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Carrier Screening Gene and Disease List

Gene	Disease	Inheritance	Gene	Disease	Inheritance
AAAS	Achalasia-addisonianism-alacrima syndrome	AR	BCS1L	GRACILE syndrome; Bjornstad syndrome; Leigh syndrome;Mitochondrial complex III deficiency, nuclear type 1	AR
ABCB11	Progressive familial intrahepatic cholestasis, type II	AR	BLM	Bloom syndrome	AR
ABCC6	Pseudoxanthoma elasticum	AR	BSND	Bartter syndrome, type IV	AR
ABCC8	Familial hyperinsulinemic hypoglycemia type 1	AR	BTD	Biotinidase deficiency	AR
ABCD1	Adrenoleukodystrophy (X-linked)	XL	CANT1	Desbuquois dysplasia, type I; Epiphyseal dysplasia, multiple, 7	AR
ACADM	Medium chain Acyl-CoA dehydrogenase deficiency	AR	CAPN3	Limb-girdle muscular dystrophy, type 2A	AR
ACADS	Short chain Acyl-CoA dehydrogenase deficiency	AR	CBS	Homocystinuria, CBS-related	AR
ACADSB	2-Methylbutyryl-CoA dehydrogenase deficiency	AR	CDH23	Usher syndrome, type ID; Deafness, autosomal recessive 12	AR
ACADVL	Very long chain Acyl-CoA dehydrogenase deficiency	AR	CEP290	Leber congenital amaurosis 10; Joubert syndrome 5; Meckel syndrome 4;Senior-Loken syndrome 6	AR
ACAT1	Beta-ketothiolase deficiency (Alpha-methylacetoacetic aciduria)	AR	CERKL	Retinitis pigmentosa 26	AR
ACOX1	Peroxisomal acyl-CoA oxidase deficiency	AR	CFTR	Cystic fibrosis;Congenital bilateral absence of vas deferens	AR

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ADA	Severe combined immunodeficiency due to ADA deficiency	AR	CHAT	Congenital myasthenic syndrome 6	AR
ADAMTS2	Ehlers Danlos syndrome, type VIIC	AR	CHM	Choroideremia	AR
ADAR	Aicardi-Goutieres syndrome 6	AR	CHRNE	Congenital myasthenic syndrome 4A; Congenital myasthenic syndrome 4B; Congenital myasthenic syndrome 4C	AR
ADGRG1	Bilateral frontoparietal polymicrogyria	AR	CLN3	Neuronal ceroid lipofuscinosis, CLN3-related	AR
AGA	Aspartylglycosaminuria	AR	CLN5	Neuronal ceroid lipofuscinosis, CLN5-related	AR
AGL	Glycogen storage disease, type III (a&b)	AR	CLN6	Neuronal ceroid lipofuscinosis, CLN6-related	AR
AGPS	Rhizomelic chondrodysplasia punctata, type III	AR	CLN8	Neuronal ceroid lipofuscinosis, CLN8-related; Northern epilepsy	AR
AGXT	Primary hyperoxaluria, type I	AR	CLRN1	Usher syndrome, type IIIA	AR
AIRE	Polyglandular autoimmune syndrome, type I (Autoimmune polyendocrinopathy syndrome type I, with or without reversible metaphyseal dysplasia)	AR	CNGA3	Achromatopsia 2, CNGA3-related	AR
ALDH3A2	Sjögren-Larsson syndrome	AR	CNGB3	Achromatopsia 3, CNGB3-related	AR
ALDH7A1	Pyridoxine-dependent epilepsy	AR	COL4A3	Alport syndrome	AR
ALDOB	Hereditary fructose intolerance	AR	COL4A5	Alport syndrome, X-linked	XL
ALG6	Congenital disorder of glycosylation, type Ic	AR	COL7A1	Dystrophic epidermolysis bullosa, autosomal recessive	AR
ALPL	Hypophosphatasia, autosomal recessive	AR	COLQ	Congenital myasthenic syndrome 5	AR

