PATERNITY For Life

Focusing on
Personalised Medicine

EXTENDED CARRIER SCREENING

Peace of Mind for Planned Pregnancies

Extended carrier screening is an important tool for prospective parents to help them determine their risk of having a child affected with a heritable disease. In many cases, parents aren't aware they are carriers and have no family history due to the rarity of some diseases in the general population.

What is covered by the screening?

Paternity For Life offers a comprehensive Extended Carrier Screening test, providing prospective parents with the information they require when planning their pregnancy. Extended Carrier Screening has been shown to detect carriers who would not have been considered candidates for traditional risk-based screening. With a simple mouth swab collection, we are able to test for over 419 genes associated with inherited diseases, including Fragile X Syndrome, Cystic Fibrosis and Spinal Muscular Atrophy. The assay has been developed in conjunction with clinical molecular geneticists, and includes genes listed in the NIH Genetic Test Registry. For a list of genes and disorders covered, please see the reverse of this brochure. If your gene of interest is not covered on our Extended Carrier Screening panel, please contact our friendly team to assist you in finding a gene test panel that suits your needs.

Why have Extended Carrier Screening?

Extended Carrier Screening prior to pregnancy enables couples to learn about their reproductive risk and consider a complete range of reproductive options, including whether or not to become pregnant, whether to use advanced reproductive technologies, such as preimplantation genetic diagnosis, or to use donor gametes. Screening also allows couples to consider prenatal diagnosis and pregnancy management options in the event of an affected fetus.

Whilst individually each disease tested is rare, around 25% of people will carry at least one abnormal mutation. These disorders are usually autosomal recessive, which means that a child must inherit a defective gene from each parent to have the disease. For autosomal recessive conditions, if a person is a carrier of the disease, they have one defective copy of the gene and one normal copy and typically don't have any symptoms of the disease. If both you and your partner are carriers of an autosomal recessive disorder, your child will have a 1 in 4 chance of inheriting one defective gene from each of you and being born with the disease. Approximately 1-2 of every 100 couples within the general population are at risk of being a carrier for an autosomal recessive genetic condition.[1]

Extended Carrier Screening for couples who are considering starting a family, aims to identify if each individual may be a carrier for the same autosomal recessive or X-linked genetic condition.

Why choose Paternity For Life?

At Paternity for Life we understand the sensitivities surrounding testing and we can assure you we strive to provide all of our patients with the highest level of professionalism, care and compassion. We offer an easy mouth swab collection kit that can be done in the comfort of your home and our laboratory handles your sample from start to finish.

Download Request Form and complete and return to Paternity For Life → Receive testing kit from Paternity for Life → Paternity For Life Receives sample for testing → Paternity For Life completes requested testing → Results sent to nominated health care professional

