MEDICOVER GENETICS

GENETIC TESTING FOR DONORS-RECIPIENTS

500 GENES TESTED

GENE	Disorder	SPECIFICATIONS
AAAS	Achalasia-Addisonianism-Alacrimia Syndrome	
ABCA12	Ichthyosis, Congenital, Autosomal Recessive 4B (Harlequin)	
ABCA3	Surfactant Metabolism Dysfunction, Pulmonary, 3	
ABCA4	Cone-Rod Dystrophy 3 • Fundus Flavimaculatus • Retinal Dystrophy, Early-Onset Severe • Retinitis Pigmentosa 19 • Stargardt Disease 1	
ABCB11	Cholestasis, Benign Recurrent Intrahepatic, 2 • Cholestasis, Progressive Familial Intrahepatic 2	
ABCB4	Cholestasis, Intrahepatic, of Pregnancy, 3 • Cholestasis, Progressive Familial Intrahepatic 3 • Gallbladder Disease 1	
ABCC6	Arterial Calcification, Generalized, of Infancy, 2 • Pseudoxanthoma Elasticum	
ABCC8	Hyperinsulinemic Hypoglycemia, Familial, 1, • Diabetes Mellitus, Permanent Neonatal 3, with or without Neurologic Features	
ABCD1	Adrenoleukodystrophy • Adrenomyeloneuropathy, Adult	◆
ACAD9	Mitochondrial Complex I Deficiency, Nuclear Type 20	
ACADM	Acyl-CoA Dehydrogenase, Medium Chain, Deficiency of	
ACADS	Acyl-CoA Dehydrogenase, Short-Chain, Deficiency of	
ACADSB	2-Methylbutyrylglycinuria	
ACADVL	Very Long-Chain Acyl-CoA Dehydrogenase Deficiency	
ACAT1	Alpha-Methylacetoacetic Aciduria	
ACOX1	Peroxisomal Acyl-CoA Oxidase Deficiency	
ACSF3	Combined Malonic and Methylmalonic Acidemia	
ADA	Adenosine Deaminase Deficiency, Partial • Severe Combined Immunodeficiency due to ADA Deficiency	
ADAMTS2	Ehlers-Danlos Syndrome, Dermatosparaxis Type	
ADGRG1	Polymicrogyria, Bilateral Frontoparietal	
AGA	Aspartylglucosaminuria	
AGL	Glycogen Storage Disease IIIa • Glycogen Storage Disease IIIb	
AGPS	Rhizomelic Chondrodysplasia Punctata, Type 3	
AGXT	Hyperoxaluria, Primary, Type 1	
AHI1	Joubert Syndrome 3	
AIRE	Autoimmune Polyendocrinopathy Syndrome, Type I, with or without Reversible Metaphyseal Dysplasia	
ALDH3A2	Sjogren-Larsson Syndrome	
ALDH6A1	Methylmalonate Semialdehyde Dehydrogenase Deficiency	
ALDH7A1	Epilepsy, Pyridoxine-Dependent	
ALDOA	Glycogen Storage Disease XII	
ALDOB	Fructose Intolerance, Hereditary	
ALG11	Congenital Disorder of Glycosylation, Type Ip	
ALG12	Congenital Disorder of Glycosylation, Type Ig	
ALG2	Congenital Disorder of Glycosylation, Type Ii • Myasthenic Syndrome, Congenital, 14, with Tubular Aggregates	
ALG3	Congenital Disorder of Glycosylation, Type Id	
ALG6	Congenital Disorder of Glycosylation, Type Ic	
ALG9	Congenital Disorder of Glycosylation, Type Il • Gillessen-Kaesbach-Nishimura Syndrome	
ALMS1	Alstrom Syndrome	
ALPL	Hypophosphatasia, Adult • Hypophosphatasia, Childhood • Hypophosphatasia, Infantile • Odontohypophosphatasia	
AMACR	Alpha-Methylacyl-CoA Racemase Deficiency • Bile Acid Synthesis Defect, Congenital, 4	
AMH	Persistent Mullerian Duct Syndrome, Type I	

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GENE	Disorder	SPECIFICATIONS
AMHR2	Persistent Mullerian Duct Syndrome, Type II	
AMT	Glycine Encephalopathy 2	
ANO10	Spinocerebellar Ataxia, Autosomal Recessive 10	
AP1S1	MEDNIK Syndrome	
AQP2	Diabetes Insipidus, Nephrogenic, 2	
AR	Androgen Insensitivity, • Hypospadias 1 • Spinal and Bulbar Muscular Atrophy of Kennedy	◆
ARG1	Argininemia	
ARSA	Metachromatic Leukodystrophy	
ARSB	Mucopolysaccharidosis Type VI (Maroteaux-Lamy)	
ASL	Argininosuccinic Aciduria	
ASNS	Asparagine Synthetase Deficiency	
ASPA	Canavan Disease	
ASS1	Citrullinemia	
ATM	Ataxia Telangiectasia	
ATP6V1B1	Distal Renal Tubular Acidosis 2 with Progressive Sensorineural Hearing Loss	
ATP7A	Menkes Disease, • Occipital Horn Syndrome, • Spinal Muscular Atrophy, Distal, 3	◆
ATP7B	Wilson Disease	
ATP8B1	Cholestasis, Benign Recurrent Intrahepatic • Cholestasis, Progressive Familial Intrahepatic 1	
ATRX	Intellectual Disability-Hypotonic Facies Syndrome	◆
AUH	3-Methylglutaconic Aciduria, Type I	
B4GALT1	Combined low LDL and Fibrinogen • Congenital Disorder of Glycosylation, Type IId	
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	
BBS9	Bardet-Biedl Syndrome 9	
BCHE	Butyrylcholinesterase Deficiency	
BCKDHA	Maple Syrup Urine Disease, Type Ia	
BCKDHB	Maple Syrup Urine Disease, Type Ib	
BCS1L	Bjornstad Syndrome • GRACILE Syndrome • Mitochondrial Complex III Deficiency, Nuclear Type 1	
BLM	Bloom Syndrome	
BRIP1	Fanconi Anemia, Complementation Group J	
BSND	Bartter Syndrome, Type 4A • Sensorineural Deafness with Mild Renal Dysfunction	
BTD	Biotinidase Deficiency	
BTK	Agammaglobulinemia, 1 • Isolated Growth Hormone Deficiency, Type III, with Agammaglobulinemia	◆
CAD	Developmental and Epileptic Encephalopathy 50	
CANT1	Desbuquois Dysplasia 1 • Epiphyseal Dysplasia, Multiple, 7	
CAPN3	Muscular Dystrophy, Limb-Girdle, Autosomal Recessive 1	
CASQ2	Ventricular Tachycardia, Catecholaminergic Polymorphic, 2	
CBS	Homocystinuria due to Cystathionine Beta-Synthase Deficiency	
CC2D1A	Intellectual Developmental Disorder, Autosomal Recessive 3	
CCDC115	Congenital Disorder of Glycosylation, Type IIo	
CCDC88C	Hydrocephalus, Congenital, 1	
CCN6	Progressive Pseudorheumatoid Dysplasia	
CD320	Methylmalonic Aciduria, Transient, due to Transcobalamin Receptor Defect	