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AMT	Glycine encephalopathy, AMT-related	AR	CPT1A	Carnitine palmitoyltransferase IA deficiency	AR
AP1S1	MEDNIK syndrome	AR	CPT2	Carnitine palmitoyltransferase II deficiency	AR
AP3B1	Hermansky-Pudlak syndrome, type 2	AR	CRB1	Leber congenital amaurosis 8;Retinitis pigmentosa-12	AR
AR	Androgen insensitivity syndrome	XL	CTNS	Cystinosis, atypical nephropathic;Cystinosis, late-onset juvenile or adolescent nephropathic; Cystinosis, nephropathic; Cystinosis, ocular nonnephropathic	AR
ARSA	Metachromatic leukodystrophy	AR	CTSD	Neuronal ceroid lipofuscinosis 10	AR
ARSB	Mucopolysaccharidosis, type VI	AR	CTSF	Neuronal ceroid lipofuscinosis 13	AR
ASL	Argininosuccinic aciduria	AR	CTSK	Pycnodysostosis	AR
ASNS	Asparagine Synthetase deficiency	AR	CYP11B2	Cortisone methyl oxidase type II deficiency; Cortisone methyl oxidase type I deficiency	AR
ASPA	Canavan disease	AR	CYP17A1	Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	AR
ASS1	Citrullinemia, type I	AR	CYP19A1	Aromatase deficiency	AR
ATM	Ataxia-telangiectasia	AR	CYP1B1	Primary congenital glaucoma 3A	AR
ATP13A2	Kufor-Rakeb syndrome (KRS); Autosomal recessive spastic paraplegia-78 (SPG78)	AR	CYP21A2	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	AR
ATP6V1B1	Renal tubular acidosis and deafness, ATP6V1B1-related	AR	CYP27A1	Cerebrotendinous xanthomatosis	AR
ATP7B	Wilson disease	AR	CYP27B1	Vitamin D-dependent rickets, type I	AR

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BBS1	Bardet-Biedl syndrome 1	AR	DBT	Maple syrup urine disease, type II	AR
BBS2	Bardet-Biedl syndrome 2	AR	DCLRE1C	Omenn syndrome;Severe combined immunodeficiency, Athabascan type	AR
BBS4	Bardet-Biedl syndrome 4	AR	DHCR7	Smith-Lemli-Opitz syndrome	AR
BBS7	Bardet-Biedl syndrome 7	AR	DHDDS	Retinitis pigmentosa 59	AR
BBS9	Bardet-Biedl syndrome 9	AR	DKC1	Dyskeratosis congenita, X-linked	XL
BBS10	Bardet-Biedl syndrome 10	AR	DLD	Dihydrolipoamide dehydrogenase deficiency	AR
BBS12	Bardet-Biedl syndrome 12	AR	DMD	Duchenne/Becker muscular dystrophy	XL
BCHE	Pseudocholinesterase deficiency	AR	DNAH5	Primary ciliary dyskinesia type 3, DNAH5-related	AR
BCKDHA	Maple syrup urine disease, type Ia	AR	DNAI1	Primary ciliary dyskinesia type 1, DNAI1-related	AR
BCKDHB	Maple syrup urine disease, type Ib	AR	DNAI2	Primary ciliary dyskinesia type 9, DNAI2-related	AR
DNAJC5	Neuronal ceroid lipofuscinosis type 4	AR	GNPTG	Mucolipidosis III gamma	AR
DOK7	Fetal akinesia deformation sequence, DOK7-related; Congenital myasthenic syndrome, 10	AR	GNS	Mucopolysaccharidosis type IIID	AR
DPYD	Dihydropyrimidine dehydrogenase deficiency	AR	GORAB	Geroderma osteodysplastica	AR
DYSF	Limb-girdle muscular dystrophy, type 2B; Miyoshi myopathy and distal myopathy with anterior tibial onset	AR	GRHPR	Primary hyperoxaluria, type II	AR
EDA	Hypohidrotic ectodermal dysplasia, X-linked	XL	GRN	Neuronal ceroid lipofuscinosis type 11	AR

