CarrierCheck Expanded (462 genes) Comprehensive Carrier Screening Test

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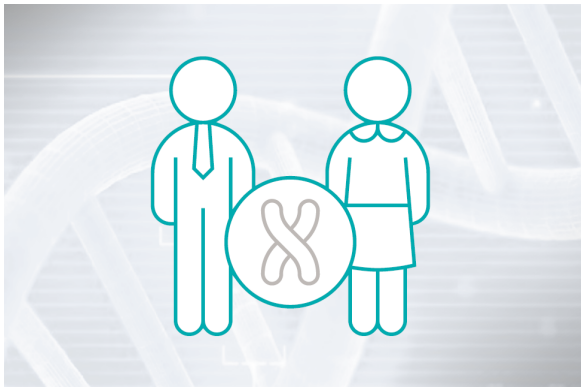
Who is a carrier?

A carrier is an individual who has a mutation in one of the alleles of a gene associated with a genetic disease.



> *"CarrierCheck enables genetic matching of gamete donors to their recipients"*

What is the CarrierCheck test?

Expanded preconceptional carrier test enables the detection of couples at risk for single gene diseases. It allows couples to make the right reproductive decision and reduces the risk of having a affected child. It specifically screens autosomal recessive and X-linked recessive inherited disorders. Next-Generation Sequencing (NGS) empowered by powerful bioinformatic tools enable simultaneous screening of hundreds of diseases with a single universal method.

> *"Carrier Tests can aid to prevent genetic disorders"*

Why CarrierCheck test?

- Comprehensive screening capacity with expanded gene panel - 462 genes
- High variant detection sensitivity,
- Simultaneous detecion of CNVs, SNPs with a single NGS based test
- Special analysis algorithms for efficient diagnosis of challenging gene regions –pseudogenes and homologous genes - SMN1, HBA1/2, CYP21A2, DMD, CFTR, GBA
- Additional MLPA tests for detection of deletion/duplication in SMN1/2 and HBA1/2
- Addtional TP-PCR for triplet nucleotide repeat detection in FMR1 gene.
- Exon level CNV detection for genes related to critical diseases - DMD, CFTR
- Fast and reliable results with exclusive analysis tool developed by Franklin by Genoox.

> *"Next generation risk reduction for consanguineous couples"*

Who are the candidates of carrier screening?

All couples who want to reduce the risk of having a child with genetic disease can be referred.

- Consanguineous individuals
- People at high risk for a particular disease, based on their ethnic background
- Before IVF applications and donor cycles

Novel reporting module:

> *"Personalized genetic counselling with residual carrier risk"*

- Artificial intelligance supported variant classification
- Ethnic specific residual carrier risk reporting
- Tracking of test process
- Patient specific variant reporting algorithm
- Custom tailored genetic counselling: To inform patients about the test-limitations and couple spesific risks
- Genetic matching is enabled via duo-analysis of gamete donors and their recipients

