eugene

Flinders Fertility Donor Panel (550 genes)		
Gene	**Condition Name**	**Inheritance**
LDLR	Familial Hypercholesterolemia	AD, AR
LDLRAP1	Familial Hypercholesterolemia	AD, AR
ABCA12	Congenital ichthyosis, ABCA12-related	AR
ABCA3	Surfactant metabolism dysfunction, pulmonary 3	AR
ABCA4	Stargardt disease	AR
ABCB11	Progressive familial intrahepatic cholestasis	AR
ABCC8	Familial hyperinsulinism	AR
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type	AR
ACAD9	Acyl-CoA dehydrogenase-9 deficiency	AR
ACADM	Medium-chain acyl-CoA dehydrogenase deficiency	AR
ACADS	Short-chain acyl-CoA dehydrogenase deficiency	AR
ACADSB	Short branched chain acyl-CoA dehydrogenase deficiency	AR
ACADVL	Very long-chain acyl-CoA dehydrogenase deficiency	AR
ACAT1	3-ketothiolase deficiency	AR
ACOX1	Peroxisomal acyl-CoA oxidase deficiency	AR
ACSF3	Combined malonic and methylmalonic aciduria	AR
ADA	Adenosine deaminase deficiency	AR
ADAMTS2	Ehlers-Danlos syndrome, dermatosparaxis type	AR
ADGRG1	Bilateral frontoparietal polymicrogyria	AR
ADK	Hypermethioninemia due to adenosine kinase deficiency	AR
AGA	Aspartylglucosaminuria	AR
AGL	Glycogen storage disease type III	AR
AGPS	Rhizomelic chondrodysplasia punctata, type 3	AR
AGXT	Primary hyperoxaluria type 1	AR
AHCY	Hypermethioninemia due to deficiency of S-adenosylhomocysteine hydrolase	AR
AHI1	Joubert syndrome, AHI1-related	AR
AIPL1	Childhood-onset severe retinal dystrophy, AIPL1-related	AR
AIRE	Autoimmune polyendocrinopathy syndrome type I	AR
ALDH3A2	Sjogren-Larsson syndrome	AR
ALDH4A1	Hyperprolinemia type II	AR
ALDH7A1	Pyridoxine-dependent epilepsy	AR
ALDOB	Hereditary fructose intolerance	AR
ALG6	Congenital disorder of glycosylation type Ic	AR
ALMS1	Alstrom syndrome	AR
ALPL	Hypophosphatasia	AR
AMN	Megaloblastic anemia 1	AR
AMT	Glycine encephalopathy	AR
ANO10	Spinocerebellar ataxia 10	AR
AP1S1	MEDNIK syndrome	AR
AQP2	Nephrogenic diabetes insipidus	AR
ARG1	Arginase deficiency	AR
ARL13B	Joubert syndrome, ARL13B-related	AR

ARL6	ARL6-related disorders	AR
ARSA	Metachromatic leukodystrophy	AR
ARSB	Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome)	AR
ASL	Argininosuccinate lyase deficiency	AR
ASNS	Asparagine synthetase deficiency	AR
ASPA	Canavan disease	AR
ASS1	Citrullinemia	AR
ATM	Ataxia-telangiectasia	AR
ATP6V1B1	Renal tubular acidosis with deafness	AR
ATP7B	Wilson disease	AR
ATP8B1	Progressive familial intrahepatic cholestasis	AR
BBS1	Bardet-Biedl syndrome type 1	AR
BBS10	Bardet-Biedl syndrome type 10	AR
BBS12	Bardet-Biedl syndrome type 12	AR
BBS2	BBS2-related ciliopathies	AR
BBS4	Bardet-Biedl syndrome 4	AR
BCHE	Butyrylcholinesterase deficiency	AR
BCKDHA	Maple syrup urine disease type Ia	AR
BCKDHB	Maple syrup urine disease type Ib	AR
BCS1L	Mitochondrial complex III deficiency	AR
BLM	Bloom syndrome	AR
BSND	Bartter syndrome	AR
BTD	Biotinidase deficiency	AR
CANT1	Desbuquois dysplasia 1	AR
CAPN3	Limb-girdle muscular dystrophy type 2A	AR
CASQ2	Catecholaminergic polymorphic ventricular tachycardia	AR