Email: admin@paternityforlife.co.uk | Web: www.paternityforlife.co.uk

Condition	Gene	Condition	Gene
Progressive Familial Intrahepatic Cholestasis, Type 3	*ABCB4*	Maple Syrup Urine Disease, Type 1A	*BCKDHA*
Pseudoxanthoma elasticum	*ABCC6*	Maple Syrup Urine Disease, Type 1B	*BCKDHB*
Familial Hyperinsulinism, ABCC8-Related	*ABCC8*	GRACILE Syndrome	*BCS1L*
Adrenoleukodystrophy, X-Linked	*ABCD1*	Bloom Syndrome	*BLM*
Mitochondrial Complex I Deficiency, ACAD9-Related	*ACAD9*	Fanconi anemia, Group J	*BRIP1*
Medium Chain Acyl-CoA Dehydrogenase Deficiency	*ACADM*	Bartter syndrome, Type 4a	*BSND*
Short Chain Acyl-CoA Dehydrogenase Deficiency	*ACADS*	Biotinidase Deficiency	*BTD*
Short/branched chain acyl-CoA dehydrogenase	*ACADSB*	Isolated growth hormone deficiency, Type III, X-linked	*BTK*
Very Long-Chain Acyl-CoA Dehydrogenase Deficiency	*ACADVL*	Desbuquois dysplasia 1	*CANT1*
Beta-Ketothiolase Deficiency	*ACAT1*	Limb-Girdle Muscular Dystrophy, Type 2A	*CAPN3*
Acyl-CoA Oxidase I Deficiency	*ACOX1*	Catecholaminergic polymorphic ventricular tachycardia	*CASQ2*
Combined Malonic and Methylmalonic Aciduria	*ACSF3*	Homocystinuria, CBS-Related	*CBS*
Severe Combined Immunodeficiency, ADA-Related	*ADA*	Mental retardation, autosomal recessive 3	*CC2D1A (CNV)*
Ehlers-Danlos Syndrome, Type VIIC	*ADAMTS2*	Usher Syndrome, Type 1D	*CDH23*
Bilateral Frontoparietal Polymicrogyria	*ADGRG1*	Leber Congenital Amaurosis, Type CEP290	*CEP290*
Aspartylglucosaminuria	*AGA*	Retinitis Pigmentosa 26	*CERKL*
Glycogen Storage Disease, Type III (Cori/Forbes)	*AGL*	Cystic Fibrosis	*CFTR (CNV)*
Rhizomelic Chondrodysplasia Punctata, Type 3	*AGPS*	Choroideremia, X-Linked	*CHM*
Hyperoxaluria, Primary, Type 1	*AGXT*	Congenital Myasthenic Syndrome, CHRNE-Related	*CHRNE*
Autoimmune polyendocrinopathy syndrome, type I	*AIRE*	Escobar Syndrome	*CHRNG*
Sjogren-Larsson Syndrome	*ALDH3A2*	Bare Lymphocyte Syndrome, CIITA-Related	*CIITA*
Pyridoxine-dependent epilepsy	*ALDH7A1*	Ceroid Lipofuscinosis, Neuronal, 3	*CLN3 (CNV)*
Hereditary Fructose Intolerance	*ALDOB*	Ceroid Lipofuscinosis, Neuronal, 5	*CLN5*
Congenital Disorder of Glycosylation, Type 1C	*ALG6*	Ceroid Lipofuscinosis, Neuronal, 6	*CLN6*
Alstrom Syndrome	*ALMS1*	Ceroid Lipofuscinosis, Neuronal, 8 (a.ka. Northern Epilepsy)	*CLN8*
Hypophosphatasia, ALPL-Related	*ALPL*	Usher Syndrome, Type 3	*CLRN1*