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GENES	CONDITIONS
ABCA3	Surfactant metabolism dysfunction, pulmonary, 3
ABCA4	Stargardt disease, type 1
ABCB11	Progressive familial intrahepatic cholestasis, type 2
ABCC8	Familial hyperinsulinism, ABCC8-related
ABCD1	Adrenoleukodystrophy, X-linked
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type
ACAD9	Mitochondrial complex I deficiency, ACAD9-related
ACADM	Medium chain acyl-CoA dehydrogenase deficiency
ACADS	Short chain acyl-CoA dehydrogenase deficiency
ACADSB	Short/branched chain acyl-CoA dehydrogenase deficiency
ACADVL	Very long chain acyl-CoA dehydrogenase deficiency
ACAT1	Beta-ketothiolase deficiency
ACOX1	Acyl-CoA oxidase I deficiency
ACSF3	Combined malonic and methylmalonic aciduria
ADA	Severe combined immunodeficiency, ADA-related
ADAMTS2	Ehlers-Danlos syndrome, type VIIC
ADGRG1	Bilateral frontoparietal polymicrogyria
ADK	Hypermethioninemia due to adenosine kinase deficiency
AFF2	Intellectual developmental disorder, X-linked 109
AGA	Aspartylglucosaminuria
AGL	Glycogen storage disease, type III (Cori/Forbes)
AGPS	Rhizomelic chondrodysplasia punctata, type 3
AGXT	Hyperoxaluria, primary, type 1
AHCY	Hypermethioninemia with deficiency of S-adenosyl-homocysteine hydrolase
AHI1	Joubert syndrome 3
AIPL1	Cone-rod dystrophy; Leber congenital amaurosis 4; Retinitis pigmentosa, juvenile
AIRE	Autoimmune polyendocrinopathy syndrome, type I
ALDH3A2	Sjögren-Larsson syndrome
ALDH4A1	Hyperprolinemia, type II
ALDOB	Hereditary fructose intolerance
ALG6	Congenital disorder of glycosylation, type 1C
ALMS1	Alström syndrome
ALPL	Hypophosphatasia, ALPL-related
AMT	Glycine encephalopathy, AMT-related
ANO10	Spinocerebellar ataxia, autosomal recessive 10
AP1S2	Mental retardation, X-linked syndromic 5
AQP2	Familial nephrogenic diabetes insipidus, AQP2-related
ARG1	Argininemia
ARL13B	Joubert syndrome 8
ARSA	Argininemia
ARSB	Mucopolysaccharidosis, type VI (Maroteaux-Lamy)
ARSE	Chondrodysplasia punctata, X-linked recessive
ARX	Developmental and epileptic encephalopathy 1; Hydranencephaly with abnormal genitalia; Lissencephaly, X-linked 2; Mental retardation, X-linked 29 and others; Partington syndrome; Proud syndrome
ASL	Argininosuccinate lyase deficiency
ASNS	Asparagine synthetase deficiency
ASPA	Canavan disease
ASS1	Citrullinemia, type 1
ATM	Ataxia-telangiectasia
ATP6V1B1	Renal tubular acidosis and deafness, ATP6V1B1-related
ATP7A	Menkes syndrome, X-linked
ATP7B	Wilson disease
ATRX	Alpha-thalassemia intellectual disability syndrome, X-linked

GENES	CONDITIONS
BBS1	Bardet-Biedl syndrome 1
BBS10	Bardet-Biedl syndrome 10
BBS12	Bardet-Biedl syndrome 12
BBS2	Bardet-Biedl syndrome 2
BBS4	Bardet-Biedl syndrome 4
BCHE	Pseudocholinesterase deficiency
BCKDHA	Maple syrup urine disease, type 1A
BCKDHB	Maple syrup urine disease, type 1B
BCS1L	GRACILE syndrome
BLM	Bloom syndrome
BRWD3	Mental retardation, X-linked 93
BSND	Bartter syndrome, type 4a
BTD	Biotinidase deficiency
CANT1	Desbuquois dysplasia 1
CAPN3	Limb-girdle muscular dystrophy, type 2A
CASQ2	Catecholaminergic polymorphic ventricular tachycardia
CBS	Homocystinuria, CBS-related
CC2D1A	Mental retardation, autosomal recessive 3
CC2D2A	COACH syndrome 2; Joubert syndrome 9; Meckel syndrome 6; Retinitis pigmentosa 93
CCDC103	Ciliary dyskinesia, primary, 17
CCDC151	Ciliary dyskinesia, primary, 30
CCDC39	Ciliary dyskinesia, primary, 14
CCDC88C	Hydrocephalus, congenital, 1
CD40LG	Immunodeficiency, X-linked, with hyper-IgM
CDH23	Usher syndrome, type 1D
CEP290	Leber congenital amaurosis, type CEP290
CERKL	Retinitis pigmentosa 26
CFTR	Cystic fibrosis
CHM	Choroideremia, X-linked
CHRNE	Congenital myasthenic syndrome, CHRNE-related
CHRNG	Escobar syndrome
CHST6	Macular corneal dystrophy
CIITA	Bare lymphocyte syndrome, CIITA-related
CLCN1	Myotonia congenita, recessive
CLN3	Ceroid lipofuscinosis, neuronal, 3
CLN5	Ceroid lipofuscinosis, neuronal, 5
CLN6	Ceroid lipofuscinosis, neuronal, 6
CLN8	Ceroid lipofuscinosis, neuronal, 8 (a.k.a. Northern epilepsy)
CLRN1	Usher syndrome, type 3
CNGA1	Retinitis pigmentosa 49
CNGA3	Achromatopsia, CNGA3-related
CNGB1	Retinitis pigmentosa 45
CNGB3	Achromatopsia, CNGB3-related
COL27A1	Steel syndrome
COL4A3	Alport syndrome, COL4A3-related
COL4A4	Alport syndrome, COL4A4-related
COL4A5	Alport syndrome, X-linked
COL7A1	Dystrophic epidermolysis bullosa, COL7A1-related
CPS1	Carbamoyl phosphate synthetase I deficiency
CPT1A	Carnitine palmitoyltransferase IA deficiency
CPT2	Carnitine palmitoyltransferase II deficiency
CRADD	Intellectual developmental disorder, autosomal recessive 34, with variant lissencephaly
CRB1	Leber congenital amaurosis 8
CRYL1	Keratoderma, Palmoplantar, with Deafness
CTNS	Cystinosis
CTSK	Pycnodysostosis