CBS	Homocystinuria due to cystathionine beta-synthase deficiency	AR
CC2D1A	Autosomal recessive intellectual developmental disorder 3	AR
CC2D2A	Joubert syndrome 9	AR
CCDC103	Primary ciliary dyskinesia, type 17	AR
CCDC151	Primary ciliary dyskinesia, type 30	AR
CCDC39	Primary ciliary dyskinesia, type 14	AR
CCDC88C	Congenital hydrocephalus 1	AR
CD3D	Severe Combined Immunodeficiency	AR
CD3E	Severe Combined Immunodeficiency	AR
CD59	CD59 deficiency	AR
CDH23	Usher syndrome, type 1D	AR
CEP152	CEP152-related disorders	AR
CEP290	CEP290-related Ciliopathies	AR
CERKL	Retinitis pigmentosa 26	AR
CFTR	Cystic Fibrosis	AR
CHAT	Congenital myasthenic syndrome 6	AR
CHRNE	Congenital myasthenic syndrome	AR
CHRNG	Multiple pterygium syndrome	AR
CHST6	Macular corneal dystrophy	AR
CIITA	Bare lymphocyte syndrome, type II	AR
CLCN1	Autosomal recessive congenital	AR

CLN3	Neuronal ceroid lipofuscinosis	AR
CLN5	Neuronal ceroid lipofuscinosis 5	AR
CLN6	Neuronal ceroid lipofuscinosis,	AR
CLN8	Neuronal ceroid lipofuscinosis, CLN8-related	AR
CLRN1	Usher syndrome, type 3A	AR
CNGA1	Retinitis Pigmentosa, CNGA1-related	AR
CNGA3	CNGA3-related retinopathy	AR
CNGB1	Retinitis Pigmentosa, CNGB1-related	AR
CNGB3	CNGB3-related retinopathy	AR
COL11A2	COL11A2-related disorders	AR
COL17A1	Junctional epidermolysis bullosa	AR
COL27A1	Steel syndrome	AR
COL4A3	Alport syndrome, COL4A3-related	AR
COL4A4	Alport syndrome, COL4A4-related	AR
COL7A1	Dystrophic epidermolysis bullosa	AR
COX15	Mitochondrial complex IV deficiency	AR
CPS1	Carbamoylphosphate synthetase I deficiency	AR
CPT1A	Carnitine palmitoyltransferase IA deficiency	AR
CPT2	Carnitine palmitoyltransferase II deficiency	AR
CRADD	Intellectual developmental disorder with variant lissencephaly	AR
CRB1	CRB1-related retinopathy	AR
CRYL1	GJB6-CRYL1 related nonsyndromic hearing loss	AR
CTNS	Cystinosis	AR
CTSA	Galactosialidosis	AR
CTSC	Papillon-Lefevre syndrome	AR
CTSK	Pycnodysostosis	AR
CYBA	Chronic granulomatous disease	AR
CYP11A1	Congenital adrenal insufficiency	AR
CYP11B1	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	AR
CYP11B2	Corticosterone methyloxidase deficiency	AR
CYP17A1	Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	AR
CYP19A1	Aromatase deficiency	AR
CYP1B1	Primary congenital glaucoma	AR
CYP21A2	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	AR
CYP27A1	Cerebrotendinous xanthomatosis	AR
CYP27B1	Vitamin D–dependent rickets, type 1	AR
DBT	Maple syrup urine disease, type II	AR
DCAF17	Woodhouse-Sakati syndrome	AR
DCLRE1C	Severe combined immunodeficiency with sensitivity to ionizing radiation	AR
DDX11	Warsaw breakage syndrome	AR
DGUOK	Mitochondrial DNA depletion syndrome 3	AR
DHCR7	Smith-Lemli-Opitz syndrome	AR
DHDDS	Retinitis pigmentosa 59	AR
DLD	Dihydrolipoamide dehydrogenase deficiency	AR
DLL3	Spondylocostal dysostosis 1	AR
DNAH5	Primary ciliary dyskinesia, DNAH5-related	AR
DNAI1	Primary ciliary dyskinesia, DNAI1-related	AR