Persistent Müllerian duct syndrome type 1	*AMH*	Achromatopsia, CNGA3-Related	*CNGA3*
Persistent Müllerian duct syndrome type 2	*AMHR2*	Achromatopsia, CNGB3-Related	*CNGB3*
Glycine Encephalopathy, AMT-Related	*AMT*	Fibrochondrogenesis type 2	*COL11A2*
Mental retardation, enteropathy, deafness, peripheral neuropathy, ichthyosis, and keratoderma (MEDNIK)	*AP1S1*	Alport Syndrome, COL4A3-Related	*COL4A3*
Familial Nephrogenic Diabetes Insipidus, AQP2-Related	*AQP2*	Alport Syndrome, COL4A4-Related	*COL4A4*
Androgen insensitivity syndrome, X-Linked	*AR*	Alport Syndrome, X-Linked	*COL4A5*
Argininemia	*ARG1*	Dystrophic Epidermolysis Bullosa, COL7A1-Related	*COL7A1*
Metachromatic Leukodystrophy, ARSA-Related	*ARSA*	Carbamoyl Phosphate Synthetase I Deficiency	*CPS1*
Mucopolysaccharidosis, Type VI (Maroteaux-Lamy)	*ARSB*	Carnitine Palmitoyltransferase IA Deficiency	*CPT1A*
Argininosuccinate Lyase Deficiency	*ASL*	Carnitine Palmitoyltransferase II Deficiency	*CPT2*
Asparagine Synthetase Deficiency	*ASNS*	Leber congenital amaurosis 8	*CRB1*
Canavan Disease	*ASPA*	Cystinosis	*CTNS (CNV)*
Citrullinemia, Type 1	*ASS1*	Papillon-Lefevre Syndrome	*CTSC*
Ataxia-Telangiectasia	*ATM (CNV)*	Ceroid Lipofuscinosis, Neuronal, 10 (CLN10 Disease)	*CTSD*
Renal Tubular Acidosis and Deafness, ATP6V1B1-Related	*ATP6V1B1*	Pycnodysostosis	*CTSK*
Menkes Syndrome, X-Linked	*ATP7A*	Chronic Granulomatous Disease, CYBA-Related	*CYBA*
Wilson Disease	*ATP7B*	Chronic Granulomatous Disease, X-Linked	*CYBB*
Progressive Familial Intrahepatic Cholestasis, Type 1	*ATP8B1*	Congenital Adrenal Hyperplasia, 11-beta-hydroxylase-deficient	*CYP11B1*
Alpha-Thalassemia Intellectual Disability Syndrome, X-Linked	*ATRX*	Corticosterone Methyloxidase Deficiency	*CYP11B2*
Bardet-Biedl Syndrome 1	*BBS1*	Congenital Adrenal Hyperplasia, 17-Alpha-Hydroxylase Deficiency	*CYP17A1*
Bardet-Biedl Syndrome 10	*BBS10*	Aromatase Deficiency	*CYP19A1*
Bardet-Biedl Syndrome 12	*BBS12*	Primary Congenital Glaucoma	*CYP1B1*
Bardet-Biedl Syndrome 2	*BBS2*	Congenital Adrenal Hyperplasia, 21-hydroxylase-deficient	CYP21A2 (CNV)
Bardet-Biedl Syndrome 4	*BBS4 (CNV)*	Cerebrotendinous Xanthomatosis	*CYP27A1*
Bardet-Biedl Syndrome 9	*BBS9*	Vitamin D-dependent rickets type 1A	*CYP27B1*
Pseudocholinesterase Deficiency	*BCHE*	Maple Syrup Urine Disease, Type 2	*DBT*