GENES	CONDITIONS
CUL4B	Mental retardation, X-linked, syndromic 15 (Cabezas type)
CYBA	Chronic granulomatous disease, CYBA-related
CYBB	Chronic granulomatous disease, X-linked
CYP11A1	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete
CYP11B1	Congenital adrenal hyperplasia, 11-beta-hydroxylase-deficient
CYP11B2	Corticosterone methyloxidase deficiency
CYP17A1	Congenital adrenal hyperplasia, 17-alpha-hydroxylase deficiency
CYP19A1	Aromatase deficiency
CYP1B1	Primary congenital glaucoma
CYP21A2	Congenital adrenal hyperplasia, 21-hydroxylase-deficient
CYP27A1	Cerebrotendinous xanthomatosis
CYP27B1	Vitamin D-dependent rickets, type I
DBT	Maple syrup urine disease, type 2
DCLRE1C	Severe combined immunodeficiency, type athabaskan
DCX	Lissencephaly, X-linked; Subcortical laminal heterotopia, X-linked
DDX11	Warsaw breakage syndrome
DHCR7	Smith-Lemli-Opitz syndrome
DHDDS	Retinitis pigmentosa 59
DLD	Dihydrolipoamide dehydrogenase deficiency
DLG3	Mental retardation, X-linked 90
DMD	Duchenne/Becker muscular dystrophy
DNAH5	Ciliary dyskinesia, primary 3
DNAI1	Ciliary dyskinesia, primary 1
DNAI2	Ciliary dyskinesia, primary 9
DNAL1	Ciliary dyskinesia, primary, 16
DPYD	Dihydropyrimidine dehydrogenase deficiency
DUOX2	Thyroid dyshormonogenesis 6
DUOXA2	Thyroid dyshormonogenesis 5
DYNC2H1	Short-rib thoracic dysplasia 3 with or without polydactyly
DYSF	Limb-girdle muscular dystrophy, type 2B
EDA	Hypohidrotic ectodermal dysplasia, X-linked
EIF2AK3	Wolcott-Rallison syndrome
EIF2B5	Leukoencephalopathy with vanishing white matter
ELP1	Dysautonomia, familial
EMD	Emery-Dreifuss muscular dystrophy 1, X-linked
ERCC2	Xeroderma pigmentosum, group D; Trichothiodystrophy 1, photosensitive; Cerebrooculofacioskeletal syndrome 2
ERCC6	Cockayne syndrome, type B
ERCC8	Cockayne syndrome, type A
ESCO2	Roberts syndrome
ETFA	Glutaric acidemia, type 2A
ETFB	Glutaric acidemia, type 2B
ETFDH	Glutaric acidemia, type 2C
ETHE1	Ethylmalonic encephalopathy
EVC	Ellis-van Creveld syndrome, EVC-related
EVC2	Ellis-van Creveld syndrome, EVC2-related
EXOSC3	Pontocerebellar hypoplasia, type 1B
F11	Factor XI deficiency
F2	Prothrombin deficiency
F5	Factor V deficiency; Thrombophilia due to thrombin defect
F8	Hemophilia A
F9	Hemophilia B
FAH	Tyrosinemia, type I