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EDAR	Hypohidrotic ectodermal dysplasia 10B	AR	GUCY2D	Leber congenital amaurosis 1; Choroidal dystrophy, central areolar 1	AR
EMD	Emery-Dreifuss muscular dystrophy	XL	HADH	Familial hyperinsulinemic hypoglycemia, familial 4; 3-hydroxyacyl-CoA dehydrogenase deficiency	AR
ERCC2	Xeroderma pigmentosum	AR	HADHA	Long-chain 3-Hydroxyacyl-CoA dehydrogenase deficiency; Trifunctional protein deficiency	AR
ETFA	Glutaric acidemia, Type IIA	AR	HAX1	Severe congenital neutropenia 3, autosomal recessive	AR
ETFB	Glutaric acidemia, Type IIB	AR	HBA1	Alpha-thalassemia	AR
ETFDH	Glutaric acidemia, Type IIC	AR	HBA2	Alpha-thalassemia	AR
ETHE1	Ethylmalonic encephalopathy	AR	HBB	Beta-thalassemia, and other hemoglobinopathies	AR
EXOSC3	Pontocerebellar hypoplasia type 1B	AR	HEXA	Megalencephalic Leukoencephalopathy with Subcortical Cysts, types 2A & 2B	AR
EYS	Retinitis pigmentosa 25, EYS-related	AR	HEPACAM	Tay-Sachs disease; GM2-ganlgliosidosis	AR
F2	Hypoprothrombinemia	AR	HEXB	Sandhoff disease	AR
F5	Factor V deficiency	AR	HFE	Hereditary hemochromatosis type 1, HFE-related	AR
F8	Hemophilia A	XL	HFE2	Hereditary hemochromatosis type 2A, HFE2-related	AR
F9	Hemophilia B	XL	HGD	Alkaptonuria	AR
F11	Factor XI Deficiency	AR	HGSNAT	Mucopolysaccharidosis, type IIIC	AR
FAH	Tyrosinemia, type I	AR	HLCS	Holocarboxylase synthetase deficiency	AR

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FAM161A	Retinitis pigmentosa 28	AR	HMGCL	3-hydroxy-3-methylglutaryl CoA lyase deficiency	AR
FANCA	Fanconi anemia, complementation group A	AR	HOGA1	Primary hyperoxaluria, type III	AR
FANCC	Fanconi anemia, complementation group C	AR	HPRT1	Lesch-Nyhan syndrome; HPRT-related gout	XL
FANCG	Fanconi Anemia, complementation group G	AR	HPS1	Hermansky-Pudlak syndrome, type 1	AR
FH	Fumarase deficiency	AR	HPS3	Hermansky-Pudlak syndrome, type 3	AR
FKRP	Limb-girdle muscular dystrophy, type 2I; 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	AR	HPS4	Hermansky-Pudlak syndrome, type 4	AR
FKTN	Muscular dystrophy-dystroglycanopathy type 4A (Walker-Warburg syndrome); Muscular dystrophy-dystroglycanopathy type 4B; Muscular dystrophy-dystroglycanopathy type 4C; Cardiomyopathy, dilated, 1X	AR	HPS5	Hermansky-Pudlak syndrome, type 5	AR
FMR1	Fragile X syndrome;FMR1-related primary ovarian insufficiency; Fragile X-associated tremor/ataxia syndrome	XL	HPS6	Hermansky-Pudlak syndrome, type 6	AR
G6PC	Glycogen storage disease, type IA	AR	HSD17B3	17-beta-hydroxysteroid dehydrogenase deficiency, type III	AR
G6PD	Glucose-6-phosphate dehydrogenase deficiency; Hemolytic anemia due to G6PD deficiency	XL	HSD17B4	D-bifunctional protein deficiency; Perrault syndrome 1	AR

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GAA	Glycogen storage disease, type II	AR	HSD3B2	Congential adrenal hyperplasia due to 3-Beta-hydroxysteroid dehydrogenase deficiency, type II	AR
GALC	Krabbe disease	AR	IDS	Mucopolysaccharidosis, type II (Hunter syndrome)	XL
GALK1	Galactokinase deficiency	AR	IDUA	Mucopolysachharidosis, type I (Hurler syndrome)	AR
GALNT3	Hyperphospatemic tumoral calcinosis, familial	AR	IKBKAP	Familial dysautonomia (HSAN3)	AR
GALT	Galactosemia	AR	IL2RG	Severe combined immunodeficiency, X-linked	XL
GAMT	Guanidinoacetate methyltransferase deficiency	AR	ISPD	Walker-Warburg (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7	AR
GBA	Gaucher disease	AR	IVD	Isovaleric acidemia	AR
GBE1	Glycogen storage disease, type IV	AR	KCNJ11	Familial hyperinsulinemic hypoglycemia type 2, KCNJ11-related	AR
GCDH	Glutaric acidemia, type I	AR	KCTD7	Neuronal ceroid lipofuscinosis 14 (progressive myoclonic epilepsy type 3)	AR
GDF5	Du Pan syndrome; Chondrodysplasia, Grebe type; Brachydactyly type A1,C; Acromesomelic dysplasia, Hunter-Thompson type	AR	L1CAM	L1 syndrome; MASA syndrome, CRASH syndrome	XL
GFPT1	Congenital myasthenic syndrome 12	AR	LAMA3	Herlitz junctional epidermolysis bullosa, LAMA3-related; Laryngoonychocutaneous syndrome; Epidermolysis bullosa, generalized atrophic benign	AR
GJB1	Charcot-Marie-Tooth disease, GJB1-related	XL	LAMB3	Herlitz junctional epidermolysis bullosa; non-Herlitz junctional epidermolysis bullosa	AR