DNAI2	Primary ciliary dyskinesia, DNAI2-related	AR
DNAL1	Primary ciliary dyskinesia, DNAL1-related	AR
DOK7	Congenital myasthenic syndrome, DOK7-related	AR
DPYD	Dihydropyrimidine dehydrogenase deficiency	AR
DUOX2	Congenital hypothyroidism, DUOX2-related	AR
DUOXA2	Congenital hypothyroidism, DUOXA2-related	AR
DYNC2H1	Short-rib thoracic dysplasia 3 with or without polydactyly	AR
DYSF	Limb-girdle muscular dystrophy type 2B	AR
EIF2AK3	Wolcott-Rallison Syndrome	AR
EIF2B5	Leukoencephalopathy with vanishing white matter	AR
ELP1	Familial Dysautonomia	AR
ERCC2	ERCC2-related disorders	AR
ERCC6	Cockayne syndrome type A	AR
ERCC8	Cockayne syndrome type A	AR
ESCO2	Roberts syndrome	AR
ETFA	Glutaric aciduria IIA	AR
ETFB	Glutaric aciduria IIB	AR
ETFDH	Glutaric aciduria IIC	AR
ETHE1	Ethylmalonic encephalopathy	AR
EVC	EVC-related bone growth disorders	AR
EVC2	EVC2-related bone growth disorders	AR
EXOSC3	Pontocerebellar hypoplasia type 1B	AR
EYS	Retinitis pigmentosa 25	AR
F11	Factor XI deficiency	AR
F5	Factor V deficiency	AR
FAH	Tyrosinemia, type 1	AR
FAM161A	Retinitis pigmentosa 28	AR
FANCA	Fanconi anemia group A	AR
FANCC	Fanconi anemia group C	AR
FANCG	Fanconi anemia group G	AR
FBP1	Fructose-1,6-bisphosphatase deficiency	AR
FH	Fumarase deficiency	AR
FKBP10	Osteogenesis imperfecta type XI	AR
FKRP	FKRP Alpha-dystroglycanopathies	AR
FKTN	FKTN Alpha-dystroglycanopathies	AR
FMO3	Trimethylaminuria	AR
FOXRED1	Mitochondrial complex I deficiency	AR
FTCD	Glutamate formiminotransferase deficiency	AR
FUCA1	Fucosidosis	AR
G6PC	Glycogen storage disease, type 1a	AR
GAA	Pompe disease	AR
GALC	Krabbe disease	AR
GALE	Galactose epimerase deficiency	AR
GALK1	Galactokinase deficiency	AR
GALNS	Mucopolysaccharidosis IVA (Morquio syndrome A)	AR
GALNT3	Familial hyperphosphatemic tumoral calcinosis	AR
GALT	Galactosemia	AR

GAMT	Guanidinoacetate methyltransferase deficiency	AR
GATM	Cerebral creatine deficiency syndrome 3	AR
GBA	Gaucher disease	AR
GBE1	Glycogen storage disease IV	AR
GCDH	Glutaric aciduria, type I	AR
GDAP1	Charcot-Marie-Tooth disease, GDAP1-related	AR
GFM1	Combined oxidative phosphorylation deficiency, GFM1-related	AR
GHR	Growth hormone insensitivity syndrome	AR
GJB2	Nonsyndromic hearing loss 1A	AR
GLB1	GLB1-related gangliosidoses	AR
GLDC	Glycine encephalopathy, GLDC-related	AR
GLE1	Lethal congenital contracture syndrome 1	AR
GNE	Inclusion body myopathy type 2 (Nonaka myopathy)	AR
GNPAT	Rhizomelic chondrodysplasia punctata, type 2	AR
GNPTAB	Mucolipidosis II & III	AR
GNPTG	Mucolipidosis III gamma	AR
GNRHR	Hypogonadotropic hypogonadism, GNRHR-related	AR
GNS	Mucopolysaccharidosis IIID (Sanfilippo syndrome D)	AR
GORAB	Geroderma osteodysplasticum	AR
GP1BA	Bernard-Soulier syndrome type A1	AR
GP9	Bernard-Soulier syndrome type C	AR
GRHPR	Primary hyperoxaluria type II	AR
GRIP1	Fraser syndrome	AR
GSS	Glutathione synthetase deficiency	AR
GUCY2D	Leber congenital amaurosis 1	AR
GUSB	Mucopolysaccharidosis type VII	AR
HADH	Familial hyperinsulinemic hypoglycemia 4	AR