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GENES	CONDITIONS
FAM161A	Retinitis pigmentosa 28
FANCA	Fanconi anemia, group A
FANCC	Fanconi anemia, group C
FANCG	Fanconi anemia, group G
FGD1	Aarskog-Scott syndrome; Mental retardation, X-linked syndromic 16
FH	Fumarase deficiency
FKRP	Limb-girdle muscular dystrophy, type 2I
FKTN	Walker-Warburg syndrome, FKTN-related
FMO3	Trimethylaminuria
FMR1	Fragile X syndrome
FTCD	Glutamate formiminotransferase deficiency
FTSJ1	Mental retardation, X-linked 9/44
FXN	Friedreich ataxia
G6PC	Glycogen storage disease, type IA
G6PD	Glucose-6-phosphate dehydrogenase deficiency
GAA	Glycogen storage disease, type II (Pompe disease)
GALC	Krabbe disease
GALE	Galactose epimerase deficiency
GALK1	Galactokinase deficiency (galactosemia, type II)
GALNS	Mucopolysaccharidosis, type IVA
GALT	Galactosemia
GAMT	Guanidinoacetate methyltransferase deficiency
GBA	Gaucher disease
GBE1	Glycogen storage disease, type IV
GCDH	Glutaric acidemia, type 1
GDAP1	Charcot-Marie-Tooth disease, axonal, type 2K; Charcot-Marie-Tooth disease, recessive intermediate, A; Charcot-Marie-Tooth disease, axonal, with vocal cord paresis
GFM1	Combined oxidative phosphorylation deficiency 1
GH1	Isolated growth hormone deficiency, type IA/II
GJB1	Charcot-Marie-Tooth disease with deafness, X-linked
GJB2	Non-syndromic hearing loss (a.k.a. connexin 26)
GJB6	Non-syndromic hearing loss (a.k.a. connexin 30)
GLA	Fabry disease
GLB1	Mucopolysaccharidosis, type IVB / GM1 ganglio-sidosis
GLDC	Glycine encephalopathy, GLDC-related
GLE1	Lethal congenital contracture syndrome 1
GNE	Inclusion body myopathy 2
GNPAT	Rhizomelic chondrodysplasia punctata, type 2
GNPTAB	Mucolipidosis II/IIIA
GNPTG	Mucolipidosis III gamma
GNRHR	Hypogonadotropic hypogonadism 7 without anosmia
GNS	Mucopolysaccharidosis, type IIID (Sanfilippo D)
GP1BA	Bernard-Soulier syndrome, type A2
GP9	Bernard-Soulier syndrome, type C
GPR143	Nystagmus 6, congenital, X-linked; Ocular albinism, type I, Nettleship-Falls type
GRHPR	Primary hyperoxaluria, type 2
GRIP1	Fraser syndrome 3
GUSB	Mucopolysaccharidosis, type VII
HADHA	Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency
HAX1	Congenital neutropenia, HAX1-related
HBA1	Alpha-thalassemia
HBA2	Alpha-thalassemia
HBB	Beta-hemoglobinopathies

GENES	CONDITIONS
HEXA	Tay-Sachs disease
HEXB	Sandhoff disease
HFE	Hemochromatosis, type 1
HGD	Alkaptonuria
HGSNAT	Mucopolysaccharidosis, type IIIC (Sanfilippo C)
HJV	Hemochromatosis, type 2A
HLCS	Holocarboxylase synthetase deficiency
HMGCL	3-hydroxy-3-methylglutaryl-coenzyme A lyase deficiency
HOGA1	Primary hyperoxaluria, type 3
HPS1	Hermansky-Pudlak syndrome 1
HPS3	Hermansky-Pudlak syndrome 3
HSD17B4	D-bifunctional protein deficiency
HSD3B2	3-beta-hydroxysteroid dehydrogenase type II deficiency
HYAL1	Mucopolysaccharidosis type IX
HYLS1	Hydrolethalus syndrome
IDH3B	Retinitis pigmentosa 46
IDS	Mucopolysaccharidosis, type II (Hunter syndrome)
IDUA	Mucopolysaccharidosis, type I (Hurler syndrome)
IL1RAPL1	Intellectual developmental disorder, X-linked 21
IL2RG	Severe combined immunodeficiency, X-linked
ITGB3	Glanzmann thrombasthenia
IVD	Isovaleric acidemia
IYD	Thyroid dyshormonogenesis 4
JAK3	SCID, autosomal recessive, T-negative/B-positive type
KCNJ11	Congenital hyperinsulinism, KCNJ11-related
KDM5C	Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type
L1CAM	CRASH syndrome; Corpus callosum, partial agenesis of; Hydrocephalus with Hirschsprung disease; MASA syndrome
LAMA2	LAMA2-related muscular dystrophy
LAMA3	Herlitz junctional epidermolysis bullosa, LAMA3-related
LAMB3	Herlitz junctional epidermolysis bullosa, LAMB3-related
LAMC2	Herlitz junctional epidermolysis bullosa, LAMC2-related
LCA5	Leber congenital amaurosis, type LCA5
LDLRAP1	Familial hypercholesterolemia, LDLRAP1-related
LHX3	Pituitary hormone deficiency, combined, 3
LIFR	Stuve-Wiedemann syndrome
LIPA	Lysosomal acid lipase deficiency
LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type
LOXHD1	Deafness, autosomal recessive 77
LPL	Lipoprotein lipase deficiency
LRP2	Donnai-Barrow syndrome
LRPPRC	Leigh syndrome, French-Canadian type
LYST	Chediak-Higashi syndrome
MAN2B1	Alpha-mannosidosis
MCCC1	3-methylcrotonyl-CoA carboxylase 1 deficiency
MCCC2	3-methylcrotonyl-CoA carboxylase 2 deficiency
MCEE	Methylmalonyl-CoA epimerase deficiency
MCOLN1	Mucolipidosis, type IV
MCPH1	Microcephaly 1, primary