Condition	Gene	Condition	Gene
Severe Combined Immunodeficiency, Type Athabaskan	*DCLRE1C*	Galactose epimerase deficiency	*GALE*
Xeroderma Pigmentosum Group E	*DDB2*	Galactokinase Deficiency (Galactosemia, Type II)	*GALK1*
Smith-Lemli-Opitz Syndrome	*DHCR7*	Mucopolysaccharidosis, Type IVA	*GALNS*
Retinitis Pigmentosa 59	*DHDDS*	Hyperphosphatemic familial tumoral calcinosis	*GALNT3*
Dyskeratosis congenita, X-Linked	*DKC1*	Galactosemia	*GALT (CNV)*
Dihydrolipoamide Dehydrogenase Deficiency	*DLD*	Guanidinoacetate Methyltransferase Deficiency	*GAMT*
Duchenne/Becker Muscular Dystrophy	*DMD (CNV)*	Gaucher Disease	*GBA*
Ciliary Dyskinesia, Primary 3	*DNAH5*	Glycogen Storage Disease, Type IV	*GBE1*
Ciliary Dyskinesia, Primary 1	*DNAI1*	Glutaric Acidemia, Type 1	*GCDH*
Ciliary Dyskinesia, Primary 9	*DNAI2*	Dopa-responsive dystonia	*GCH1*
Ciliary Dyskinesia, Primary, 16	*DNAL1*	Grebe syndrome	*GDF5*
Congenital Myasthenic Syndrome, DOK7-Related	*DOK7*	Combined Oxidative Phosphorylation Deficiency 1	*GFM1*
Dihydropyrimidine Dehydrogenase Deficiency	*DPYD*	Isolated growth hormone deficiency, Type IA/II	*GH1 (CNV)*
Limb-Girdle Muscular Dystrophy, Type 2B	*DYSF*	Isolated growth hormone deficiency, Type IB	*GHRHR*
Hypohidrotic Ectodermal Dysplasia, X-Linked	*EDA*	Charcot-Marie-Tooth Disease with Deafness, X-Linked	*GJB1*
Hypohidrotic Ectodermal Dysplasia	*EDAR*	Non-Syndromic Hearing Loss (a.k.a. Connexin 26)	*GJB2*
Wolcott-Rallison Syndrome	*EIF2AK3*	Erythrokeratodermia variabilis et progressiva	*GJB3*
Leukoencephalopathy with Vanishing White Matter	*EIF2B5*	Non-Syndromic Hearing Loss (a.k.a. Connexin 30)	*GJB6 (CNV)*
Dysautonomia, familial	*ELP1*	Fabry Disease	*GLA (CNV)*
Emery-Dreifuss Muscular Dystrophy 1, X-Linked	*EMD*	Mucopolysaccharidosis, Type IVB / GM1 Gangliosidosis	*GLB1*
Xeroderma Pigmentosum Group D	*ERCC2*	Glycine Encephalopathy, GLDC-Related	*GLDC*
Xeroderma Pigmentosum Group B	*ERCC3*	Lethal Congenital Contracture Syndrome 1	*GLE1*
Xeroderma Pigmentosum Group F	*ERCC4*	Inclusion Body Myopathy 2	*GNE*
Xeroderma pigmentosum Group G	*ERCC5*	Mucolipidosis II/IIIA	*GNPTAB*
Cockayne syndrome, type B	*ERCC6*	Mucolipidosis III gamma	*GNPTG*
Cockayne syndrome, type A	*ERCC8*	Mucopolysaccharidosis, Type IIID (Sanfilippo D)	*GNS*
Roberts Syndrome	*ESCO2*	Geroderma osteodysplastica	*GORAB*
Glutaric Acidemia, Type 2A	*ETFA*	Bernard-Soulier Syndrome, Type A2	*GP1BA*
Glutaric Acidemia, Type 2B	*ETFB*	Bernard-Soulier Syndrome, Type B	*GP1BB*
Glutaric Acidemia, Type 2C	*ETFDH*	Bernard-Soulier Syndrome, Type C	*GP9*
Ethylmalonic Encephalopathy	*ETHE1*	Primary Hyperoxaluria, Type 2	*GRHPR*
Ellis-van Creveld Syndrome, EVC-Related	*EVC*	Leber congenital amaurosis 1	*GUCY2D*
Ellis-van Creveld Syndrome, EVC2-related	*EVC2*	Mucopolysaccharidosis, Type VII	*GUSB*
Pontocerebellar Hypoplasia, Type 1B	*EXOSC3*	Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	*HADHA*
Retinitis Pigmentosa 25	*EYS*	Trifunctional protein deficiency	*HADHB*
Factor XI deficiency	*F11*	Congenital Neutropenia, HAX1-Related	*HAX1*
Prothrombin deficiency	*F2*	Alpha-Thalassemia	*HBA1 (CNV)*
Hemophilia A	*F8*	Alpha-Thalassemia	*HBA2 (CNV)*
Hemophilia B	*F9*	Beta-Hemoglobinopathies	*HBB (CNV)*
Tyrosinemia, Type I	*FAH*	Tay-Sachs Disease	*HEXA (CNV)*
Retinitis Pigmentosa 28	*FAM161A*	Sandhoff Disease	*HEXB*
Fanconi Anemia, Group A	*FANCA (CNV)*	Hemochromatosis, Type 1	*HFE*
Fanconi Anemia, Group C	*FANCC (CNV)*	Hemochromatosis, Type 2A	*HFE2*
Fanconi Anemia, Group G	*FANCG*	Alkaptonuria	*HGD*
Fumarase Deficiency	*FH*	Mucopolysaccharidosis, Type IIIC (Sanfilippo C)	*HGSNAT*
Limb-Girdle Muscular Dystrophy, Type 2I	*FKRP*	Holocarboxylase Synthetase Deficiency	*HLCS*
Walker-Warburg Syndrome, FKTN-Related	*FKTN*	3-Hydroxy-3-Methylglutaryl-Coenzyme A Lyase Deficiency	*HMGCL*
Glycogen Storage Disease, Type IA	*G6PC*	Heme Oxygenase-1 Deficiency	*HMOX1*
Glucose-6-Phosphate Dehydrogenase Deficiency*	*G6PD*	Primary Hyperoxaluria, Type 3	*HOGA1*
Glycogen Storage Disease, Type II (Pompe Disease)	*GAA (CNV)*	Tyrosinemia, Type III	*HPD*
Krabbe Disease	*GALC (CNV)*	Hermansky-Pudlak Syndrome 1	*HPS1*