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CDH23	Deafness, Autosomal Recessive 12 • Usher Syndrome, Type 1D • Usher Syndrome, Type 1D/F Digenic	
CEP290	Joubert Syndrome 5 • Meckel Syndrome 4 • Senior-Loken Syndrome 6	
CERKL	Retinitis Pigmentosa 26	
CFTR	Cystic Fibrosis	
CHM	Choroideremia	◆
CHRNE	Myasthenic Syndrome, Congenital, 4A, Slow-Channel • Myasthenic Syndrome, Congenital, 4B, Fast-Channel • Myasthenic Syndrome, Congenital, 4C, associated with Acetylcholine Receptor Deficiency	
CHRNG	Escobar Syndrome • Multiple Pterygium Syndrome, Lethal Type	
CIITA	Bare Lymphocyte Syndrome, Type II, Complementation Group A	
CLCN1	Myotonia Congenita, Recessive	
CLN3	Ceroid Lipofuscinosis, Neuronal, 3	
CLN5	Ceroid Lipofuscinosis, Neuronal, 5	
CLN6	Ceroid Lipofuscinosis, Neuronal, 6A • Ceroid Lipofuscinosis, Neuronal, 6B (Kufs Type)	
CLN8	Ceroid Lipofuscinosis, Neuronal, 8 • Ceroid Lipofuscinosis, Neuronal, 8, Northern Epilepsy Variant	
CLRN1	Usher Syndrome, Type 3A	
CNGA3	Achromatopsia 2	
CNGB3	Achromatopsia 3	
COG1	Congenital Disorder of Glycosylation, Type IIg	
COL11A2	Deafness, Autosomal Recessive 53 • Fibrochondrogenesis 2 • Otospondylomegaepiphyseal Dysplasia, Autosomal Recessive	
COL4A3	Alport Syndrome 2, COL4A3-Related	
COL4A4	Alport Syndrome 2, COL4A4-Related	
COL4A5	Alport Syndrome 1	◆
COL7A1	Epidermolysis Bullosa Dystrophica Inversa • Epidermolysis Bullosa Dystrophica, Autosomal Recessive • Epidermolysis Bullosa Dystrophica, Localisata Variant • Epidermolysis Bullosa Pruriginosa • Epidermolysis Bullosa, Pretibial • Transient Bullous of the Newborn	
CPS1	Carbamoylphosphate Synthetase I Deficiency	
CPT1A	CPT Deficiency, Hepatic, Type IA	
CPT2	CPT II Deficiency, Infantile • CPT II Deficiency, Lethal Neonatal • CPT II Deficiency, Myopathic, Stress-Induced	
CRB1	Leber Congenital Amaurosis 8 • Retinitis Pigmentosa-12	
CTNS	Cystinosis, Atypical Nephropathic • Cystinosis, Late-Onset Juvenile or Adolescent Nephropathic • Cystinosis, Nephropathic • Cystinosis, Ocular Nonnephropathic	
CTSA	Galactosialidosis	
CTSC	Haim-Munk Syndrome • Papillon-Lefevre Syndrome • Periodontitis 1, Juvenile	
CTSD	Ceroid Lipofuscinosis, Neuronal, 10	
CTSK	Pycnodysostosis	
CYBA	Chronic Granulomatous Disease 4, Autosomal Recessive	
CYBB	Chronic Granulomatous Disease • Immunodeficiency 34, Mycobacteriosis	◆
CYP11A1	Adrenal Insufficiency, Congenital, with 46 XY Sex Reversal, Partial or Complete	
CYP11B1	Adrenal Hyperplasia, Congenital, due to 11-Beta-Hydroxylase Deficiency	
CYP11B2	Hypoaldosteronism, Congenital, due to CMO I Deficiency • Hypoaldosteronism, Congenital, due to CMO II Deficiency	
CYP17A1	17,20-Lyase Deficiency, Isolated • 17-Alpha-Hydroxylase • 17,20-Lyase Deficiency	
CYP19A1	Aromatase Deficiency	
CYP1B1	Anterior Segment Dysgenesis 6, Multiple Subtypes • Glaucoma 3A, Primary Open Angle, Congenital, Juvenile, or Adult Onset	
CYP21A2	Adrenal Hyperplasia, Congenital, due to 21-Hydroxylase Deficiency • Hyperandrogenism, Nonclassic Type, due to 21-Hydroxylase Deficiency	
CYP27A1	Cerebrotendinous Xanthomatosis	
CYP27B1	Vitamin D-Dependent Rickets, Type I	
DBT	Maple Syrup Urine Disease, Type II	