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GJB2	Nonsyndromic hearing loss, GJB2-related	AR	LAMC2	Herlitz junctional epidermolysis bullosa; non-Herlitz junctional epidermolysis bullosa	AR
GJB6	Nonsyndromic Hearing Loss, GJB6-related	AR	LARGE1	Muscular dystrophy-dystroglycanopathy, congenital with brain and eye anomalies, type 6A (Walker-Warburg); Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type 6B	AR
GLA	Fabry disease	XL	LCA5	Leber congenital amaurosis 5, LCA5-related	AR
GLB1	GM1-gangliosidosis; Mucopolysaccharidosis type IVB	AR	LHCGR	Leydig cell hypoplasia; Luteinizing hormone resistance	AR
GLDC	Glycine encephalopathy, GLDC-related	AR	LIFR	Stuve-Wiedemann syndrome	AR
GNE	Inclusion body myopathy, type II	AR	LIPA	Cholesteryl ester storage disease	AR
GNPAT	Rhizomelic chondrodysplasia punctata, type II	AR	LIPH	Woolly hair/hypotrichosis, autosomal recessive	AR
GNPTAB	Mucolipidosis, type II alpha/beta; Mucolipidosis, type III alpha/beta	AR	LOXHD1	Autosomal recessive deafness 77	AR
LRPPRC	Leigh syndrome with COX deficiency (French Canadian type)	AR	LPL	Lipoprotein lipase deficiency	AR
LYST	Chediak-Higashi syndrome	AR	PEX12	Zellweger syndrome spectrum, PEX12- related	AR
MAN2B1	Alpha-mannosidosis type I & II	AR	PEX26	Zellweger syndrome spectrum, PEX26- related	AR
MCCC1	3-Methylcrontonyl-CoA carboxylase 1 deficiency	AR	PFKM	Glycogen storage disease, type VII	AR
MCCC2	3-Methylcrontonyl-CoA carboxylase 2 deficiency	AR	PHGDH	3-phosphoglycerate dehydrogenase deficiency; Neu-Laxova syndrome 1	AR

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MCOLN1	Mucolipidosis type IV	AR	PKHD1	Polycystic kidney disease, autosomal recessive	AR
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy	AR	PMM2	Congenital disorder of glycosylation, type IA	AR
MEFV	Familial Mediterranean fever	AR	POLG	Mitochondrial DNA depletion syndrome type 4A (Alpers type); Mitochondrial DNA depletion syndrome type 4B (MNGIE type); Mitochondrial Recessive Ataxia Syndrome (includes SANDO and SCAE); Progressive external ophthalmoplegia with mitochondrial deletions autosomal recessive type 1	AR
MFSD8	Neuronal ceroid-lipofuscinosis 7, MFSD8-related; Macular dystrophy with central cone involvement	AR	POMGNT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type 3A; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type 3B; Muscular dystrophy-dystroglycanopathy (limb-girdle), type 3C	AR
MKKS	Bardet-Biedl syndrome 6; McKusick-Kaufman syndrome	AR	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	AR
MKS1	Bardet-Biedl syndrome 13; Joubert syndrome 28; Meckel syndrome 1	AR	POMT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2; Muscular dystrophy-dystroglycanopathy	AR