HADHA	Trifunctional protein deficiency	AR
HADHB	Trifunctional protein deficiency	AR
HAX1	Severe congenital neutropenia, HAX1-related	AR
HBA1	Alpha thalassemia	AR
HBA2	Alpha thalassemia	AR
HBB	Sickle cell disease	AR
HEXA	Tay-Sachs disease	AR
HEXB	Sandhoff disease	AR
HGD	Alkaptonuria	AR
HGSNAT	Mucopolysaccharidosis IIIC (Sanfilippo syndrome C)	AR
HJV	Hemochromatosis, type 2A	AR
HLCS	Holocarboxylase synthetase deficiency	AR
HMGCL	3-hydroxy-3-methylglutaryl-CoA lyase deficiency	AR
HOGA1	Primary hyperoxaluria type III	AR
HPD	Tyrosinemia type III	AR
HPS1	Hermansky-Pudlak syndrome 1	AR
HPS3	Hermansky-Pudlak syndrome 3	AR
HPS4	Hermansky-Pudlak syndrome 4	AR
HPS6	Hermansky-Pudlak syndrome 6	AR
HSD17B4	D-bifunctional protein deficiency	AR

HSD3B2	Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase 2 deficie	AR
HYAL1	Mucopolysaccharidosis type IX	AR
HYLS1	Hydrolethalus syndrome	AR
IDH3B	Retinitis pigmentosa, IDH3B-related	AR
IDUA	Mucopolysaccharidosis, type I (Hurler syndrome)	AR
IGHMBP2	IGHMBP2-related neuropathies	AR
INVS	Nephronophthisis 2	AR
ITGA6	Junctional epidermolysis bullosa	AR
ITGB3	Glanzmann thrombasthenia	AR
ITGB4	Junctional epidermolysis bullosa	AR
IVD	Isovaleric Acidemia	AR
IYD	Thyroid dyshormonogenesis, IYD-related	AR
JAK3	Severe combined immunodeficiency, JAK3-related	AR
KCNJ11	KCNJ11-related hyperinsulinism	AR
LAMA2	Muscular dystrophy, LAMA2-related	AR
LAMA3	Junctional epidermolysis bullosa 2	AR
LAMB3	Junctional epidermolysis bullosa, LAMB3-related	AR
LAMC2	Junctional epidermolysis bullosa, LAMC2-related	AR
LCA5	Leber congenital amaurosis 5	AR
LHX3	Combined pituitary hormone deficiency 3	AR
LIFR	Stuve-Wiedemann syndrome	AR
LIPA	Lysosomal acid lipase deficiency	AR
LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type	AR
LOXHD1	Nonsyndromic hearing loss 77	AR
LPL	Familial lipoprotein lipase deficiency	AR
LRP2	Donnai–Barrow syndrome	AR
LRPPRC	Leigh syndrome with Complex IV deficiency	AR
LYST	Chediak-Higashi syndrome	AR
MAN2B1	Alpha-Mannosidosis	AR
MANBA	Beta-Mannosidosis	AR
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency (3-MCC deficiency)	AR
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency (3-MCC deficiency)	AR
MCEE	Methylmalonyl-CoA epimerase deficiency	AR
MCOLN1	Mucolipidosis IV	AR
MCPH1	Primary microcephaly 1, recessive	AR
MED17	Postnatal Progressive Microcephaly with Seizures and Brain Atrophy	AR
MEFV	Familial Mediterranean fever	AR
MESP2	Spondylocostal dysostosis	AR
MFSD8	Neuronal ceroid lipofuscinosis, MFSD8-related	AR
MKS1	MKS1-related ciliopathies	AR
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts	AR
MLYCD	Malonyl-CoA decarboxylase deficiency	AR
MMAA	Methylmalonic aciduria, cblA type	AR
MMAB	Methylmalonic aciduria, cblB type	AR
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type	AR
MMADHC	Methylmalonic aciduria and homocystinuria, cblD type	AR
MPI	Congenital disorder of glycosylation type Ib	AR

MPL	Congenital amegakaryocytic thrombocytopenia	AR
