CENTOGENE
THE RARE DISEASE COMPANY

CentoScreen® Carrier Screening

DETECTS 332 AUTOSOMAL AND X-LINKED RECESSIVE DISORDERS

GENE	DISEASE	INHERITANCE	SEVERITY		
			MILD	MODERATE	SEVERE
AAAS	Achalasia-addisonianism-alacrima syndrome	AR			
ABCB11	Progressive familial intrahepatic cholestasis, type II	AR			
ABCC6	Pseudoxanthoma elasticum	AR			
ABCC8	Familial hyperinsulinemic hypoglycemia type 1	AR			
ABCD1	Adrenoleukodystrophy (X-linked)	XL			
ACADM	Medium chain Acyl-CoA dehydrogenase deficiency	AR			
ACADS	Short chain Acyl-CoA dehydrogenase deficiency	AR			
ACADSB	2-Methylbutyryl-CoA dehydrogenase deficiency	AR			
ACADVL	Very long chain Acyl-CoA dehydrogenase deficiency	AR			
ACAT1	Beta-ketothiolase deficiency (Alpha-methylacetoacetic aciduria)	AR			
ACOX1	Peroxisomal acyl-CoA oxidase deficiency	AR			
ADA	Severe combined immunodeficiency due to ADA deficiency	AR			
ADAMTS2	Ehlers Danlos syndrome, type VIIC	AR			
ADAR	Aicardi-Goutieres syndrome 6	AR			
ADGRG1	Bilateral frontoparietal polymicrogyria	AR			
AGA	Aspartylglycosaminuria	AR			
AGL	Glycogen storage disease, type III (a&b)	AR			
AGPS	Rhizomelic chondrodysplasia punctata, type III	AR			
AGXT	Primary hyperoxaluria, type I	AR			
AIRE	Polyglandular autoimmune syndrome, type I (Autoimmune polyendocrinopathy syndrome type I, with or without reversible metaphyseal dysplasia)	AR			
ALDH3A2	Sjögren-Larsson syndrome	AR			
ALDH7A1	Pyridoxine-dependent epilepsy	AR			
ALDOB	Hereditary fructose intolerance	AR			
ALG6	Congenital disorder of glycosylation, type Ic	AR			
ALPL	Hypophosphatasia, autosomal recessive	AR			
AMT	Glycine encephalopathy, AMT-related	AR			
AP1S1	MEDNIK syndrome	AR			
AP3B1	Hermansky-Pudlak syndrome, type 2	AR			
AR	Androgen insensitivity syndrome	XL			
ARSA	Metachromatic leukodystrophy	AR			
ARSB	Mucopolysaccharidosis, type VI	AR			
ASL	Argininosuccinic aciduria	AR			
ASNS	Asparagine Synthetase deficiency	AR			
ASPA	Canavan disease	AR			
ASS1	Citrullinemia, type I	AR			
ATM	Ataxia-telangiectasia	AR			
ATP13A2	Kufor-Rakeb syndrome (KRS); Autosomal recessive spastic paraplegia-78 (SPG78)	AR			
ATP6V1B1	Renal tubular acidosis and deafness, ATP6V1B1-related	AR			
ATP7B	Wilson disease	AR			
BBS1	Bardet-Biedl syndrome 1	AR			
BBS2	Bardet-Biedl syndrome 2	AR			
BBS4	Bardet-Biedl syndrome 4	AR			

MILD = Normal lifespan but affects quality of life
MODERATE = Moderately affects lifespan (early adulthood) and/or quality of life
SEVERE = Significantly affects lifespan (infancy/childhood) and quality of life

Some diseases can present with varying degrees of severity in different individuals.