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DCLRE1C	Omenn Syndrome • Severe Combined Immunodeficiency, Athabascan Type	
DDB2	Xeroderma Pigmentosum, Group E, DDB-Negative Subtype	
DDOST	Congenital Disorder of Glycosylation, Type Ir	
DHCR7	Smith-Lemli-Opitz Syndrome	
DHDDS	Retinitis Pigmentosa 59	
DKC1	Dyskeratosis Congenital	◆
DLD	Dihydrolipoamide Dehydrogenase Deficiency	
DMD	Duchenne Muscular Dystrophy • Becker Muscular Dystrophy	◆■
DNAH5	Ciliary Dyskinesia, Primary, 3, with or without Situs Inversus	
DNAI1	Ciliary Dyskinesia, Primary, 1, with or without Situs Inversus	
DNAI2	Ciliary Dyskinesia, Primary, 9, with or without Situs Inversus	
DNAJC12	Hyperphenylalaninemia, Mild, Non-BH4-Deficient	
DNAJC19	3-Methylglutaconic Aciduria, Type V	
DNAL1	Ciliary Dyskinesia, Primary, 16	
DOK7	Fetal Akinesia Deformation Sequence 3 • Myasthenic Syndrome, Congenital, 10	
DOLK	Congenital Disorder of Glycosylation, Type Im	
DPAGT1	Congenital Disorder of Glycosylation, Type Ij • Myasthenic Syndrome, Congenital, 13, with Tubular Aggregates	
DPM2	Congenital Disorder of Glycosylation, Type Iu	
DPM3	Muscular Dystrophy-Dystroglycanopathy (Limb-Girdle), Type C, 15	
DPYD	5-Fluorouracil Toxicity • Dihydropyrimidine Dehydrogenase Deficiency	
DYSF	Muscular Dystrophy, Limb-Girdle, Autosomal Recessive 2 • Miyoshi Muscular Dystrophy 1 • Myopathy, Distal, with Anterior Tibial Onset	
EDA	Ectodermal Dysplasia 1, Hypohidrotic	◆
EDAR	Ectodermal Dysplasia 10B, Hypohidrotic/Hair/Tooth Type, Autosomal Recessive	
EIF2AK3	Wolcott-Rallison Syndrome	
EIF2B5	Leukoencephalopathy with Vanishing White Matter 5, with or without Ovarian Failure	
ELP1	Dysautonomia, Familial	
EMD	Emery-Dreifuss Muscular Dystrophy 1	◆
ENO3	Glycogen Storage Disease XIII	
ERCC2	Trichothiodystrophy 1, Photosensitive • Xeroderma Pigmentosum, Group D	
ERCC3	Trichothiodystrophy 2, Photosensitive • Xeroderma Pigmentosum, Group B	
ERCC4	Fanconi Anemia, Complementation Group Q • Xeroderma Pigmentosum, Type F/Cockayne Syndrome • XFE Progeroid Syndrome	
ERCC5	Cerebrooculofacioskeletal Syndrome 3 • Xeroderma Pigmentosum, Group G/Cockayne Syndrome	
ERCC6	Cerebrooculofacioskeletal Syndrome 1 • Cockayne Syndrome, Type B • UV-Sensitive Syndrome 1	
ERCC8	Cockayne Syndrome, Type A • UV-Sensitive Syndrome 2	
ESCO2	Juberg-Hayward Syndrome • Roberts-SC Phocomelia Syndrome	
ETFA	Glutaric Acidemia IIA	
ETFB	Glutaric Acidemia IIB	
ETFDH	Glutaric Acidemia IIC	
ETHE1	Ethylmalonic Encephalopathy	
EVC	Ellis-Van Creveld Syndrome, EVC-Related	
EVC2	Ellis-Van Creveld Syndrome, EVC2-related	
EXOSC3	Pontocerebellar Hypoplasia, Type 1B	
EYS	Retinitis Pigmentosa 25	
F11	Factor XI Deficiency, Autosomal Recessive	
F2	Dysprothrombinemia • Hypoprothrombinemia	