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				(congenital with mental retardation), type B, 2; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2	
MLC1	Megalencephalic Leukoencephalopathy with subcortical cysts, type I	AR	PPT1	Neuronal ceroid lipofuscinosis 1, PPT1-related	AR
MMAA	Methylmalonic aciduria, cblA type	AR	PRPS1	Arts Syndrome;Charcot-Marie-Tooth disease, PRPS1-related; Gout, PRPS1-related	XL
MMAB	Methylmalonic aciduria, cblB type	AR	PSAP	Metachromatic leukodystrophy due to SAP-b deficiency; Atypical Gaucher disease; Atypical Krabbe disease	AR
MMADHC	Methylmalonic aciduria, cblD type	AR	PUS1	Mitochondrial myopathy and sideroblastic anemia 1	AR
MPI	Congenital disorder of glycosylation, type IB	AR	PYGL	Glycogen storage disease VI	AR
MPL	Congenital amegakaryocytic thrombocytopenia	AR	PYGM	Glycogen storage disease, type V	AR
MPV17	Hepatocerebral mitochondrial DNA depletion syndrome 6, MPV17-related	AR	RAB23	Carpenter syndrome	AR
MRE11A	Ataxia-telangiectasia-like disorder 1	AR	RAG1	Severe combined immunodeficiency, RAG1-related; Omenn syndrome;Combined cellular and humoral immune defects with granulomas	AR
MTHFR	Homocystinuria due to MTHFR deficiency, severe; Neural tube defects folate-sensitive	AR	RAG2	Omenn syndrome;Severe combined immunodeficiency, Athabascan type	AR
MTM1	Myotubular myopathy, MTM1-related	XL	RAPSN	Congenital myasthenic syndrome 11, RAPSN-related; Fetal akinesia deformation sequence	AR
MTTP	Abetalipoproteinemia	AR	RARS2	Pontocerebellar hypoplasia type 6	AR

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MUT	Methylmalonic aciduria mut(0) type, MUT-related	AR	RDH12	Leber congenital amaurosis 13	AR
MYO7A	Usher syndrome, type IB; Deafness, autosomal recessive 2	AR	RNASEH2A	Aicardi-Goutieres syndrome 4	AR
NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo B)	AR	RNASEH2B	Aicardi-Goutieres syndrome 2	AR
NAGS	N-acetylglutamate synthase deficiency	AR	RNASEH2C	Aicardi-Goutieres syndrome 3, RNASEH2C-related	AR
NBN	Nijmegen breakage syndrome	AR	RPE65	Leber congenital amaurosis 2; Retinitis pigmentosa 20	AR
NDUFAF6	Mitochondrial complex 1 deficiency, NDUFAF6-related	AR	RS1	Juvenile retinoschisis, X-linked	XL
NEB	Nemaline myopathy 2	AR	RTEL1	Dyskeratosis congenita, autosomal recessive 5	AR
NPC1	Niemann-pick disease, type C1	AR	SACS	Spastic ataxia of Charlevoix-Saguenay, autosomal recessive	AR
NPC2	Niemann-pick disease, type C2	AR	SAMD9	Familial tumoral calcinosis, normophosphatemic	AR
NPHS1	Nephrotic syndrome, type I	AR	SAMHD1	Aicardi-Goutieres syndrome 5	AR
NPHS2	Nephrotic syndrome, type II	AR	SBDS	Shwachman-Diamond syndrome	AR
NR2E3	Enhanced S-cone syndrome	AR	SEPSECS	Pontocerebellar hypoplasia 2D	AR
NTRK1	Congenital insensitivity to pain with anhidrosis	AR	SERPINA1	Alpha-1 antitrypsin deficiency	AR
OPA3	3-methylglutaconic aciduria, type III	AR	SGCA	Limb-girdle muscular dystrophy, type 2D	AR
OTC	Ornithine transcarbamylase deficiency	XL	SGCB	Limb-girdle muscular dystrophy, type 2E	AR
PAH	Phenylalanine hydroxylase deficiency	AR	SGCG	Limb-girdle muscular dystrophy, type 2C	AR

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PCCA	Propionic acidemia, PCCA-related	AR	SGSH	Mucopolysaccharidosis type IIIA (Sanfilippo A)	AR
PCCB	Propionic acidemia, PCCB-related	AR	SLC12A3	Gitelman syndrome	AR
PCDH15	Usher syndrome, type IF; Deafness, autosomal recessive 23	AR	SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy (Andermann syndrome)	AR
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency, X-Linked	XL	SLC17A5	Salla disease;Infantile sialic acid storage disorder	AR
PDHB	Pyruvate dehydrogenase E1-beta deficiency, autosomal recessive	AR	SLC22A5	Primary carnitine deficiency	AR
PEPD	Prolidase deficiency	AR	SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome (Ornithine translocase deficiency)	AR
PET100	Mitochondrial complex IV deficiency	AR	SLC26A2	Sulfate transporter-related osteochondrodysplasia; Achondrogenesis Ib; Atelosteogenesis II; Diastrophic dysplasia; Epiphyseal dysplasia, multiple, 4	AR
PEX1	Zellweger syndrome spectrum, PEX1-related	AR	SLC26A4	Pendred syndrome; Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	AR
PEX2	Zellweger syndrome spectrum, PEX2-related	AR	SLC35A3	Arthrogryposis, mental retardation and seizures	AR
PEX6	Zellweger syndrome spectrum, PEX6-related	AR	SLC37A4	Glycogen storage disease, type 1b; Gycogen storage disease type 1c	AR
PEX7	Rhizomelic chondrodysplasia punctata, type I; Peroxisome biogenesis disorder 9B	AR	SLC39A4	Acrodermatitis enteropathica	AR