AR = Autosomal recessive
XL = X-linked recessive

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GENE	DISEASE	INHERITANCE	SEVERITY		
			MILD	MODERATE	SEVERE
BBS7	Bardet-Biedl syndrome 7	AR			
BBS9	Bardet-Biedl syndrome 9	AR			
BBS10	Bardet-Biedl syndrome 10	AR			
BBS12	Bardet-Biedl syndrome 12	AR			
BCKDHA	Maple syrup urine disease, type Ia	AR			
BCKDHB	Maple syrup urine disease, type Ib	AR			
BCS1L	GRACILE syndrome; Bjornstad syndrome; Leigh syndrome; Mitochondrial complex III deficiency, nuclear type 1	AR			
BLM	Bloom syndrome	AR			
BSND	Bartter syndrome, type IV	AR			
BTD	Biotinidase deficiency	AR			
CANT1	Desbuquois dysplasia, type I; Epiphyseal dysplasia, multiple, 7	AR			
CAPN3	Limb-girdle muscular dystrophy, type 2A	AR			
CBS	Homocystinuria, CBS-related	AR			
CDH23	Usher syndrome, type ID; Deafness, autosomal recessive 12	AR			
CEP290	Leber congenital amaurosis 10; Joubert syndrome 5; Meckel syndrome 4; Senior-Loken syndrome 6	AR			
CERKL	Retinitis pigmentosa 26	AR			
CFTR	Cystic fibrosis; Congenital bilateral absence of vas deferens	AR			
CHAT	Congenital myasthenic syndrome 6	AR			
CHM	Choroideremia	AR			
CHRNE	Congenital myasthenic syndrome 4A; Congenital myasthenic syndrome 4B; Congenital myasthenic syndrome 4C	AR			
CLN3	Neuronal ceroid lipofuscinosis, CLN3-related	AR			
CLN5	Neuronal ceroid lipofuscinosis, CLN5-related	AR			
CLN6	Neuronal ceroid lipofuscinosis, CLN6-related	AR			
CLN8	Neuronal ceroid lipofuscinosis, CLN8-related; Northern epilepsy	AR			
CLRN1	Usher syndrome, type IIIA	AR			
CNGA3	Achromatopsia 2, CNGA3-related	AR			
CNGB3	Achromatopsia 3, CNGB3-related	AR			
COL4A3	Alport syndrome	AR			
COL4A4	Alport syndrome 2	AR			
COL4A5	Alport syndrome, X-linked	XL			
COL7A1	Dystrophic epidermolysis bullosa, autosomal recessive	AR			
COLQ	Congenital myasthenic syndrome 5	AR			
CPT1A	Carnitine palmitoyltransferase IA deficiency	AR			
CPT2	Carnitine palmitoyltransferase II deficiency	AR			
CRB1	Leber congenital amaurosis 8; Retinitis pigmentosa-12, autosomal recessive	AR			
CTNS	Cystinosis, atypical nephropathic; Cystinosis, late-onset juvenile or adolescent nephropathic; Cystinosis, nephropathic; Cystinosis, ocular nonnephropathic	AR			
CTSD	Neuronal ceroid lipofuscinosis 10	AR			
CTSF	Neuronal ceroid lipofuscinosis 13	AR			
CTSK	Pycnodysostosis	AR			
CYP11B1	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	AR			

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			MILD	MODERATE	SEVERE
CYP11B2	Cortisone methyl oxidase type II deficiency; Cortisone methyl oxidase type I deficiency	AR			
CYP17A1	Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	AR			
CYP19A1	Aromatase deficiency	AR			
CYP1B1	Primary congenital glaucoma 3A	AR			
CYP21A2	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	AR			
CYP27A1	Cerebrotendinous xanthomatosis	AR			
CYP27B1	Vitamin D-dependent rickets, type I	AR			
DBT	Maple syrup urine disease, type II	AR			
DCLRE1C	Omenn syndrome; Severe combined immunodeficiency, Athabascan type	AR			
DHCR7	Smith-Lemli-Opitz syndrome	AR			
DHDDS	Retinitis pigmentosa 59	AR			
DKC1	Dyskeratosis congenita, X-linked	XL			
DLD	Dihydrolipoamide dehydrogenase deficiency	AR			
DMD	Duchenne/Becker muscular dystrophy	XL			
DNAH5	Primary ciliary dyskinesia type 3, DNAH5-related	AR			
DNAI1	Primary ciliary dyskinesia type 1, DNAI1-related	AR			
DNAI2	Primary ciliary dyskinesia type 9, DNAI2-related	AR			
DNAJC5	Neuronal ceroid lipofuscinosis type 4	AR			
DOK7	Fetal akinesia deformation sequence, DOK7-related; Congenital myasthenic syndrome, 10	AR			
DPYD	Dihydropyrimidine dehydrogenase deficiency	AR			
DYSF	Limb-girdle muscular dystrophy, type 2B; Miyoshi myopathy and distal myopathy with anterior tibial onset	AR			
EDA	Hypohidrotic ectodermal dysplasia, X-linked	XL			
EDAR	Hypohidrotic ectodermal dysplasia 10B	AR			
EMD	Emery-Dreifuss muscular dystrophy	XL			
ERCC2	Xeroderma pigmentosum	AR			
ETFA	Glutaric acidemia, Type IIA	AR			
ETFB	Glutaric acidemia, Type IIB	AR			
ETFDH	Glutaric acidemia, Type IIC	AR			
ETHE1	Ethylmalonic encephalopathy	AR			
EXOSC3	Pontocerebellar hypoplasia type 1B	AR			
EYS	Retinitis pigmentosa 25, EYS-related	AR			
F8	Hemophilia A	XL			
F9	Hemophilia B	XL			
F11	Factor XI Deficiency	AR			
FAH	Tyrosinemia, type I	AR			
FAM161A	Retinitis pigmentosa 28	AR			
FANCA	Fanconi anemia, complementation group A	AR			
FANCC	Fanconi anemia, complementation group C	AR			
FANCG	Fanconi Anemia, complementation group G	AR			
FH	Fumarase deficiency	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			
FKTN	Muscular dystrophy-dystroglycanopathy type 4A (Walker-Warburg syndrome); Muscular dystrophy-dystroglycanopathy type 4B; Muscular dystrophy-dystroglycanopathy type 4C; Cardiomyopathy, dilated, 1X	AR			

