				
TCIRG1	Osteopetrosis, recessive 1	X			X	
TCOF1	Treacher Collins syndrome 1	X				
TCS1	Treacher Collins syndrome 1; Dyskeratosis congenita	X	X		X	
TECTA	Deafness, autosomal dominant 8/12; Deafness, autosomal recessive 21	X			X	
TEK	Venous malformations, multiple cutaneous and mucosal	X				
TERC	Dyskeratosis congenita, autosomal dominant; Aplasticanemia		X			
TERT	Dyskeratosis congenita		X			
TFAP2A	Branchiooculofacial syndrome	X				
TFAP2B	Char syndrome	X				
TFR2	Hemochromatosis, type 3		X		X	
TG	Thyroid dyshormonogenesis 3	X	X		X	
TGFB1	Camurati-Engelmann disease	X	X			
TGFBI	Corneal dystrophy, Groenouw type I; Corneal dystrophy, lattice type I; Corneal dystrophy, Reis-Bucklers type; Corneal dystrophy, Avellino type; Corneal dystrophy, lattice type IIIA; Corneal dystrophy, Thiel-Behnke type; Corneal dystrophy, epithelial basement membrane	X	X			
TGFBR1	Marfan syndrome; Loeys-Dietz syndrome; Familial thoracic aortic aneurysms and dissections	X				X
TGFBR2	Marfan syndrome; Loeys-Dietz syndrome; Familial thoracic aortic aneurysms and dissections	X				X

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TGIF1	Holoprosencephaly-4	X				
TGM1	Ichthyosis, lamellar, autosomal recessive; Ichthyosiform erythroderma, congenital; Self-healing collodion baby	X			X	
TH	Segawa syndrome, recessive	X			X	
THAP1	Dystonia 6, torsion	X	X			
THPO	Thrombocythemia, essential		X			
TIMM8A	Deafness, X-linked 1, progressive; Mohr-Tranebjaerg syndrome; Jensen syndrome	X			X	
TINF2	Dyskeratosis congenita, autosomal dominant; Revesz syndrome	X				
TK2	Mitochondrial DNA depletion syndrome 2 (myopathic type)	X			X	
TMC1	Deafness, autosomal recessive 7; Deafness, autosomal dominant 36	X			X	
TMEM216	Meckel syndrome, type 2; Joubert syndrome 2	X			X	
TMEM43	Arrhythmogenic right venticular cardiomyopathy; Emery-Dreifuss muscular dystrophy 7		X			X
TMEM67	Meckel syndrome, type 3; Joubert syndrome 6; COACH syndrome; Nephronophthisis 11	X			X	
TMIE	Deafness, autosomal recessive 6	X			X	
TMPRSS3	Deafness, autosomal recessive 8, childhood onset; Deafness, autosomal recessive 10, congenital	X			X	
TNFRSF11A	Osteolysis, familial expansile; Paget disease of bone; Osteopetrosis, autosomal recessive 7	X	X		X	
TNFRSF11B	Paget disease, juvenile	X			X	
TNFRSF13B	Immunoglobulin A deficiency 2; Immunodeficiency, commonvariable, 2	X			X	
TNFRSF1A	Periodic fever, familial	X	X			
TNNI2	Arthrogryposis multiplex congenita, distal, type 2B	X			X	
TNNI2	Arthrogryposis multiplex congenita, distal, type 2B	X				
TNNI3	Familial hypertrophic cardiomyopathy; Dilated cardiomyopathy	X	X		X	X
TNNT2	Familial hypertrophic cardiomyopathy; Dilated cardiomyopathy	X	X			X
TNNT3	Arthrogryposis multiplex congenita, distal, type 2B	X				
TNXB	Ehlers-Danlos due to tenascin X deficiency; Ehlers-Danlos syndrome, hypermobility type	X			X	
TOR1A	Dystonia-1, torsion; Dystonia, early-onset atypical, with myoclonic features	X	X			
TP53	Li-Fraumeni syndrome	X	X	X		X
TP63	Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome3; Split-hand/foot malformation 4; Hay-Wells syndrome; ADULT syndrome; Limb-mammary syndrome; Rapp-Hodgkin syndrome; Orofacial cleft 8	X				
TPM1	Familial hypertrophic cardiomyopathy; Dilated cardiomyopathy	X	X			X
TPM2	Arthrogryposis multiplex congenita, distal, type 1; Nemaline myopathy; Arthrogryposis, distal, type 2B	X				
TPM3	Nemaline myopathy 1, autosomal dominant	X			X	
TPO	Thyroid dyshormonogenesis 2A	X			X	
TPP1	Ceroid-lipofuscinosis, neuronal 2, classic late infantile	X			X	