MPV17	Hepatocerebral mitochondrial DNA depletion syndrome, MPV17-related	AR
MRE11	Ataxia-Telangiectasia-Like Disorder 1	AR
MTHFR	Homocystinuria, MTHFR-related	AR
MTMR2	Charcot-Marie-Tooth disease, type 4B1	AR
MTR	Methylcobalamin deficiency, type cblG	AR
MTRR	Homocystinuria-megaloblastic anemia, cobalamin E type	AR
MTTP	Abetalipoproteinemia	AR
MUT	Methylmalonic aciduria–methylmalonyl–CoA mutase deficiency	AR
MVK	Mevalonate kinase deficiency	AR
MYO15A	Nonsyndromic hearing loss, MYO15A-related	AR
MYO7A	MYO7A-related disorders	AR
NAGA	Schindler disease types 1 and 3	AR
NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo syndrome B)	AR
NAGS	N-acetylglutamate synthase deficiency	AR
NBN	Nijmegen breakage syndrome	AR
NDRG1	Charcot-Marie-Tooth disease, type 4D	AR
NDUFAF2	Mitochondrial complex I deficiency (Leigh syndrome)	AR
NDUFAF5	Mitochondrial complex I deficiency	AR
NDUFS4	Mitochondrial complex I deficiency (Leigh syndrome)	AR
NDUFS6	Mitochondrial complex I deficiency	AR
NDUFS7	Mitochondrial complex I deficiency	AR
NDUFV1	Mitochondrial complex I deficiency, nuclear type 4	AR
NEB	Nemaline myopathy	AR
NEU1	Sialidosis, type I and II	AR
NGLY1	Congenital disorder of deglycosylation	AR
NPC1	Niemann-Pick disease, type C1	AR
NPC2	Niemann-Pick disease, type C2	AR
NPHP1	NPHP1-related ciliopathies	AR
NPHS1	Congenital nephrotic syndrome, type 1	AR
NPHS2	Congenital nephrotic syndrome, type 2	AR
NR2E3	NR2E3-related retinal dystrophies	AR
NTRK1	Congenital insensitivity to pain with anhidrosis	AR
OAT	Gyrate atrophy of choroid and retina	AR
OCA2	Oculocutaneous albinism type II	AR
OPA3	Costeff syndrome	AR
OTOF	Nonsyndromic hearing loss, OTOF- related	AR
P3H1	Osteogenesis imperfecta, type VIII	AR
PAH	Phenylketonuria (PKU)	AR
PANK2	Pantothenate kinase-associated neurodegeneration	AR
PC	Pyruvate carboxylase deficiency	AR
PCBD1	Tetrahydrobiopterin deficiency	AR
PCCA	Propionic acidemia, PCCA-related	AR
PCCB	Propionic acidemia, PCCB-related	AR
PCDH15	PCDH15-related sensory loss	AR
PDE6A	Retinitis pigmentosa, PDE6A-related	AR
PDHB	Pyruvate dehydrogenase E1-beta deficiency	AR

PEPD	Prolidase deficiency	AR
PET100	Mitochondrial complex IV deficiency	AR
PEX1	Zellweger syndrome, PEX1-related	AR
PEX10	Zellweger syndrome, PEX10-related	AR
PEX12	Zellweger syndrome, PEX12-related	AR
PEX2	Zellweger syndrome, PEX2-related	AR
PEX26	Zellweger syndrome	AR
PEX6	Zellweger syndrome, PEX6-related	AR
PEX7	Rhizomelic chondrodysplasia punctata, type 1	AR
PFKM	Glycogen storage disease VII	AR
PHGDH	Phosphoglycerate dehydrogenase deficiency	AR
PHKB	Glycogen storage disease type IXb	AR
PIGN	Multiple congenital anomalies hypotonia seizures syndrome 1	AR
PJVK	Nonsyndromic hearing loss 59	AR
PKHD1	Polycystic kidney disease, PKHD1-related	AR
PLA2G6	Infantile neuroaxonal dystrophy	AR
PLOD1	Ehlers-Danlos syndrome with kyphoscoliosis, PLOD1-related	AR
PMM2	Congenital disorder of glycosylation type 1a	AR