Condition	Gene	Condition	Gene
17-beta hydroxysteroid dehydrogenase 3 deficiency	*HSD17B3*	Deafness, autosomal recessive, 3	*MYO15A*
D-Bifunctional Protein Deficiency	*HSD17B4*	Usher Syndrome, Type 1B	*MYO7A*
3-Beta-Hydroxysteroid Dehydrogenase Type II Deficiency	*HSD3B2*	Mucopolysaccharidosis, Type IIIB (Sanfilippo B)	*NAGLU*
Hydrolethalus Syndrome	*HYLS1*	N-acetylglutamate Synthase Deficiency	*NAGS*
Mucopolysaccharidosis, Type II (Hunter Syndrome)	*IDS*	Nijmegen Breakage Syndrome	*NBN*
Mucopolysaccharidosis, Type I (Hurler Syndrome)	*IDUA*	Charcot-Marie-Tooth Disease type 4D	*NDRG1*
Severe Combined Immunodeficiency, X-Linked	*IL2RG*	Mitochondrial Complex I Deficiency, NDUFAF5-Related	*NDUFAF5*
Glanzmann thrombasthenia	*ITGB3 (CNV)*	Mitochondrial complex I deficiency	*NDUFS4*
Isovaleric Acidemia	*IVD*	Mitochondrial Complex I Deficiency, NDUFS6-Related	*NDUFS6*
Congenital Hyperinsulinism, KCNJ11-Related	*KCNJ11*	Nemaline Myopathy, NEB-Related	*NEB (CNV)*
LAMA2-related Muscular Dystrophy	*LAMA2*	Sialidosis	*NEU1*
Herlitz Junctional Epidermolysis Bullosa, LAMA3-Related	*LAMA3*	Hydatidiform Mole, Recurrent	*NLRP7*
Herlitz Junctional Epidermolysis Bullosa, LAMB3-Related	*LAMB3*	Niemann-Pick Disease, Type C1/D	*NPC1*
Herlitz Junctional Epidermolysis Bullosa, LAMC2-Related	*LAMC2*	Niemann-Pick Disease, Type C2	*NPC2*
Leber Congenital Amaurosis, Type LCA5	*LCA5*	Juvenile Nephronophthisis	*NPHP1*
Familial Hypercholesterolemia, LDLR-Related	*LDLR*	Congenital Finnish Nephrosis	*NPHS1*
Familial Hypercholesterolemia, LDLRAP1-Related	*LDLRAP1*	Steroid-Resistant Nephrotic Syndrome	*NPHS2*
Leydig cell hypoplasia	*LHCGR*	Congenital Adrenal Hypoplasia, X-linked	*NR0B1*
Stuve-Wiedemann Syndrome	*LIFR*	Enhanced S-Cone Syndrome	*NR2E3*
Lysosomal Acid Lipase Deficiency	*LIPA*	Congenital Insensitivity to Pain with Anhidrosis (CIPA)	*NTRK1*
Woolly Hair/Hypotrichosis Syndrome	*LIPH*	Ornithine Aminotransferase Deficiency	*OAT*
Deafness, Autosomal Recessive 77	*LOXHD1*	Lowe syndrome, X-Linked	*OCRL*
Lipoprotein Lipase Deficiency	*LPL*	Costeff Syndrome (3-Methylglutaconic Aciduria, Type 3)	*OPA3*
Leigh Syndrome, French-Canadian Type	*LRPPRC*	Ornithine Transcarbamylase Deficiency	*OTC*
Chediak-Higashi syndrome	*LYST*	Phenylketonuria	*PAH (CNV)*
Alpha-Mannosidosis	*MAN2B1*	Pantothenate Kinase-Associated Neurodegeneration	*PANK2*
Hypermethioninemia	*MAT1A*	Pyruvate Carboxylase Deficiency	*PC*