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			MILD	MODERATE	SEVERE
FMR1	Fragile X syndrome; FMR1-related primary ovarian insufficiency; Fragile X-associated tremor/ataxia syndrome	XL			
G6PC	Glycogen storage disease, type IA	AR			
G6PD	Glucose-6-phosphate dehydrogenase deficiency; Hemolytic anemia due to G6PD deficiency	XL			
GAA	Glycogen storage disease, type II	AR			
GALC	Krabbe disease	AR			
GALE	Galactose epimerase deficiency	AR			
GALK1	Galactokinase deficiency	AR			
GALNT3	Hyperphospatemic tumoral calcinosis, familial	AR			
GALT	Galactosemia	AR			
GAMT	Guanidinoacetate methyltransferase deficiency	AR			
GBA	Gaucher disease	AR			
GBE1	Glycogen storage disease, type IV	AR			
GCDH	Glutaric acidemia, type I	AR			
GCSH	Nonketotic hyperglycinemia	AR			
GDF5	Du Pan syndrome; Chondrodysplasia, Grebe type; Brachydactyly type A1,C; Acromesomelic dysplasia, Hunter-Thompson type	AR			
GFPT1	Congenital myasthenic syndrome 12	AR			
GJB1	Charcot-Marie-Tooth disease, GJB1-related	XL			
GJB2	Nonsyndromic hearing loss, GJB2-related	AR			
GJB6	Nonsyndromic Hearing Loss, GJB6-related	AR			
GLA	Fabry disease	XL			
GLB1	GM1-gangliosidosis; Mucopolysaccharidosis type IVB	AR			
GLDC	Glycine encephalopathy, GLDC-related	AR			
GNE	Inclusion body myopathy, type II	AR			
GNPAT	Rhizomelic chondrodysplasia punctata, type II	AR			
GNPTAB	Mucolipidosis, type II alpha/beta; Mucolipidosis, type III alpha/beta	AR			
GNPTG	Mucolipidosis III gamma	AR			
GNS	Mucopolysaccharidosis type IIID	AR			
GORAB	Geroderma osteodysplastica	AR			
GRHPR	Primary hyperoxaluria, type II	AR			
GRN	Neuronal ceroid lipofuscinosis type 11	AR			
GUCY2D	Leber congenital amaurosis 1; Choroidal dystrophy, central areolar 1	AR			
HADH	Familial hyperinsulinemic hypoglycemia, familial 4; 3-hydroxyacyl-CoA dehydrogenase deficiency	AR			
HADHA	Long-chain 3-Hydroxyacyl-CoA dehydrogenase deficiency; Trifunctional protein deficiency	AR			
HADHB	Mitochondrial trifunctional protein deficiency	AR			
HAX1	Severe congenital neutropenia 3, autosomal recessive	AR			
HBA1	Alpha-thalassemia	AR			
HBA2	Alpha-thalassemia	AR			
HBB	Beta-thalassemia, and other hemoglobinopathies	AR			
HEPACAM	Megalencephalic Leukoencephalopathy with Subcortical Cysts, types 2A & 2B	AR			
HEXA	Tay-Sachs disease; GM2-ganlgliosidosis	AR			
HEXB	Sandhoff disease	AR			
HFE2	Hereditary hemochromatosis type 2A, HFE2-related	AR			
HGD	Alkaptonuria	AR			