GENES	CONDITIONS
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy
MEFV	Familial Mediterranean fever
MESP2	Spondylothoracic dysostosis, MESP2-related
MFSD8	Ceroid lipofuscinosis, neuronal, 7
MID1	Opitz GBBB syndrome
MKS1	Meckel-Gruber syndrome, type 1
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts
MMAA	Methylmalonic aciduria, MMAA-related
MMAB	Methylmalonic aciduria, MMAB-related
MMACHC	Methylmalonic aciduria and homocystinuria, type cblC
MMADHC	Methylmalonic aciduria and homocystinuria, type cblD
MPI	Congenital disorder of glycosylation, type 1B
MPL	Thrombocytopenia, congenital amegakaryocytic
MPV17	Hepatocerebral mitochondrial DNA depletion syndrome, MPV17-related
MTHFR	Homocystinuria due to deficiency of MTHFR
MTM1	Myotubular myopathy, X-linked
MTMR2	Charcot-Marie-Tooth disease, type 4B1
MTRR	Homocystinuria, type cblE
MTTP	Abetalipoproteinemia
MUT	Methylmalonic aciduria, type mut(0)
MVK	Mevalonic aciduria; Hyper-IgD syndrome; Porokera-tosis 3, multiple types
MYO7A	Usher syndrome, type 1B
NAGA	Schindler disease; Kanzaki disease
NAGLU	Mucopolysaccharidosis, type IIIB (Sanfilippo B)
NAGS	N-acetylglutamate synthase deficiency
NBN	Nijmegen breakage syndrome
NCF1	Chronic granulomatous disease 1, autosomal recessive
NDP	Exudative vitreoretinopathy 2, X-linked; Norrie disease
NDRG1	Charcot-Marie-Tooth disease type 4D
NDUFAF5	Mitochondrial complex I deficiency, nuclear type 16
NDUFAF6	Mitochondrial complex 1 deficiency, NDUFAF6-re-lated
NDUFS4	Mitochondrial complex I deficiency
NDUFS6	Mitochondrial complex I deficiency, NDUFS6-related
NEB	Nemaline myopathy, NEB-related
NPC1	Niemann-Pick disease, type C1/D
NPC2	Niemann-Pick disease, type C2
NPHP1	Juvenile nephronophthisis
NPHS1	Congenital Finnish nephrosis
NPHS2	Steroid-resistant nephrotic syndrome
NR0B1	Congenital adrenal hypoplasia, X-linked
NR2E3	Enhanced S-cone syndrome
NTRK1	Insensitivity to pain, congenital, with anhidrosis
OAT	Ornithine aminotransferase deficiency
OCA2	Albinism
OCRL	Lowe syndrome, X-linked
OPA3	Costeff syndrome (3-methylglutaconic aciduria, type 3)
OPHN1	Intellectual developmental disorder, X-linked syndromic, Billuart type
OTC	Ornithine transcarbamylase deficiency
OTOF	Deafness; Auditory neuropathy
OXTR	Long Qt Syndrome 9; Rippling Muscle Disease 2