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PEX10	Zellweger syndrome spectrum, PEX10- related	AR	SLC4A11	Corneal dystrophy and perceptive deafness syndrome; Autosomal recessive corneal dystrophy	AR
SLC6A8	Creatine transporter defect, SLC6A8-related (Cerebral creatine deficiency syndrome 1)	XL	UGT1A1	Crigler-Najjar syndrome, type I; Crigler-Najjar syndrome, type II;Hyperbilirubinemia, familial transient neonatal Gilbert syndrome	AR
SMN1	Spinal muscular atrophy	AR	USH1C	Usher syndrome, type IC; Deafness, autosomal recessive 18A	AR
SMPD1	Niemann-Pick disease type A; Niemann-Pick disease type B	AR	USH2A	Usher syndrome, Type 2A; Retinitis pigmentosa 39	AR
ST3GAL5	Amish infantile epilepsy syndrome	AR	VPS13A	Choreoacanthocytosis	AR
STAR	Congenital lipoid adrenal hyperplasia	AR	VPS53	Pontocerebellar hypoplasia 2E	AR
STS	X-linked ichthyosis	XL	VRK1	Pontocerebellar hypoplasia, type 1A	AR
SUMF1	Multiple sulphatase deficiency	AR	XPA	Xeroderma pigmentosum group A	AR
TAT	Tyrosinemia type II (Richner-Hanhart syndrome)	AR	XPC	Xeroderma pigmentosum group C	AR
TCIRG1	Osteopetrosis type 1, infantile malignant	AR	ZFYVE26	Spastic paraplegia type 15, ZFYVE26-related	AR
TECPR2	Hereditary spastic paraparesis, type 49	AR	CYP11B1	11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia	AR
TFR2	Hereditary hemochromatosis type 3, TFR2-related	AR	PTS	6-pyruvoyl-tetrahydropterin Synthase Deficiency	AR
TGM1	Lamellar ichthyosis type I	AR	ALMS1	Alstrom Syndrome	AR
TH	Segawa syndrome (tyrosine hydroxylase deficiency)	AR	ARG1	Argininemia	AR

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TMEM216	Joubert syndrome 2; Meckel syndrome 2	AR	ATP7A	Menkes disease;Occipital horn syndrome;Spinal muscular atrophy, distal, X-linked 3	XLR
TPP1	Neuronal ceroid lipofuscinosis 2, TPP1-related; Spinocerebellar ataxia type 7	AR	CPS1	Carbamoylphosphate Synthetase I Deficiency	AR
TREX1	Aicardi-Goutieres syndrome type 1, TREX1-related	AR	VPS13B	Cohen Syndrome	AR
TRIM37	Mulibrey nanism	AR	COL4A4	Alport syndrome 2	AR
TSEN2	Pontocerebellar hypoplasia type 2B	AR	PROP1	Combined Pituitary Hormone Deficiency,PROP1-related	AR
TSEN34	Pontocerebellar hypoplasia type 2C	AR	SGCD	Muscular dystrophy, limb-girdle, autosomal recessive 6	AR
TSEN54	Pontocerebellar hypoplasia type 2A; pontocerebellar hypoplasia type 4; Pontocerebellar hypoplasia type 5	AR	ERCC6	Cerebrooculofacioskeletal syndrome 1;Cockayne syndrome, type B;De Sanctis-Cacchione syndrome;UV-sensitive syndrome 1	AR
TTC8	Bardet-Biedl syndrome 8; Retinitis pigmentosa 51	AR	ERCC8	Cockayne syndrome, type A;UV-sensitive syndrome 2	AR
TTN	Early onset myopathy with fatal cardiomyopathy; Limb-girdle muscualr dystrophy 2J;Salih myopathy	AR	EVC	EVC-related Ellis-van Creveld Syndrome	AR
TTPA	Ataxia with vitamin E deficiency	AR	EVC2	EVC2-related Ellis-van Creveld Syndrome	AR
UBR1	Johanson-Blizzard syndrome	AR	HYLS1	Hydrolethalus Syndrome	AR
LAMA2	LAMA2-related Muscular Dystrophy	AR	CCDC151	Primary ciliary dyskinesia-30	AR
MMACHC	Methylmalonic Aciduria and Homocystinuria,cblC Type	AR	GALNS	Mucopolysaccharidosis IVA	AR
PC	Pyruvate Carboxylase Deficiency	AR	CD40LG	Immunodeficiency with hyper-IgM	XLR