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TRAPPC2	Spondyloepiphyseal dysplasia tarda	X			X	
TRAPPC9	Mental retardation, autosomal recessive 13	X			X	
TREM2	Nasu-Hakola disease	X			X	
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive; Chilblain lupus; Vasculopathy, retinal, with cerebral leukodystrophy	X	X		X	
TRIM37	Mulibrey nanism	X			X	
TRIOBP	Deafness, autosomal recessive 28	X			X	
TRPM1	Night blindness, congenital stationary, type IC	X			X	
TRPS1	Trichorhinophalangeal syndrome, type I; Trichorhinophalangeal syndrome, type III	X				
TRPV4	Brachyolmia type 3; Spondylometaphyseal dysplasia,Kozlowski type; Metatropic dysplasia; Hereditary motor and sensory neuropathy, type IIc; Scapuloperoneal spinalmuscular atrophy; Parastremmatic dwarfism; SED, Maroteaux type	X	X			
TSC1	Tuberous sclerosis complex	X		X		X
TSC2	Tuberous sclerosis complex	X		X		X
TSEN2	Pontocerebellar hypoplasia type 2B	X			X	
TSEN34	Pontocerebellar hypoplasia type 2C	X			X	
TSEN54	Pontocerebellar hypoplasia type 2A; Pontocerebellarhypoplasia type 4	X			X	
TSFM	Combined oxidative phosphorylation deficiency 3	X			X	
TSHB	Hypothryoidism, congenital, nongoitrous 4	X			X	
TSHR	Hypothyroidism, congenital, nongoitrous; Hyperthyroidism, nonautoimmune; Thyroid carcinoma with thyrotoxicosis; Hyperthyroidism, familial gestational	X	X		X	
TSPYL1	Sudden infant death with dysgenesis of the testes syndrome	X			X	
TTN	Cardiomyopathy, familial hypertrophic, 9; Cardiomyopathy,dilated, 1G; Tibial muscular dystrophy, tardive; Muscular dystrophy, limb-girdle, type 2J; Myopathy, proximal, with early respiratory muscle involvement; Myopathy, early-onset, with fatal cardiomyopathy	X	X		X	
TTPA	Ataxia with isolated vitamin E deficiency	X			X	
TTR	Amyloidosis, hereditary, transthyretin-related; Carpal tunnel syndrome, familial	X	X			
TUBA1A	Lissencephaly 3	X				
TUFM	Combined oxidative phosphorylation deficiency 4	X			X	
TUSC3	Mental retardation, autosomal recessive 7	X			X	
TWIST1	Saethre-Chotzen syndrome; Saethre-Chotzen syndrome with eyelid anomalies; Craniosynostosis, type 1	X				
TYK2	Tyrosine kinase 2 deficiency	X	X		X	
TYMP	Mitochondrial DNA depletion syndrome 1 (MNGIE type)		X		X	
TYR	Albinism, oculocutaneous, type IA; Waardenburgsyndrome/albinism, digenic; Albinism, oculocutaneous, type IB	X			X	
TYROBP	Nasu-Hakola disease		X		X	
TYRP1	Albinism, oculocutaneous, type III	X			X	
UBA1	Spinal muscular atrophy, X-linked 2, infantile	X			X	