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			MILD	MODERATE	SEVERE
HGSNAT	Mucopolysaccharidosis, type IIIC	AR			
HLCS	Holocarboxylase synthetase deficiency	AR			
HMGCL	3-hydroxy-3-methylglutaryl CoA lyase deficiency	AR			
HOGA1	Primary hyperoxaluria, type III	AR			
HPRT1	Lesch-Nyhan syndrome; HPRT-related gout	XL			
HPS1	Hermansky-Pudlak syndrome, type 1	AR			
HPS3	Hermansky-Pudlak syndrome, type 3	AR			
HPS4	Hermansky-Pudlak syndrome, type 4	AR			
HPS5	Hermansky-Pudlak syndrome, type 5	AR			
HPS6	Hermansky-Pudlak syndrome, type 6	AR			
HSD17B3	17-beta-hydroxysteroid dehydrogenase deficiency, type III	AR			
HSD17B4	D-bifunctional protein deficiency; Perrault syndrome 1	AR			
HSD3B2	Congential adrenal hyperplasia due to 3-Beta-hydroxysteroid dehydrogenase deficiency, type II	AR			
IDS	Mucopolysaccharidosis, type II (Hunter syndrome)	XL			
IDUA	Mucopolysachharidosis, type I (Hurler syndrome)	AR			
IKBKAP	Familial dysautonomia (HSAN3)	AR			
IL2RG	Severe combined immunodeficiency, X-linked	XL			
ISPD	Walker-Warburg (Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7	AR			
IVD	Isovaleric acidemia	AR			
KCNJ11	Familial hyperinsulinemic hypoglycemia type 2, KCNJ11-related	AR			
KCTD7	Neuronal ceroid lipofuscinosis 14 (progressive myoclonic epilepsy type 3)	AR			
L1CAM	L1 syndrome; MASA syndrome, CRASH syndrome	XL			
LAMA3	Herlitz junctional epidermolysis bullosa, LAMA3-related; Laryngoonychocutaneous syndrome; Epidermolysis bullosa, generalized atrophic benign	AR			
LAMB3	Herlitz junctional epidermolysis bullosa; non-Herlitz junctional epidermolysis bullosa	AR			
LAMC2	Herlitz junctional epidermolysis bullosa; non-Herlitz junctional epidermolysis bullosa	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			
LCA5	Leber congenital amaurosis 5, LCA5-related	AR			
LHCGR	Leydig cell hypoplasia; Luteinizing hormone resistance	AR			
LIFR	Stuve-Wiedemann syndrome	AR			
LIPA	Cholesteryl ester storage disease	AR			
LIPH	Woolly hair/hypotrichosis, autosomal recessive	AR			
LOXHD1	Autosomal recessive deafness 77	AR			
LPL	Lipoprotein lipase deficiency	AR			
LRPPRC	Leigh syndrome with COX deficiency (French Canadian type)	AR			
LYST	Chediak-Higashi syndrome	AR			
MAN2B1	Alpha-mannosidosis type I & II	AR			
MCCC1	3-Methylcrontonyl-CoA carboxylase 1 deficiency	AR			
MCCC2	3-Methylcrontonyl-CoA carboxylase 2 deficiency	AR			
MCOLN1	Mucolipidosis type IV	AR			
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy	AR			
MEFV	Familial Mediterranean fever	AR			
MFSD8	Neuronal ceroid-lipofuscinosis 7, MFSD8-related; Macular dystrophy with central cone involvement	AR			
MKKS	Bardet-Biedl syndrome 6; McKusick-Kaufman syndrome	AR			