3-Methylcrotonyl-CoA Carboxylase 1 Deficiency	*MCCC1*	Propionic Acidemia, PCCA-Related	*PCCA*
3-Methylcrotonyl-CoA Carboxylase 2 Deficiency	*MCCC2*	Propionic Acidemia, PCCB-Related	*PCCB*
Mucolipidosis, Type IV	*MCOLN1 (CNV)*	Usher Syndrome, Type 1F	*PCDH15 (CNV)*
RETT Syndrome	*MECP2*	Pyruvate Dehydrogenase Deficiency, X-Linked	*PDHA1*
Microcephaly, postnatal progressive, with seizures and brain atrophy	*MED17*	Pyruvate Dehydrogenase Deficiency, PDHB-Related	*PDHB*
Familial Mediterranean Fever	*MEFV*	Prolidase deficiency	*PEPD*
Spondylothoracic Dysostosis, MESP2-Related	*MESP2*	Cytochrome-c oxidase deficiency	*PET100*
Ceroid Lipofuscinosis, Neuronal, 7	*MFSD8*	Peroxisome Biogenesis Disorder 1A (Zellweger)	*PEX1*
Bardet-Biedl Syndrome 6	*MKKS*	Peroxisome Biogenesis Disorder 6A (Zellweger)	*PEX10*
Meckel-Gruber Syndrome, Type 1	*MKS1*	Peroxisome Biogenesis Disorder 3A (Zellweger)	*PEX12*
Megalencephalic Leukoencephalopathy with Subcortical Cysts	*MLC1*	Peroxisome Biogenesis Disorder 5A (Zellweger)	*PEX2*
Malonyl-CoA decarboxylase deficiency	*MLYCD*	Peroxisome Biogenesis Disorder 4A (Zellweger)	*PEX6*
Methylmalonic Aciduria, MMAA-Related	*MMAA*	Rhizomelic Chondrodysplasia Punctata, Type 1	*PEX7*
Methylmalonic Aciduria, MMAB-Related	*MMAB*	Glycogen Storage Disease, Type VII	*PFKM*
Methylmalonic Aciduria and Homocystinuria, Type cblC	*MMACHC*	Phosphoglycerate Dehydrogenase Deficiency	*PHGDH*
Methylmalonic Aciduria and Homocystinuria, Type cblD	*MMADHC*	Multiple congenital anomalies-hypotonia-seizures syndrome 1	*PIGN*
Molybdenum cofactor deficiency	*MOCS1*	Polycystic Kidney Disease, Autosomal Recessive	*PKHD1*
Congenital Disorder of Glycosylation, Type 1B	*MPI*	Infantile neuroaxonal dystrophy 1	*PLA2G6*
Congenital Amegakaryocytic Thrombocytopenia	*MPL*	Congenital Disorder of Glycosylation, Type 1A, PMM2-Related	*PMM2*
Hepatocerebral Mitochondrial DNA Depletion Syndrome, MPV17-Related	*MPV17*	Pyridoxal 5'-phosphate-dependent epilepsy	*PNPO*
Ataxia-telangiectasia-like disorder 1	*MRE11*	POLG-Related Disorders	*POLG*
Homocystinuria due to Deficiency of MTHFR	*MTHFR*	Xeroderma pigmentosum Variant	*POLH*
Myotubular Myopathy, X-Linked	*MTM1*	Muscle-Eye-Brain Disease, POMGNT1-Related	*POMGNT1*
Homocystinuria, Type cblE	*MTRR*	Cytochrome P450 oxidoreductase deficiency	*POR*
Abetalipoproteinemia	*MTTP*	Ceroid Lipofuscinosis, Neuronal, 1	*PPT1*
Methylmalonic Aciduria, Type mut(0)	*MUT*	Myasthenic syndrome, congenital, 22	*PREPL (CNV)*