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GENES	CONDITIONS
P3H1	Osteogenesis imperfecta, type VIII
PAH	Phenylketonuria
PAK3	Intellectual developmental disorder, X-linked 30
PANK2	Pantothenate kinase-associated neurodegeneration
PC	Pyruvate carboxylase deficiency
PCBD1	Hyperphenylalaninemia, BH4-deficient, D
PCCA	Propionic acidemia, PCCA-related
PCCB	Propionic acidemia, PCCB-related
PCDH15	Usher syndrome, type 1F
PDE6A	Retinitis pigmentosa 43
PDHA1	Pyruvate dehydrogenase deficiency, X-linked
PDHB	Pyruvate dehydrogenase deficiency, PDHB-related
PEX1	Peroxisome biogenesis disorder 1A (Zellweger)
PEX10	Peroxisome biogenesis disorder 6A (Zellweger)
PEX12	Peroxisome biogenesis disorder 3A (Zellweger)
PEX2	Peroxisome biogenesis disorder 5A (Zellweger)
PEX6	Peroxisome biogenesis disorder 4A (Zellweger)
PEX7	Rhizomelic chondrodysplasia punctata, type 1
PFKM	Glycogen storage disease, type VII
PGK1	Phosphoglycerate kinase 1 deficiency
PHF8	Intellectual developmental disorder, X-linked syndromic, Siderius type
PHGDH	Phosphoglycerate dehydrogenase deficiency
PKHD1	Polycystic kidney disease, autosomal recessive
PLA2G6	Infantile neuroaxonal dystrophy 1
PLOD1	Ehlers-Danlos syndrome, kyphoscoliotic type, 1
PLP1	Spastic paraplegia 2, X-linked; Pelizaeus-Merzbacher disease
PMM2	Congenital disorder of glycosylation, type 1A, PMM2-related
POLG	POLG-related disorders
POLR1C	Leukodystrophy, hypomyelinating, 11; Treacher Collins syndrome 3
POMGNT1	Muscle-eye-brain disease, POMGNT1-related
POMT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, B,C, 1
POMT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, B, C, 2
POU3F4	Deafness, X-linked 2
PPT1	Ceroid lipofuscinosis, neuronal, 1
PQBP1	Renpenning syndrome
PRDX1	Methylmalonic aciduria and homocystinuria, cblC type
PREPL	Myasthenic syndrome, congenital, 22
PRF1	Hemophagocytic lymphohistiocytosis, familial, 2; Aplastic anemia; Lymphoma, non-Hodgkin
PROP1	Combined pituitary hormone deficiency 2
PRPS1	Arts syndrome, X-linked
PSAP	Metachromatic leukodystrophy, PSAP-related
PTS	6-pyruvoyl-tetrahydropterin synthase (PTPS) deficiency
PUS1	Mitochondrial myopathy and sideroblastic anemia (MLASA1)
PYGM	Glycogen storage disease, type V (McArdle disease)
QDPR	Hyperphenylalaninemia, BH4-deficient, C
RAB23	Carpenter syndrome
RAG1	Omenn syndrome, RAG1-related
RAG2	Omenn syndrome, RAG2-related
RAPSN	Congenital myasthenic syndrome, RAPSN-related
RARS2	Pontocerebellar hypoplasia, type 1 and 6, RARS2-related
RAX	Microphthalmia, isolated 3
RDH12	Leber congenital amaurosis, type RDH12
RMRP	Cartilage-hair hypoplasia
RNASEH2B	Aicardi-Goutieres syndrome 2
RP2	Retinitis pigmentosa 2
RPE65	Leber congenital amaurosis 2
RPGR	Macular degeneration, X-linked atrophic; Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness; Cone-rod dystrophy, X-linked, 1
RPGRIP1L	Ciliopathies, RPGRIP1L-related
RS1	Juvenile retinoschisis, X-linked
RTEL1	Dyskeratosis congenita, RTEL1-related
SACS	Autosomal recessive spastic ataxia of Charlevoix-Saguenay
SAMD9	MIRAGE syndrome
SAMHD1	Aicardi-Goutires syndrome
SCO2	Mitochondrial complex IV deficiency, nuclear type 2
SEPSECS	Pontocerebellar hypoplasia, type 2D
SERPINA1	Alpha-1-antitrypsin deficiency
SGCA	Limb-girdle muscular dystrophy, type 2D
SGCB	Limb-girdle muscular dystrophy, type 2E
SGCD	Limb-girdle muscular dystrophy, type 2F
SGCG	Limb-girdle muscular dystrophy, type 2C
SGSH	Mucopolysaccharidosis, type IIIA (Sanfilippo A)