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			MILD	MODERATE	SEVERE
MKS1	Bardet-Biedl syndrome 13; Joubert syndrome 28; Meckel syndrome 1	AR			
MLC1	Megalencephalic Leukoencephalopathy with subcortical cysts, type I	AR			
MMAA	Methylmalonic aciduria, cblA type	AR			
MMAB	Methylmalonic aciduria, cblB type	AR			
MMADHC	Methylmalonic aciduria, cblD type	AR			
MPI	Congenital disorder of glycosylation, type IB	AR			
MPL	Congenital amegakaryocytic thrombocytopenia	AR			
MPV17	Hepatocerebral mitochondrial DNA depletion syndrome 6, MPV17-related	AR			
MRE11A	Ataxia-telangiectasia-like disorder 1	AR			
MTHFR	Homocystinuria due to MTHFR deficiency, severe; Neural tube defects folate-sensitive	AR			
MTM1	Myotubular myopathy, MTM1-related	XL			
MTTP	Abetalipoproteinemia	AR			
MUT	Methylmalonic aciduria mut(0) type, MUT-related	AR			
MYO7A	Usher syndrome, type IB; Deafness, autosomal recessive 2	AR			
NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo B)	AR			
NAGS	N-acetylglutamate synthase deficiency	AR			
NBN	Nijmegen breakage syndrome	AR			
NDUFAF6	Mitochondrial complex 1 deficiency, NDUFAF6-related	AR			
NEB	Nemaline myopathy 2	AR			
NPC1	Niemann-pick disease, type C1	AR			
NPC2	Niemann-pick disease, type C2	AR			
NPHS1	Nephrotic syndrome, type I	AR			
NPHS2	Nephrotic syndrome, type II	AR			
NR2E3	Enhanced S-cone syndrome	AR			
NTRK1	Congenital insensitivity to pain with anhidrosis	AR			
OPA3	3-methylglutaconic aciduria, type III	AR			
OTC	Ornithine transcarbamylase deficiency	XL			
PAH	Phenylalanine hydroxylase deficiency	AR			
PCCA	Propionic acidemia, PCCA-related	AR			
PCCB	Propionic acidemia, PCCB-related	AR			
PCDH15	Usher syndrome, type IF; Deafness, autosomal recessive 23	AR			
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency, X-Linked	XL			
PDHB	Pyruvate dehydrogenase E1-beta deficiency, autosomal recessive	AR			
PEPD	Prolidase deficiency	AR			
PET100	Mitochondrial complex IV deficiency	AR			
PEX1	Zellweger syndrome spectrum, PEX1-related	AR			
PEX2	Zellweger syndrome spectrum, PEX2-related	AR			
PEX6	Zellweger syndrome spectrum, PEX6-related	AR			
PEX7	Rhizomelic chondrodysplasia punctata, type I; Peroxisome biogenesis disorder 9B	AR			
PEX10	Zellweger syndrome spectrum, PEX10- related	AR			
PEX12	Zellweger syndrome spectrum, PEX12- related	AR			
PEX26	Zellweger syndrome spectrum, PEX26- related	AR			
PFKM	Glycogen storage disease, type VII	AR			
PHGDH	3-phosphoglycerate dehydrogenase deficiency; Neu-Laxova syndrome 1	AR			
PKHD1	Polycystic kidney disease, autosomal recessive	AR			

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			MILD	MODERATE	SEVERE
PMM2	Congenital disorder of glycosylation, type IA	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			
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			
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			
POMT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2	AR			
PPT1	Neuronal ceroid lipofuscinosis 1, PPT1-related	AR			
PRPS1	Arts Syndrome; Charcot-Marie-Tooth disease, PRPS1-related; Gout, PRPS1-related	XL			
PSAP	Metachromatic leukodystrophy due to SAP-b deficiency; Atypical Gaucher disease; Atypical Krabbe disease	AR			
PUS1	Mitochondrial myopathy and sideroblastic anemia 1	AR			
PYGL	Glycogen storage disease VI	AR			
PYGM	Glycogen storage disease, type V	AR			
RAB23	Carpenter syndrome	AR			
RAG1	Severe combined immunodeficiency, RAG1-related; Omenn syndrome; Combined cellular and humoral immune defects with granulomas	AR			
RAG2	Omenn syndrome; Severe combined immunodeficiency, Athabascan type	AR			
RAPSN	Congenital myasthenic syndrome 11, RAPSN-related; Fetal akinesia deformation sequence	AR			
RARS2	Pontocerebellar hypoplasia type 6	AR			
RDH12	Leber congenital amaurosis 13	AR			
RNASEH2A	Aicardi-Goutieres syndrome 4	AR			
RNASEH2B	Aicardi-Goutieres syndrome 2	AR			
RNASEH2C	Aicardi-Goutieres syndrome 3, RNASEH2C-related	AR			
RPE65	Leber congenital amaurosis 2; Retinitis pigmentosa 20	AR			
RS1	Juvenile retinoschisis, X-linked	XL			
RTEL1	Dyskeratosis congenita, autosomal recessive 5	AR			
SACS	Spastic ataxia of Charlevoix-Saguenay, autosomal recessive	AR			
SAMD9	Familial tumoral calcinosis, normophosphatemic	AR			
SAMHD1	Aicardi-Goutieres syndrome 5	AR			
SBDS	Shwachman-Diamond syndrome	AR			
SEPSECS	Pontocerebellar hypoplasia 2D	AR			
SERPINA1	Alpha-1 antitrypsin deficiency	AR			
SGCA	Limb-girdle muscular dystrophy, type 2D	AR			
SGCB	Limb-girdle muscular dystrophy, type 2E	AR			
SGCG	Limb-girdle muscular dystrophy, type 2C	AR			
SGSH	Mucopolysaccharidosis type IIIA (Sanfilippo A)	AR			
SLC12A3	Gitelman syndrome	AR			
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy (Andermann syndrome)	AR			
SLC17A5	Salla disease; Infantile sialic acid storage disorder	AR			
SLC22A5	Primary carnitine deficiency	AR			
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome (Ornithine translocase deficiency)	AR			