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GFM1	Combined oxidative phosphorylation deficiency 1	AR	CHRNG	Escobar syndrome;Multiple pterygium syndrome	AR
NR0B1	X-linked Congenital Adrenal Hypoplasia	XLR	COL27A1	Steel syndrome	AR
ACAD9	Mitochondrial complex I deficiency, nuclear type 20	AR	CYBA	Chronic granulomatous disease , due to deficiency of CYBA	AR
ESCO2	Roberts syndrome;SC phocomelia syndrome	AR	CYBB	Chronic granulomatous disease;Immunodeficiency 34, mycobacteriosis	XLR
AQP2	Nephrogenic diabetes insipidus	AD,AR	DCX	Lissencephaly;Subcortical laminal heterotopia	XL
ARSE	Chondrodysplasia punctata	XLR	DNAL1	Primary ciliary dyskinesia-16	AR
ATRX	Alpha-thalassemia/mental retardation syndrome	XLD	EIF2AK3	Wolcott-Rallison syndrome	AR
CCDC103	Primary ciliary dyskinesia-17	AR	EIF2B5	Leukoencephalopathy with vanishing white matter	AR
GLE1	Congenital arthrogryposis with anterior horn cell disease;Lethal congenital contracture syndrome 1	AR	HYAL1	Mucopolysaccharidosis type IX	AR
GP1BA	Bernard-Soulier syndrome, type A1;Nonarteritic anterior ischemic optic neuropathy, susceptibility to	AR	LDLRAP1	Autosomal recessive hypercholesterolemia	AR
GP9	Bernard-Soulier syndrome, type C	AR	LHX3	Pituitary hormone deficiency, combined, 3	AR
GUSB	Mucopolysaccharidosis VII	AR	MTRR	Homocystinuria-megaloblastic anemia, cbl E type;Neural tube defects, folate-sensitive, susceptibility to	AR
HJV	Hemochromatosis, type 2A	AR	NDRG1	Charcot-Marie-Tooth disease, type 4D	AR
TSFM	Combined oxidative phosphorylation deficiency 3	AR	NDUFAF5	Mitochondrial complex I deficiency, nuclear type 16	AR

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TTC37	Trichohepatoenteric syndrome 1	AR	NDUFS6	Mitochondrial complex I deficiency, nuclear type 9	AR
TYMP	Mitochondrial DNA depletion syndrome 1	AR	NPHP1	Nephronophthisis 1, juvenile	AR
VPS45	Neutropenia, severe congenital, 5	AR	OAT	Gyrate atrophy of choroid and retina with or without ornithinemia	AR
WAS	Neutropenia, severe congenital, X-linked;Thrombocytopenia, X-linked;Wiskott-Aldrich syndrome	XLR	OCRL	Dent disease 2;Lowe syndrome	AR
WNT10A	Odontoonychodermal dysplasia;Schopf-Schulz-Passarge syndrome	AR	PLA2G6	Infantile neuroaxonal dystrophy 1;Neurodegeneration with brain iron accumulation 2B;Parkinson disease 14	AR
SLC7A7	Lysinuric protein intolerance	AR	RPGRIP1L	COACH syndrome;Joubert syndrome 7;Meckel syndrome 5	AR
SMARCAL1	Schimke immunoosseous dysplasia	AR	SLC25A13	Citrullinemia, adult-onset type II;Citrullinemia, type II, neonatal-onset	AR
TRIM32	Bardet-Biedl syndrome 11;Muscular dystrophy, limb-girdle, autosomal recessive 8	AR	SLC25A20	Carnitine-acylcarnitine translocase deficiency	AR
TRMU	Liver failure, transient infantile	AR	SLC26A3	Congenital secretory chloride diarrhea	AR