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UBE2A	Mental retardation, X-linked syndromic	X			X	
UBE3A	Angelman syndrome	X				
UBR1	Johanson-Blizzard syndrome	X			X	
UGT1A1	Crigler-Najjar syndrome, type I; Crigler-Najjar syndrome, type II; Hyperbilirubinemia, familial transcient neonatal	X			X	
UMOD	Hyperuricemic nephropathy, familial juvenile 1; Medullary cystic kidney disease 2; Glomerulocystic kidney disease with hyperuricemia and isosthenuria	X				
UNC13D	Hemophagocytic lymphohistiocytosis, familial, 3	X			X	
UNC93B1	Herpes simplex encephalitis, susceptibility to, 1	X			X	
UPF3B	Mental retardation, X-linked, syndromic 14	X			X	
UQCRB	Mitochondrial complex III deficiency	X			X	
UQCRQ	Mitochondrial complex III deficiency	X			X	
UROD	Porphyria cutanea tarda; Porphyria, hepatoerythropoietic	X	X			
UROS	Porphyria, congenital erythropoietic	X			X	
USH1C	Usher syndrome, type 1C; Deafness, autosomal recessive 18	X			X	
USH1G	Usher syndrome, type 1G	X			X	
USH2A	Usher syndrome, type 2A; Retinitis pigmentosa-39	X			X	
USP9Y	Azoospermia		X			
VAPB	Amyotrophic lateral sclerosis 8; Spinal muscular atrophy, late-onset, Finkel type		X			
VCAN	Wagner syndrome 1	X				
VCP	Inclusion body myopathy with early-onset Paget disease andfrontotemporal dementia		X			
VDR	Rickets, vitamin D-resistant, type IIA	X	X		X	
VHL	Von Hippel Lindau syndrome	X	X	X	X	X
VIPAS39	Arthrogryposis, renal dysfunction, and cholestasis 2	X			X	
VLDLR	Cerebellar hypoplasia and mental retardation with or withoutquadrupedal locomotion 1	X			X	
VPS13A	Choreoacanthocytosis		X		X	
VPS13B	Cohen syndrome	X			X	
VPS33B	Arthrogryposis, renal dysfunction, and cholestasis 1	X			X	
VRK1	Pontocerebellar hypoplasia type 1	X			X	
VSX1	Keratoconus; Corneal dystrophy, hereditary polymorphous posterior		X			
VWF	von Willebrand disease, type 2A, 2B, 2M, and 2N; vonWillebrand disease, type 1; von Willibrand disease, type 3	X	X		X	
WAS	Wiskott-Aldrich syndrome; Thrombocytopenia, X-linked; Neutropenia, severe congenital, X-linked; Thrombocytopenia, X-linked, intermittent	X			X	
WDR35	Cranioectodermal dysplasia 2	X			X	
WFS1	Wolfram syndrome; Deafness, autosomal dominant 6/14/38; Wolfram-like syndrome, autosomal dominant	X	X		X	
WISP3	Arthropathy, progressive pseudorheumatoid, of childhood; Spondyloepiphyseal dysplasia tarda with progressive arthropathy	X			X	
WNK1	Pseudohypoaldosteronism, type IIC; Neuropathy, hereditary sensory and autonomic, type II	X			X	

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WNK4	Pseudohypoaldosteronism type II	X	X			
WNT3	Tetra-amelia, autosomal recessive	X			X	
WNT4	SERKAL syndrome; Mullerian aplasia and hyperandrogenism	X	X			
WNT7A	Ulna and fibula, absence of, with sever limb deficiency; Fuhrmann syndrome	X			X	
WRN	Werner syndrome	X			X	
WT1	WT1-related Wilms tumor	X		X		X
XIAP	Lymphoproliferative syndrome, X-linked, 2	X			X	
XK	McLeod syndrome; McLeod syndrome with neuroacanthosis		X		X	
XPA	Xeroderma pigmentosum, group A	X			X	
XPC	Xeroderma pigmentosum, group C	X			X	
YARS	Charcot-Marie-Tooth disease, dominant intermediate C; Myopathy, lactic acidosis, and sideroblastic anemia 2	X	X		X	
YARS2	Myopathy, lactic acidosis, and sideroblastic anemia 2	X			X	
ZAP70	Selective T-cell defect	X			X	
ZDHHC9	Mental retardation, X-linked, ZDHHC9-related	X			X	
ZEB2	Mowat-Wilson syndrome	X				
ZFP57	Diabetes mellitus, transient neonatal, 1	X			X	
ZIC2	Holoprosencephaly-5	X				
ZIC3	Heterotaxy, visceral, 1, X-linked; Congenital heart defects, nonsyndromic, 1, X-linked	X			X	
ZMPSTE24	Mandibuloacral dysplasia with type B lipodystrophy; Restrictive dermopathy, lethal	X			X	
ZNF41	Mental retardation, X-linked-89	X			X	
ZNF469	Brittle cornea syndrome (Ehlers-Danlos syndrome type VIB)	X			X	
ZNF674	Mental retardation, X-linked-92	X			X	
ZNF711	Mental retardation, X-linked 97	X			X	

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