Condition	Gene	Condition	Gene
Combined Pituitary Hormone Deficiency 2	*PROP1*	Schimke Immunoosseous Dysplasia	*SMARCAL1*
Arts syndrome, X-Linked	*PRPS1*	Spinal Muscular Atrophy	*SMN1 (CNV)*
Metachromatic Leukodystrophy, PSAP-Related	*PSAP*	Niemann-Pick Disease, Types A/B	*SMPD1*
6-Pyruvoyl-Tetrahydropterin Synthase (PTPS) Deficiency	*PTS*	5-alpha reductase deficiency	*SRD5A2*
Mitochondrial Myopathy and Sideroblastic Anemia (MLASA1)	*PUS1*	GM3 synthase deficiency	*ST3GAL5*
Glycogen Storage Disease, Type V (McArdle Disease)	*PYGM*	Lipoid Congenital Adrenal Hyperplasia	*STAR*
Carpenter Syndrome	*RAB23*	Deafness, autosomal recessive 16	*STRC (CNV)*
Omenn Syndrome, RAG1-Related	*RAG1*	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)	*SUCLA2*
Omenn Syndrome, RAG2-Related	*RAG2*	Multiple Sulfatase Deficiency	*SUMF1*
Congenital Myasthenic Syndrome, RAPSN-Related	*RAPSN*	Leigh Syndrome	*SURF1*
Pontocerebellar Hypoplasia, Type 1 and 6, RARS2-Related	*RARS2*	Tyrosinemia, Type II	*TAT*
Leber Congenital Amaurosis, Type RDH12	*RDH12*	Osteopetrosis, Infantile Malignant, TCIRG1-Related	*TCIRG1*
Retinal Dystrophies, RLBP1-Associated	*RLBP1*	Hereditary Spastic Paraparesis, Type 49	*TECPR2*
Cartilage-Hair Hypoplasia	*RMRP*	Hemochromatosis, Type 3, TFR2-Related	*TFR2*
Aicardi-Goutieres syndrome, RNASEH2C-related	*RNASEH2C*	Lamellar Ichthyosis, Type 1	*TGM1*
Leber Congenital Amaurosis 2	*RPE65*	Segawa Syndrome, TH-Related	*TH*
Ciliopathies, RPGRIP1L-Related	*RPGRIP1L*	Deafness, autosomal dominant 36, autosomal recessive 7	*TMC1*
Juvenile Retinoschisis, X-Linked	*RS1*	Joubert Syndrome 2 / Meckel Syndrome 2	*TMEM216*
Dyskeratosis Congenita, RTEL1-Related	*RTEL1*	Congenital hypothyroidism	*TPO*
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	*SACS*	Ceroid Lipofuscinosis, Neuronal, 2	*TPP1*
MIRAGE syndrome	*SAMD9*	Aicardi-Goutieres syndrome, TREX1-related	*TREX1*
Aicardi-Goutières Syndrome	*SAMHD1 (CNV)*	Bardet-Biedl syndrome 11	*TRIM32*
Shwachman-Diamond syndrome	*SBDS*	Mulibrey nanism syndrome	*TRIM37*
Pontocerebellar Hypoplasia, Type 2D	*SEPSECS*	Acute Infantile Liver Failure, TRMU-Related	*TRMU*
Alpha-1-Antitrypsin Deficiency	*SERPINA1*	Pontocerebellar hypoplasia	*TSEN54*