SH3TC2	Charcot-Marie-Tooth disease, type 4C; Mononeuropathy of the median nerve, mild
SLC12A3	Gitelman syndrome
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy (Andermann syndrome)
SLC16A2	Allan-Herndon-Dudley syndrome
SLC17A5	Salla disease
SLC19A3	Thiamine metabolism dysfunction syndrome 2
SLC22A5	Carnitine deficiency
SLC25A13	Citrullinemia, type II
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome
SLC25A20	Carnitine-acylcarnitine translocase deficiency
SLC26A2	Achondrogenesis, type 1B
SLC26A3	Congenital chloride diarrhea
SLC26A4	Pendred syndrome
SLC35A3	Autism spectrum, epilepsy, and arthrogryposis
SLC37A4	Glycogen storage disease, type IB
SLC39A4	Acrodermatitis enteropathica
SLC3A1	Cystinuria, type A
SLC46A1	Folate malabsorption
SLC4A11	Corneal dystrophy and perceptive deafness
SLC5A5	Thyroid dyshormonogenesis 1
SLC6A19	Hartnup disorder; Hyperglycinuria; Iminoglycinuria
SLC6A8	Creatine transporter defect (cerebral creatine deficiency syndrome 1, X-linked)
SLC7A7	Cystinuria, type B
SMARCAL1	Schimke immunoosseous dysplasia
SMN1	Spinal muscular atrophy
SMPD1	Niemann-Pick disease, types A/B
SPG11	Spastic paraplegia 11; Amyotrophic lateral sclerosis 5, juvenile; Charcot-Marie-Tooth disease, axonal, type 2X
SPG7	Spastic paraplegia 7
SPINK5	Netherton syndrome
STAR	Lipoid congenital adrenal hyperplasia
EYS	Retinitis pigmentosa 25
STX11	Hemophagocytic lymphohistiocytosis, familial, 4
STXBP2	Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease
SUMF1	Multiple sulfatase deficiency
SURF1	Leigh syndrome
SYN1	Epilepsy, X-linked, with variable learning disabilities and behavior disorders; Intellectual developmental disorder, X-linked 50
TAT	Tyrosinemia, type II
TBCD	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum
TCIRG1	Osteopetrosis, infantile malignant, TCIRG1-related
TCTN2	Joubert syndrome 24
TECPR2	Hereditary spastic paraparesis, type 49
TF	Atransferrinemia
TFR2	Hemochromatosis, type 3, TFR2-related
TG	Thyroid dyshormonogenesis 3
TGM1	Lamellar ichthyosis, type 1
TH	Segawa syndrome, TH-related
THOC2	Mental retardation, X-linked 12/35
TMC1	Deafness, autosomal dominant 36, autosomal recessive 7
TMEM216	Joubert syndrome 2/Meckel syndrome 2
TNXB	Ehlers-Danlos syndrome, classic-like, 1
TPO	Congenital hypothyroidism
TPP1	Ceroid lipofuscinosis, neuronal, 2
TRDN	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness
TRIM32	Bardet-Biedl syndrome 11
TRMU	Acute infantile liver failure, TRMU-related
TSFM	Combined oxidative phosphorylation deficiency 3
TSHB	Congenital hypothyroidism
TTC37	Tricho-hepato-enteric syndrome
TTPA	Ataxia with vitamin E deficiency
TYMP	Myoneurogastrointestinal encephalopathy (MNGIE)
TYR	Oculocutaneous albinism, type 1
UGT1A1	Crigler-Najjar syndrome
UNC13D	Hemophagocytic lymphohistiocytosis, familial, 3
UPF3B	Mental retardation, syndromic 14
USH1C	Usher syndrome, type 1C
USH1G	Usher syndrome, type 1G
USH2A	Usher syndrome, type 2A
VPS13A	Choreo-acanthocytosis
VPS13B	Cohen syndrome
VPS45	Congenital neutropenia, VPS45-related
VPS53	Pontocerebellar hypoplasia, type 2E
VRK1	Pontocerebellar hypoplasia, type 1A
VSX2	Microphthalmia/Anophthalmia, VSX2-related
WAS	Wiskott-Aldrich syndrome, X-linked
WHRN	Deafness, autosomal recessive 31, Usher syndrome, type 2D
WNT10A	Odonto-onycho-dermal dysplasia/Schopf-Schulz-Passarge syndrome
XPA	Xeroderma pigmentosum group A
XPC	Xeroderma pigmentosum group C
ZDHHC9	Mental retardation, X-linked syndromic, Raymond type
ZFYVE26	Spastic paraplegia type 15
ZNF711	Mental retardation, X-linked 97