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F8	Hemophilia A	◆
F9	Hemophilia B • Thrombophilia 8 due to Factor IX Defect	◆
FAH	Tyrosinemia, Type I	
FAM161A	Retinitis Pigmentosa 2	
FANCA	Fanconi Anemia, Complementation Group A	
FANCC	Fanconi Anemia, Complementation Group C	
FANCG	Fanconi Anemia, Complementation Group G	
FH	Fumarase Deficiency	
FKRP	Muscular Dystrophy-Dystroglycanopathy (Congenital with Brain and Eye Anomalies), Type A, 5 • Muscular Dystrophy-Dystroglycanopathy (Congenital with or without Impaired Intellectual Development), Type B, 5 • Muscular Dystrophy-Dystroglycanopathy (Limb-Girdle), Type C, 5	
FKTN	Cardiomyopathy, Dilated, 1X, • Muscular Dystrophy-Dystroglycanopathy (Congenital with Brain and Eye Anomalies), Type A, 4, • Muscular Dystrophy-Dystroglycanopathy (Congenital without impaired intellectual development), Type B, 4, • Muscular Dystrophy-Dystroglycanopathy (Limb-Girdle), Type C, 4	
FMO3	Trimethylaminuria	
FMR1	Fragile X Syndrome	◆■
FUT8	Congenital Disorder of Glycosylation with Defective Fucosylation 1	
G6PC	Glycogen Storage Disease Ia	
G6PD	Hemolytic Anemia, G6PD Deficient (Favism)	◆
GAA	Glycogen Storage Disease II	
GALC	Krabbe Disease	
GALE	Galactose Epimerase Deficiency	
GALK1	Galactokinase Deficiency with Cataracts	
GALNS	Mucopolysaccharidosis IVA	
GALNT3	Tumoral Calcinosis, Hyperphosphatemic, Familial, 1	
GALT	Galactosemia	
GAMT	Cerebral Creatine Deficiency Syndrome 2	
GBA	Gaucher Disease, Perinatal Lethal • Gaucher Disease, Type I • Gaucher Disease, Type II • Gaucher Disease, Type III • Gaucher Disease, Type IIIC	
GBE1	Glycogen Storage Disease IV • Polyglucosan Body Disease, Adult Form	
GCDH	Glutaricaciduria, Type I	
GCH1	Dystonia, DOPA-Responsive • Hyperphenylalaninemia, BH4-Deficient, B	
GDF5	Acromesomelic Dysplasia 2A • Acromesomelic Dysplasia 2B • Brachydactyly, Type A1, C	
GFM1	Combined Oxidative Phosphorylation Deficiency 1	
GH1	Growth Hormone Deficiency, Isolated, Type IA • Kowarski Syndrome	
GHRHR	Growth Hormone Deficiency, Isolated, Type IV	
GJB1	Charcot-Marie-Tooth Neuropathy, 1	◆
GJB2	Deafness, Autosomal Recessive 1A	
GJB3	Deafness, Digenic, GJB2/GJB3 • Erythrokeratodermia Variabilis et Progressiva 1	
GJB6	Deafness, Autosomal Recessive 1B • Deafness, Digenic GJB2/GJB6	
GLA	Fabry Disease • Fabry Disease, Cardiac Variant	◆
GLB1	GM1-Gangliosidosis, Type I • GM1-Gangliosidosis, Type II • GM1-Gangliosidosis, Type III • Mucopolysaccharidosis Type IVB (Morquio)	
GLDC	Glycine Encephalopathy 1	
GLE1	Congenital Arthrogryposis with Anterior Horn Cell Disease • Lethal Congenital Contracture Syndrome 1	
GM2A	GM2-Gangliosidosis, AB Variant	
GMPPA	Alacrima, Achalasia, and Impaired Intellectual Development Syndrome	
GNE	Nonaka Myopathy	

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GNPTAB	Mucolipidosis II Alpha/Beta • Mucolipidosis III Alpha/Beta	
GNPTG	Mucolipidosis III Gamma	
GNS	Mucopolysaccharidosis Type IIID	
GORAB	Geroderma Osteodysplasticum	
GP1BA	Bernard-Soulier Syndrome, Type A1 (Recessive)	
GP1BB	Bernard-Soulier Syndrome, Type B • Giant Platelet Disorder, Isolated	
GP9	Bernard-Soulier Syndrome, Type C	
GRHPR	Hyperoxaluria, Primary, Type II	
GRIP1	Fraser Syndrome 3	
GUCY2D	Cone-Rod Dystrophy 6 • Leber Congenital Amaurosis 1 • Night Blindness, Congenital Stationary, Type 1I	
GUSB	Mucopolysaccharidosis VII	
GYS1	Glycogen Storage Disease 0, Muscle	
GYS2	Glycogen Storage Disease 0, Liver	
HADH	3-Hydroxyacyl-CoA Dehydrogenase Deficiency • Hyperinsulinemic Hypoglycemia, Familial, 4	
HADHA	Fatty Liver, Acute, of Pregnancy • HELLP Syndrome, Maternal, of Pregnancy • LCHAD Deficiency • Mitochondrial Trifunctional Protein Deficiency 1	
HADHB	Mitochondrial Trifunctional Protein Deficiency 2	
HAX1	Neutropenia, Severe Congenital 3, Autosomal Recessive	
HBA1	Thalassemia, Alpha • Hemoglobin H Disease, Nondeletional	■
HBA2	Thalassemia, Alpha • Hemoglobin H Disease, Deletional and Nondeletional	■
HBB	Sickle Cell Anemia • Thalassemia, Beta	
HEXA	GM2-Gangliosidosis, Several Forms • Tay-Sachs Disease	
HEXB	Sandhoff Disease, Infantile, Juvenile, and Adult Forms	
HFE	Hemochromatosis	
HGD	Alkaptonuria	
HGSNAT	Mucopolysaccharidosis Type IIIC (Sanfilippo C) • Retinitis Pigmentosa 73	
HJV	Hemochromatosis, Type 2A	
HLCS	Holocarboxylase Synthetase Deficiency	
HMGCL	HMG-CoA Lyase Deficiency	
HMGCS2	HMG-CoA Synthase-2 Deficiency	
HMOX1	Heme Oxygenase-1 Deficiency	
HOGA1	Hyperoxaluria, Primary, Type III	
HPD	Tyrosinemia, Type III	
HPS1	Hermansky-Pudlak Syndrome 1	
HPS3	Hermansky-Pudlak Syndrome 3	
HPS4	Hermansky-Pudlak Syndrome 4	
HSD17B3	Pseudohermaphroditism, Male, with Gynecomastia	
HSD17B4	D-Bifunctional Protein Deficiency • Perrault Syndrome 1	
HSD3B2	Adrenal Hyperplasia, Congenital, due to 3-Beta-hydroxysteroid Dehydrogenase 2 Deficiency	
HYAL1	Hydrolethalus Syndrome	
HYLS1	Hydrolethalus Syndrome	
IDS	Mucopolysaccharidosis II	◆
IDUA	Mucopolysaccharidosis Ih • Mucopolysaccharidosis Ih/s • Mucopolysaccharidosis Is	
IL2RG	Combined Immunodeficiency, Moderate • Severe Combined Immunodeficiency	◆
ITGB3	Glanzmann Thrombasthenia 2	