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SLC26A2	Sulfate transporter-related osteochondrodysplasia; Achondrogenesis Ib; Atelosteogenesis II; Diastrophic dysplasia; Epiphyseal dysplasia, multiple, 4	AR			
SLC26A4	Pendred syndrome; Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	AR			
SLC35A3	Arthrogryposis, mental retardation and seizures	AR			
SLC37A4	Glycogen storage disease, type 1b; Gycogen storage disease type 1c	AR			
SLC39A4	Acrodermatitis enteropathica	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			
SMN1	Spinal muscular atrophy	AR			
SMPD1	Niemann-Pick disease type A; Niemann-Pick disease type B	AR			
ST3GAL5	Amish infantile epilepsy syndrome	AR			
STAR	Congenital lipoid adrenal hyperplasia	AR			
STS	X-linked ichthyosis	XL			
SUMF1	Multiple sulphatase deficiency	AR			
TAT	Tyrosinemia type II (Richner-Hanhart syndrome)	AR			
TCIRG1	Osteopetrosis type 1, infantile malignant	AR			
TECPR2	Hereditary spastic paraparesis, type 49	AR			
TFR2	Hereditary hemochromatosis type 3, TFR2-related	AR			
TGM1	Lamellar ichthyosis type I	AR			
TH	Segawa syndrome (tyrosine hydroxylase deficiency)	AR			
TMEM216	Joubert syndrome 2; Meckel syndrome 2	AR			
TPP1	Neuronal ceroid lipofuscinosis 2, TPP1-related; Spinocerebellar ataxia type 7	AR			
TREX1	Aicardi-Goutieres syndrome type 1, TREX1-related	AR			
TRIM37	Mulibrey nanism	AR			
TSEN2	Pontocerebellar hypoplasia type 2B	AR			
TSEN34	Pontocerebellar hypoplasia type 2C	AR			
TSEN54	Pontocerebellar hypoplasia type 2A; pontocerebellar hypoplasia type 4; Pontocerebellar hypoplasia type 5	AR			
TTC8	Bardet-Biedl syndrome 8; Retinitis pigmentosa 51	AR			
TTN	Early onset myopathy with fatal cardiomyopathy; Limb-girdle muscualr dystrophy 2J; Salih myopathy	AR			
TTPA	Ataxia with vitamin E deficiency	AR			
UBR1	Johanson-Blizzard syndrome	AR			
UGT1A1	Crigler-Najjar syndrome, type I; Crigler-Najjar syndrome, type II; Hyperbilirubinemia, familial transient neonatal Gilbert syndrome	AR			
USH1C	Usher syndrome, type IC; Deafness, autosomal recessive 18A	AR			
USH2A	Usher syndrome, Type 2A; Retinitis pigmentosa 39	AR			
VPS13A	Choreoacanthocytosis	AR			
VPS53	Pontocerebellar hypoplasia 2E	AR			
VRK1	Pontocerebellar hypoplasia, type 1A	AR			
XPA	Xeroderma pigmentosum group A	AR			
XPC	Xeroderma pigmentosum group C	AR			
ZFYVE26	Spastic paraplegia type 15, ZFYVE26-related	AR			

The 34 genes we evaluate by copy number variation (CNV) include:
ABCC6, ALDH3A2, COL4A5, CTNS, DBT, DMD, EDA, F8, FANCA, FKTN, GAA, GALC, GBE1, GJB6, GLDC, HBA1, HBA2, HBB, HEXB, HPRT1, HPS3, HSD17B4, IDS, MCOLN1,NEB, OTC, PAH, PCCA, PCDH15, PDHA1, RAPSN, SGCB, STS, and XPC.