PNPO	Pyridoxamine 5’-phosphate oxidase deficiency	AR
POLG	POLG-related disorders	AR
POLH	Xeroderma pigmentosum	AR
POLR1C	POLR1C-related disorders	AR
POMGNT1	POMGNT1 Alpha-dystroglycanopathies	AR
POMT1	POMT1 Alpha-dystroglycanopathies	AR
POMT2	POMT2 Alpha-dystroglycanopathies	AR
POR	Antley-Bixler syndrome	AR
POU1F1	Combined pituitary hormone deficiency	AR
PPT1	Neuronal ceroid lipofuscinosis, PPT1-related	AR
PRCD	Retinitis pigmentosa 36	AR
PRF1	Hemophagocytic lymphohistiocytosis, familial, 2	AR
PROP1	Combined pituitary hormone deficiency 2	AR
PSAP	Metachromatic leukodystrophy due to saposin-B deficiency	AR
PTPRC	PTPRC related-severe combined immunodeficiency	AR
PTS	Tetrahydrobiopterin deficiency	AR
PUS1	Mitochondrial myopathy and sideroblastic anemia 1	AR
PYGM	Glycogen storage disease type V	AR
QDPR	Tetrahydrobiopterin deficiency, QDPR-related	AR
RAB23	Carpenter syndrome	AR
RAG1	Omenn syndrome, RAG1-related	AR
RAG2	Omenn syndrome, RAG2-related	AR
RAPSN	RAPSN-associated acetylcholine receptor deficiency	AR
RARS2	Pontocerebellar hypoplasia type 6	AR
RAX	Microphthalmia, isolated 3	AR
RDH12	Leber congenital amaurosis type 13	AR
RLBP1	Retinal dystrophy, RLBP1-related	AR
RMRP	Cartilage-Hair Hypoplasia Anauxetic Dysplasia Spectrum Disorder	AR
RNASEH2B	Aicardi Goutieres syndrome 2	AR

RNASEH2C	Aicardi-Goutieres syndrome 3	AR
RPE65	RPE65-related retinopathy	AR
RPGRIP1L	RPGRIP1L-related ciliopathies	AR
RTEL1	Dyskeratosis congenita type 5	AR
SACS	Autosomal recessive spastic ataxia of Charlevoix-Saguenay	AR
SAMD9	Normophosphatemic Familial Tumoral Calcinosis	AR
SAMHD1	Aicardi-Goutieres syndrome	AR
SCO2	Mitochondrial complex IV deficiency	AR
SEC23B	Congenital dyserythropoietic anemia, type II	AR
SEPSECS	Pontocerebellar hypoplasia type 2D	AR
SERPINA1	Alpha-1 antitrypsin deficiency	AR
SGCA	Limb-girdle muscular dystrophy, type 2D	AR
SGCB	Limb-girdle muscular dystrophy, type 2E	AR
SGCD	Limb-girdle muscular dystrophy, type 2F	AR
SGCG	Limb-girdle muscular dystrophy, type 2C	AR
SGSH	Mucopolysaccharidosis IIIA (Sanfilippo syndrome A)	AR
SH3TC2	Charcot-Marie-Tooth disease, SH3TC2-related	AR
SLC12A3	Gitelman syndrome	AR
SLC12A6	Andermann syndrome	AR
SLC17A5	Sialic acid storage disorder	AR
SLC19A2	Thiamine-responsive megaloblastic anemia syndrome	AR
SLC19A3	Biotin-responsive basal ganglia disease	AR
SLC1A4	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly syndrome	AR
SLC22A5	Systemic primary carnitine deficiency	AR
SLC25A13	Citrin deficiency	AR
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome (Triple H syndrome)	AR
SLC25A20	Carnitine-acylcarnitine translocase deficiency	AR
SLC26A2	SLC26A2-related disorders	AR
SLC26A3	Congenital secretory chloride diarrhea	AR
SLC26A4	Pendred syndrome	AR
SLC35A3	Arthrogryposis, intellectual disability, and seizures	AR
SLC37A4	Glycogen storage disease, type Ib	AR
SLC39A4	Acrodermatitis enteropathica	AR
SLC3A1	Cystinuria, type I	AR
SLC45A2	Oculocutaneous albinism, type IV	AR