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IVD	Isovaleric Acidemia	
KCNJ11	Hyperinsulinemic Hypoglycemia, Familial, 2	
L1CAM	Hydrocephalus, Congenital, X-linked • MASA Syndrome	◆
LAMA2	Muscular Dystrophy, Congenital, Merosin Deficient or Partially Deficient • Muscular Dystrophy, Limb-Girdle, Autosomal Recessive 23	
LAMA3	Epidermolysis Bullosa, Junctional 2A, Intermediate • Epidermolysis Bullosa, Junctional 2B, Severe • Epidermolysis Bullosa, Junctional 2C, laryngoonychocutaneous	
LAMB3	Epidermolysis Bullosa, Junctional 1A, Intermediate • Epidermolysis Bullosa, Junctional 1B, Severe	
LAMC2	Epidermolysis Bullosa, Junctional 3A, Intermediate • Epidermolysis Bullosa, Junctional 3B, Severe	
LCA5	Leber Congenital Amaurosis 5	
LDHA	Glycogen Storage Disease XI	
LDLR	Hypercholesterolemia, Familial, 1 • LDL Cholesterol Level QTL2	
LDLRAP1	Hypercholesterolemia, Familial, 4	
LHCGR	Leydig Cell Hypoplasia with Hypergonadotropic Hypogonadism • Leydig Cell Hypoplasia with Pseudohermaphroditism • Luteinizing Hormone Resistance, Female	
LHX3	Pituitary Hormone Deficiency, Combined, 3	
LIAS	Hyperglycinemia, Lactic Acidosis, and Seizures	
LIFR	Stuve-Wiedemann Syndrome/Schwartz-Jampel Type 2 Syndrome	
LIPA	Cholesteryl ester Storage Disease • Wolman Disease	
LIPH	Hypotrichosis 7 • Woolly Hair, Autosomal Recessive 2 with or without Hypotrichosis	
LOXHD1	Deafness, Autosomal Recessive 77	
LPL	Lipoprotein Lipase Deficiency	
LRP2	Donnai-Barrow Syndrome	
LRPPRC	Mitochondrial Complex IV Deficiency, Nuclear Type 5, (French-Canadian)	
LYST	Chediak-Higashi Syndrome	
MAN1B1	Rafiq Syndrome	
MAN2B1	Mannosidosis, Alpha, Types I and II	
MANBA	Mannosidosis, Beta	
MAT1A	Hypermethioninemia, Persistent, Autosomal Dominant, due to Methionine Adenosyltransferase I/III Deficiency • Methionine Adenosyltransferase Deficiency, Autosomal Recessive	
MCCC1	3-Methylcrotonyl-CoA Carboxylase 1 Deficiency	
MCCC2	3-Methylcrotonyl-CoA Carboxylase 2 Deficiency	
MCOLN1	Mucolipidosis IV	
MCPH1	Microcephaly 1, Primary, Autosomal Recessive	
MECP2	Encephalopathy, Neonatal Severe • Intellectual Developmental Disorder, Syndromic 13 • Intellectual Developmental Disorder, Lubs Type	◆
MED17	Microcephaly, Postnatal Progressive, with Seizures and Brain Atrophy	
MEFV	Familial Mediterranean Fever, Autosomal Recessive	
MESP2	Spondylocostal Dysostosis 2, Autosomal Recessive	
MFSD8	Ceroid Lipofuscinosis, Neuronal, 7 • Macular Dystrophy with Central Cone Involvement	
MGAT2	Congenital Disorder of Glycosylation, Type IIa	
MID1	Opitz GBBB Syndrome	◆
MKKS	Bardet-Biedl Syndrome 6 • McKusick-Kaufman Syndrome	
MKS1	Bardet-Biedl Syndrome 13 • Joubert Syndrome 28 • Meckel Syndrome 1	
MLC1	Megalencephalic Leukoencephalopathy with Subcortical Cysts 1	
MLYCD	Malonyl-CoA Decarboxylase Deficiency	
MMAA	Methylmalonic Acidemia, cblA Type	
MMAB	Methylmalonic Acidemia, cblB Type	