Limb-Girdle Muscular Dystrophy, Type 2D	*SGCA*	Combined Oxidative Phosphorylation Deficiency 3	*TSFM*
Limb-Girdle Muscular Dystrophy, Type 2E	*SGCB*	Congenital hypothyroidism	*TSHB*
Limb-Girdle Muscular Dystrophy, Type 2F	*SGCD*	Hypothyroidism, congenital, nongoitrous, 1	*TSHR*
Limb-Girdle Muscular Dystrophy, Type 2C	*SGCG*	Tricho-Hepato-Enteric Syndrome	*TTC37*
Mucopolysaccharidosis, Type IIIA (Sanfilippo A)	*SGSH*	Familial dilated cardiomyopathy	*TTN*
Gitelman Syndrome	*SLC12A3*	Ataxia with Vitamin E Deficiency	*TTPA*
Agenesis of the Corpus Callosum with Peripheral Neuropathy (Andermann Syndrome)	*SLC12A6*	Myoneurogastrointestinal Encephalopathy (MNGIE)	*TYMP*
Salla Disease	*SLC17A5*	Oculocutaneous Albinism, Type 1	*TYR*
Megaloblastic Anemia Syndrome	*SLC19A2*	Oculocutaneous albinism, Type 3	*TYRP1*
Carnitine Deficiency	*SLC22A5*	Crigler-Najjar Syndrome	*UGT1A1*
Citrullinemia, Type II	*SLC25A13*	Beta-ureidopropionase deficiency	*UPB1*
Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH) Syndrome	*SLC25A15*	Usher Syndrome, Type 1C	*USH1C*
Carnitine-acylcarnitine translocase deficiency	*SLC25A20*	Usher Syndrome, Type 2A	*USH2A (CNV)*
Achondrogenesis, Type 1B	*SLC26A2*	Choreo-acanthocytosis	*VPS13A (CNV)*
Congenital Chloride Diarrhea	*SLC26A3*	Cohen Syndrome	*VPS13B*
Pendred Syndrome	*SLC26A4*	Congenital Neutropenia, VPS45-Related	*VPS45*
Autism Spectrum, Epilepsy and Arthrogryposis	*SLC35A3*	Pontocerebellar Hypoplasia, Type 2E	*VPS53*
Glycogen Storage Disease, Type IB	*SLC37A4*	Pontocerebellar Hypoplasia, Type 1A	*VRK1*
Acrodermatitis Enteropathica	*SLC39A4*	Microphthalmia/Anophthalmia, VSX2-Related	*VSX2*
Cystinuria, Type A	*SLC3A1 (CNV)*	Von Willebrand disease	*VWF*
Oculocutaneous albinism, Type 4	*SLC45A2*	Wiskott-Aldrich syndrome, X-Linked	*WAS*
Corneal Dystrophy and Perceptive Deafness	*SLC4A11*	Progressive Pseudorheumatoid Dysplasia	*WISP3*
Creatine Transporter Defect (Cerebral Creatine Deficiency Syndrome 1, X-Linked)	*SLC6A8*	Odonto-Onycho-Dermal Dysplasia / Schopf-Schulz-Passarge Syndrome	*WNT10A*
Lysinuric Protein Intolerance	*SLC7A7*	Werner Syndrome	*WRN*
Cystinuria, Type B	*SLC7A9*	Xeroderma pigmentosum Group A	*XPA*
		Xeroderma Pigmentosum Group C	*XPC*
		Spastic Paraplegia Type 15	*ZFYVE26*