SLC46A1	Hereditary folate malabsorption	AR
SLC4A11	Corneal endothelial dystrophy	AR
SLC5A5	Thyroid dyshormonogenesis, SLC5A5-related	AR
SLC6A19	Hartnup disorder	AR
SLC7A7	Lysinuric protein intolerance	AR
SMARCAL1	Schimke immunoosseous dysplasia	AR
SMN1	Spinal muscular atrophy	AR
SMPD1	Niemann-Pick disease, type A/B	AR
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome	AR
SPG11	SPG11-related Neuromuscular Disorders	AR
SPG7	Spastic paraplegia type 7	AR
SPINK5	Netherton syndrome	AR

SPR	Sepiapterin Reductase Deficiency	AR
ST3GAL5	Salt and pepper developmental regression syndrome	AR
STAR	Lipoid congenital adrenal hyperplasia	AR
STX11	Familial hemophagocytic lymphohistiocytosis	AR
STXBP2	Familial hemophagocytic lymphohistiocytosis	AR
SUMF1	Multiple sulfatase deficiency	AR
SURF1	Leigh syndrome, SURF1-related	AR
SYNE4	Autosomal recessive deafness 76	AR
TAT	Tyrosinemia, type II	AR
TBCE	Hypoparathyroidism-retardation-dysmorphism syndrome	AR
TCIRG1	Osteopetrosis 1	AR
TCTN2	TCTN2-related ciliopathies	AR
TECPR2	Spastic paraplegia 49	AR
TF	Atransferrinemia	AR
TFR2	Hemochromatosis, type 3	AR
TG	Thyroid dyshormonogenesis, TG- related	AR
TGM1	Congenital ichthyosis	AR
TH	Segawa syndrome	AR
TK2	Mitochondrial DNA depletion syndrome 2	AR
TMC1	Nonsyndromic hearing loss 7	AR
TMEM216	TMEM216-related ciliopathies	AR
TMPRSS3	Nonsyndromic hearing loss, TMPRSS3-related	AR
TPO	Thyroid dyshormonogenesis, TPO- related	AR
TPP1	Neuronal ceroid lipofuscinosis, TPP1- related	AR
TRDN	Catecholaminergic polymorphic ventricular tachycardia	AR
TREX1	Aicardi-Goutieres syndrome 1	AR
TRIM32	TRIM32-related disorders	AR
TRIM37	Mulibrey nanism	AR
TRMU	Liver failure, acute infantile	AR
TSEN54	Pontocerebellar hypoplasia type 2A	AR
TSFM	Combined oxidative phosphorylation deficiency, TSFM-related	AR
TSHB	Congenital hypothyroidism, TSHB- related	AR
TSHR	Congenital hypothyroidism, TSHR- related	AR
TTC37	Trichohepatoenteric syndrome	AR
TTPA	Ataxia with isolated vitamin E deficiency	AR
TULP1	TULP1-related retinal disorders	AR
TYMP	Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease	AR
TYR	Oculocutaneous albinism types 1A and 1B	AR
TYRP1	Oculocutaneous albinism, type III	AR
UGT1A1	Crigler-Najjar syndrome	AR
UNC13D	Familial hemophagocytic lymphohistiocytosis type 3	AR
USH1C	USH1C-related disorders	AR
USH1G	Usher syndrome type IG	AR
USH2A	Usher syndrome, type 2A	AR
VDR	Vitamin D-dependent rickets, type 2A	AR
VPS13A	Choreoacanthocytosis	AR
VPS13B	Cohen syndrome	AR

VPS45	Severe congenital neutropenia, VPS45-related	AR
VPS53	Pontocerebellar hypoplasia type 2E	AR
VRK1	Pontocerebellar hypoplasia type 1A	AR
VSX2	Microphthalmia with or without coloboma	AR
WHRN	Usher syndrome type 2D	AR
WISP3	Progressive pseudorheumatoid dysplasia	AR
WNT10A	WNT10A-related ectodermal dysplasias	AR
WRN	Werner syndrome	AR
XPA	Xeroderma pigmentosum, group A	AR
XPC	Xeroderma pigmentosum, group C	AR
ZFYVE26	Spastic paraplegia 15	AR
GJB6	GJB6-CRYL1 related nonsyndromic hearing loss	AR, Digenic