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GENE	Disorder	SPECIFICATIONS
MMACHC	Methylmalonic Acidemia and Homocystinuria, cblC Type	
MMADHC	Methylmalonic Acidemia and homocystinuria, cblD Type	
MOCS1	Molybdenum Cofactor Deficiency A	
MOGS	Congenital Disorder of Glycosylation, Type Iib	
MPDU1	Congenital Disorder of Glycosylation, Type If	
MPI	Congenital Disorder of Glycosylation, Type Ib	
MPL	Thrombocytopenia, Congenital Amegakaryocytic	
MPV17	Charcot-Marie-Tooth Disease, Axonal, Type 2EE • Mitochondrial DNA Depletion Syndrome 6 (Hepatocerebral Type)	
MRE11	Ataxia-Telangiectasia-Like Disorder 1	
MTHFR	Homocystinuria due to MTHFR Deficiency	
MTM1	Myopathy, Centronuclear	◆
MTR	Homocystinuria-Megaloblastic Anemia, cblG Complementation Type	
MTRR	Homocystinuria-Megaloblastic Anemia, cblE Type	
MTTP	Abetalipoproteinemia	
MMUT	Methylmalonic Acidemia due to Methylmalonyl-CoA Mutase Deficiency	
MVK	Hyper-IgD Syndrome • Mevalonic Aciduria	
MYO15A	Deafness, Autosomal Recessive 3	
MYO7A	Deafness, Autosomal Recessive 2 • Usher Syndrome, Type 1B	
NADK2	2,4-Dienoyl-CoA Reductase Deficiency	
NAGA	Kanzaki Disease • Schindler Disease, Type I • Schindler Disease, Type III	
NAGLU	Mucopolysaccharidosis Type IIIB (Sanfilippo B)	
NAGS	N-Acetylglutamate Synthase Deficiency	
NBN	Nijmegen Breakage Syndrome	
NDRG1	Charcot-Marie-Tooth Disease, Type 4D	
NDUFAF5	Mitochondrial Complex I Deficiency, Nuclear Type 16	
NDUFS4	Mitochondrial Complex I Deficiency, Nuclear Type 1	
NDUFS6	Mitochondrial Complex I Deficiency, Nuclear Type 9	
NEB	Arthrogryposis Multiplex Congenita 6 • Nemaline Myopathy 2, Autosomal Recessive	
NEU1	Sialidosis, Type I • Sialidosis, Type II	
NGLY1	Congenital Disorder of Deglycosylation 1	
NLRP7	Hydatidiform Mole, Recurrent, 1	
NPC1	Niemann-Pick Disease, Type C1 • Niemann-Pick Disease, Type D	
NPC2	Niemann-Pick Disease, Type C2	
NPHP1	Joubert Syndrome 4 • Nephronophthisis 1, Juvenile • Senior-Loken Syndrome-1	
NPHS1	Nephrotic Syndrome, Type 1	
NPHS2	Nephrotic Syndrome, Type 2	
NR0B1	46 XY Sex Reversal 2, Dosage-Sensitive • Adrenal Hypoplasia, Congenital	◆
NR2E3	Enhanced S-Cone Syndrome • Retinitis Pigmentosa 37	
NTRK1	Insensitivity to Pain, Congenital, with Anhidrosis	
OAT	Gyrate Atrophy of Choroid and Retina with or without Ornithinemia	
OCA2	Albinism, Brown Oculocutaneous • Albinism, Oculocutaneous, Type II	
OCRL	Dent Disease 2 • Lowe Syndrome	◆
OPA3	3-Methylglutaconic Aciduria, Type III	
OTC	Ornithine Transcarbamylase Deficiency	◆
PAH	Phenylketonuria	
PANK2	HARP Syndrome • Neurodegeneration with Brain Iron Accumulation 1	
PC	Pyruvate Carboxylase Deficiency	

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GENETIC TESTING FOR DONORS-RECIPIENTS

500 GENES TESTED

GENE	Disorder	SPECIFICATIONS
PCCA	Propionicacidemia	
PCCB	Propionicacidemia	
PCDH15	Deafness, Autosomal Recessive 23 • Usher Syndrome, Type 1D/F digenic • Usher Syndrome, Type 1F	
PDHA1	Pyruvate Dehydrogenase E1-Alpha Deficiency	◆
PDHB	Pyruvate Dehydrogenase E1-Beta Deficiency	
PEPD	Prolidase Deficiency	
PET100	Mitochondrial Complex IV Deficiency, Nuclear Type 12	
PEX1	Heimler Syndrome 1 • Peroxisome Biogenesis Disorder 1A (Zellweger) • Peroxisome Biogenesis Disorder 1B (NALD/IRD)	
PEX10	Peroxisome Biogenesis Disorder 6A (Zellweger) • Peroxisome Biogenesis Disorder 6B	
PEX11B	Peroxisome Biogenesis Disorder 14B	
PEX12	Peroxisome Biogenesis Disorder 3A (Zellweger) • Peroxisome Biogenesis Disorder 3B	
PEX13	Peroxisome Biogenesis Disorder 11A (Zellweger) • Peroxisome Biogenesis Disorder 11B	
PEX14	Peroxisome Biogenesis Disorder 13A (Zellweger)	
PEX16	Peroxisome Biogenesis Disorder 8A (Zellweger) • Peroxisome Biogenesis Disorder 8B	
PEX19	Peroxisome Biogenesis Disorder 12A (Zellweger)	
PEX2	Peroxisome Biogenesis Disorder 5A (Zellweger) • Peroxisome Biogenesis Disorder 5B	
PEX26	Peroxisome Biogenesis Disorder 7A (Zellweger) • Peroxisome Biogenesis Disorder 7B	
PEX3	Peroxisome Biogenesis Disorder 10A (Zellweger)	
PEX5	Peroxisome Biogenesis Disorder 2A (Zellweger) • Peroxisome Biogenesis Disorder 2B • Rhizomelic Chondrodysplasia Punctata, Type 5	
PEX6	Heimler Syndrome 2 • Peroxisome Biogenesis Disorder 4A (Zellweger) • Peroxisome Biogenesis Disorder 4B	
PEX7	Peroxisome Biogenesis Disorder 9B • Rhizomelic Chondrodysplasia punctata, Type 1	
PFKM	Glycogen Storage Disease VII	
PGAM2	Glycogen Storage Disease X	
PGM1	Congenital Disorder of Glycosylation, Type It	
PHGDH	Neu-Laxova Syndrome 1 • Phosphoglycerate dehydrogenase Deficiency	
PHKG2	Glycogen Storage Disease IXc	
PHYH	Refsum Disease	
PIGN	Multiple Congenital Anomalies-Hypotonia-Seizures Syndrome 1	
PKHD1	Polycystic Kidney Disease 4, with or without Hepatic Disease	
PLA2G6	Infantile Neuroaxonal Dystrophy 1 • Neurodegeneration with Brain Iron Accumulation 2B • Parkinson Disease 14, Autosomal Recessive	
PMM2	Congenital Disorder of Glycosylation, Type Ia	
PNPO	Pyridoxamine 5'-Phosphate Oxidase Deficiency	
POLG	Mitochondrial DNA Depletion Syndrome 4A (Alpers Type) • Mitochondrial DNA Depletion Syndrome 4B (MNGIE Type) • Mitochondrial Recessive Ataxia Syndrome (includes SANDO and SCAE) • Progressive External Ophthalmoplegia, Autosomal Recessive 1	
POLH	Xeroderma Pigmentosum, Variant Type	
POMGNT1	Muscular Dystrophy-Dystroglycanopathy (Congenital with Brain and Eye Anomalies) Type A, 3 • Muscular Dystrophy-Dystroglycanopathy (Congenital with Impaired Intellectual Development) Type B, 3 • Muscular Dystrophy-Dystroglycanopathy (Limb-Girdle), Type C, 3 • Retinitis Pigmentosa 76	
POR	Antley-Bixler Syndrome with Genital Anomalies and Disordered Steroidogenesis	
PPT1	Ceroid Lipofuscinosis, Neuronal, 1	
PREPL	Myasthenic Syndrome, Congenital, 22	
PRF1	Hemophagocytic Lymphohistiocytosis, Familial, 2	
PROP1	Pituitary Hormone Deficiency, Combined, 2	
PRPS1	Arts Syndrome • Charcot-Marie-Tooth Disease, 5 • Deafness, 1 • Gout, PRPS-related • Phosphoribosylpyrophosphate Synthetase Superactivity	◆

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PSAP	Combined SAP Deficiency • Krabbe Disease, Atypical • Metachromatic Leukodystrophy due to SAP-b Deficiency	
PTS	Hyperphenylalaninemia, BH4-Deficient, A	
PUS1	Myopathy, Lactic Acidosis, and Sideroblastic Anemia 1	
PYGM	McArdle Disease	
RAB23	Carpenter Syndrome	
RAG1	Combined Cellular and Humoral Immune Defects with Granulomas • Omenn Syndrome • Severe Combined Immunodeficiency, B Cell-Negative	
RAG2	Combined Cellular and Humoral Immune Defects with Granulomas • Omenn Syndrome • Severe Combined Immunodeficiency, B Cell-Negative	
RAPSN	Fetal Akinesia Deformation Sequence 2 • Myasthenic Syndrome, Congenital, 11, Associated with Acetylcholine Receptor Deficiency	
RARS2	Pontocerebellar Hypoplasia, Type 6	
RDH12	Leber Congenital Amaurosis 13	
RFT1	Congenital Disorder of Glycosylation, Type In	
RLBP1	Bothnia Retinal Dystrophy • Fundus Albipunctatus • Retinitis Punctata Albescens	
RMRP	Anauxetic Dysplasia 1 • Cartilage-Hair Hypoplasia • Metaphyseal Dysplasia without Hypotrichosis	
RNASEH2B	Aicardi-Goutieres Syndrome 2	
RNASEH2C	Aicardi-Goutieres Syndrome 3	
RPE65	Leber Congenital Amaurosis 2 • Retinitis pigmentosa 20	
RPGRIP1L	Joubert Syndrome 7 • Meckel Syndrome 5	
RS1	Retinoschisis	◆
RTEL1	Dyskeratosis Congenital, Autosomal Recessive 5	
SACS	Spastic Ataxia, Charlevoix-Saguenay Type	
SAMD9	Tumoral Calcinosis, Familial, Normophosphatemic	
SAMHD1	Aicardi-Goutieres Syndrome 5	
SBDS	Shwachman-Diamond Syndrome 1	
SCO2	Mitochondrial Complex IV Deficiency, Nuclear Type 2	
SEPSECS	Pontocerebellar Hypoplasia Type 2D	
SERAC1	3-Methylglutaconic Aciduria with Deafness, Encephalopathy, and Leigh-Like Syndrome	
SERPINA1	Emphysema due to AAT Deficiency • Emphysema-Cirrhosis, due to AAT Deficiency • Hemorrhagic Diathesis due to Antithrombin Pittsburgh	
SGCA	Muscular Dystrophy, Limb-Girdle, Autosomal Recessive 3	
SGCB	Muscular Dystrophy, Limb-Girdle, Autosomal Recessive 4	
SGCD	Muscular Dystrophy, Limb-Girdle, Autosomal Recessive 6	
SGCG	Muscular Dystrophy, Limb-Girdle, Autosomal Recessive 5	
SGSH	Mucopolysaccharidosis Type IIIA (Sanfilippo A)	
SKIC3	Trichohepatoenteric Syndrome 1	
SLC12A3	Gitelman Syndrome	
SLC12A6	Agenesis of the Corpus Callosum with Peripheral Neuropathy	
SLC17A5	Salla Disease • Sialic Acid Storage Disorder, Infantile	
SLC19A2	Thiamine-Responsive Megaloblastic Anemia Syndrome	
SLC19A3	Thiamine Metabolism Dysfunction Syndrome 2 (Biotin- or Thiamine-Responsive Encephalopathy Type 2)	
SLC22A5	Carnitine Deficiency, Systemic Primary	
SLC25A13	Citrullinemia, Adult-Onset Type II • Citrullinemia, Type II, Neonatal-Onset	
SLC25A15	Hyperornithinemia-Hyperammonemia-Homocitrullinemia Syndrome	
SLC25A20	Carnitine-Acylcarnitine Translocase Deficiency	

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SLC26A2	Achondrogenesis Ib • Atelosteogenesis, Type II • De la Chapelle Dysplasia • Diastrophic Dysplasia • Diastrophic Dysplasia, Broad Bone-Platyspondylic Variant • Epiphyseal Dysplasia, Multiple, 4	
SLC26A3	Diarrhea 1, Secretory Chloride, Congenital	
SLC26A4	Deafness, Autosomal Recessive 4, with Enlarged Vestibular Aqueduct • Pendred Syndrome	
SLC35A3	Arthrogryposis, Impaired Intellectual Development, and Seizures	
SLC37A4	Glycogen Storage Disease Ib • Glycogen Storage Disease Ic	
SLC39A4	Acrodermatitis enteropathica	
SLC3A1	Cystinuria, Type A	
SLC45A2	Albinism, Oculocutaneous, Type IV	
SLC4A11	Corneal Endothelial Dystrophy and Perceptive Deafness • Corneal Endothelial Dystrophy, Autosomal Recessive	
SLC6A8	Cerebral Creatine Deficiency Syndrome 1	◆
SLC7A7	Lysinuric Protein Intolerance	
SLC7A9	Cystinuria, Type B	
SMARCAL1	Schimke Immunoosseous Dysplasia	
SMN1	Spinal Muscular Atrophy 1 • Spinal Muscular Atrophy 2 • Spinal Muscular Atrophy 3 • Spinal Muscular Atrophy 4	■
SMPD1	Niemann-Pick Disease, Type A • Niemann-Pick Disease, Type B	
SRD5A2	Pseudovaginal Perineoscrotal Hypospadias	
ST3GAL5	Salt and Pepper Developmental Regression Syndrome	
STAR	STAR Syndrome	
STRC	Deafness, Autosomal Recessive 16	
SUCLA2	Mitochondrial DNA Depletion Syndrome 5 (Encephalomyopathic with or without Methylmalonic Aciduria)	
SUMF1	Multiple Sulfatase Deficiency	
SURF1	Charcot-Marie-Tooth Disease, Type 4K • Mitochondrial Complex IV Deficiency, Nuclear Type 1	
TAT	Tyrosinemia, Type II	
TCIRG1	Osteopetrosis, Autosomal Recessive 1	
TECPR2	Neuropathy, Hereditary Sensory and Autonomic, Type IX, with Developmental Delay	
TF	Atransferrinemia	
TFR2	Hemochromatosis, Type 3	
TGM1	Ichthyosis, Congenital, Autosomal Recessive 1	
TH	Segawa Syndrome, Recessive	
TMC1	Deafness, Autosomal Recessive 7	
TMEM216	Joubert Syndrome 2 • Meckel Syndrome 2	
TPO	Thyroid Dyshormonogenesis 2A	
TPP1	Ceroid Lipofuscinosis, Neuronal, 2 • Spinocerebellar ataxia, Autosomal Recessive 7	
TPRN	Deafness, Autosomal Recessive 79	
TREX1	Aicardi-Goutieres Syndrome 1, Dominant and Recessive	
TRIM32	Muscular dystrophy, Limb-Girdle, Autosomal Recessive 8	
TRIM37	Mulibrey Nanism	
TRIOBP	Deafness, Autosomal Recessive 28	
TRMU	Liver Failure, Transient Infantile	
TSEN54	Pontocerebellar Hypoplasia Type 2A • Pontocerebellar Hypoplasia Type 4	
TSFM	Combined Oxidative Phosphorylation Deficiency 3	
TSHB	Hypothyroidism, Congenital, Nongoitrous 4	
TSHR	Hypothyroidism, Congenital, Nongoitrous, 1	
TTN	Congenital Myopathy 5 with Cardiomyopathy • Muscular Dystrophy, Limb-Girdle, Autosomal Recessive 10	
TTPA	Ataxia with Isolated Vitamin E Deficiency	

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GENE	Disorder	SPECIFICATIONS
TYMP	Mitochondrial DNA Depletion Syndrome 1 (MNGIE Type)	
TYR	Albinism, Oculocutaneous, Type IA • Albinism, Oculocutaneous, Type IB	
TYRP1	Albinism, Oculocutaneous, Type III	
UGT1A1	Crigler-Najjar Syndrome, Type I • Crigler-Najjar Syndrome, Type II • Hyperbilirubinemia, Familial Transient Neonatal	
UPB1	Beta-Ureidopropionase Deficiency	
USH1C	Deafness, Autosomal Recessive 18A • Usher Syndrome, Type 1C	
USH1G	Usher Syndrome, Type 1G	
USH2A	Retinitis Pigmentosa 39 • Usher Syndrome, Type 2A	
VPS13A	Choreoacanthocytosis	
VPS13B	Cohen Syndrome	
VPS45	Neutropenia, Severe Congenital, 5, Autosomal Recessive	
VPS53	Pontocerebellar Hypoplasia, Type 2E	
VRK1	Pontocerebellar Hypoplasia Type 1A	
VSX2	Microphthalmia with Coloboma 3 • Microphthalmia, Isolated 2	
VWF	von Willebrand Disease, Type 3 • von Willebrand Disease, Types 2A, 2B, 2M, and 2N	
WAS	Neutropenia, Severe Congenital • Thrombocytopenia • Thrombocytopenia, Intermittent • Wiskott-Aldrich Syndrome	◆
WNT10A	Ectodermal Dysplasia 16 (Odontoonychodermal Dysplasia) • Schopf-Schulz-Passarge Syndrome • Tooth Agenesis, Selective, 4	
WRN	Werner Syndrome	
XPA	Xeroderma Pigmentosum, Group A	
XPC	Xeroderma Pigmentosum, Group C	
ZFYVE26	Spastic Paraplegia 15, Autosomal Recessive	

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