	TOTAL Autosomal recessive genes	**477**
ABCD1	Adrenoleukodystrophy, X-linked	XL
AP1S2	X-linked Intellectual disability, AP1S2-related	XL
ARSE	Chondrodysplasia punctata type 1, X-linked	XL
ARX	X-linked intellectual disability, ARX-related	XL
ATP7A	Menkes disease	XL
ATRX	Alpha thalassemia X-linked intellectual disability syndrome	XL
BRWD3	X-linked intellectual disability, BRWD3-related	XL
BTK	X-linked agammaglobulinemia	XL
CD40LG	Hyper IgM syndrome, X-linked	XL
CHM	Choroideremia	XL
COL4A5	Alport syndrome, COL4A5-related	XL
CUL4B	X-linked intellectual disability, CUL4B-related	XL
CYBB	Chronic granulomatous disease, X-linked	XL
DCX	Lissencephaly, X-linked	XL
DKC1	X-linked dyskeratosis congenita	XL
DLG3	X-linked intellectual disability, DLG3-related	XL
DMD	Dystrophinopathies	XL
EDA	Hypohidrotic ectodermal dysplasia	XL
EMD	Emery-Dreifuss muscular dystrophy	XL
F8	Hemophilia A	XL
F9	Hemophilia B	XL
FGD1	X-linked Aarskog-Scott syndrome	XL
FMR1	Fragile X Syndrome	XL
FTSJ1	X-linked intellectual disability, FTSJ1-related	XL
G6PD	Glucose-6-phosphate dehydrogenase deficiency	XL
GJB1	Charcot-Marie-Tooth disease, X-linked type 1	XL
GLA	Fabry disease	XL
IDS	Mucopolysaccharidosis type II (Hunter syndrome)	XL
IL1RAPL1	X-linked intellectual disability, IL1RAPL1-related	XL
IL2RG	X-linked severe combined immunodeficiency	XL
L1CAM	L1 syndrome	XL
MID1	Opitz GBBB syndrome, type I	XL

MTM1	Myotubular myopathy, X-linked	XL
NR0B1	Congenital adrenal hypoplasia, X-linked	XL
OCRL	OCRL-related disorders	XL
OPHN1	X-linked intellectual disability- cerebellar hypoplasia syndrome	XL
OTC	Ornithine transcarbamylase deficiency	XL
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency	XL
PGK1	Phosphoglycerate kinase 1 deficiency	XL
PLP1	PLP1-related disorders	XL
POU3F4	X-linked hearing loss, POU3F4-related	XL
PQBP1	Renpenning syndrome	XL
PRPS1	PRPS1-related disorders	XL
RP2	X-linked Retinitis pigmentosa, RP2-related	XL
RPGR	X-linked Retinitis pigmentosa, RPGR-related	XL
RS1	Juvenile retinoschisis, X-linked	XL
SLC16A2	Allan-Herndon-Dudley syndrome	XL
SLC6A8	Creatine deficiency syndrome	XL
SYN1	X-linked epilepsy with variable learning disabilities	XL
TAZ	Barth syndrome	XL
THOC2	X-linked Intellectual disability, THOC2-related	XL
UPF3B	Lujan-Fryns syndrome, UPF3B- related	XL
WAS	WAS-related hematopoietic disorder	XL
ZDHHC9	X-linked intellectual disability, ZDHHC9-related	XL
AFF2	Fragile XE syndrome	XL
AR	Androgen insensitivity syndrome	XL
AVPR2	Nephrogenic diabetes insipidus	XL
CLCN5	Dent disease	XL
FANCB	Fanconi anemia group B	XL
FHL1	FHL1-related neuromuscular disorders	XL
FOXP3	IPEX syndrome	XL
GPR143	X-linked Ocular albinism, GPR143-related	XL
HCFC1	Methylmalonic acidemia with homocystinuria, type cblX	XL
HSD17B10	HSD10 mitochondrial disease	XL
IGSF1	X-linked central hypothyroidism and testicular enlargement	XL
KDM5C	X-linked intellectual disability, KDM5C-related	XL
NDP	Norrie disease	XL
NONO	X-linked intellectual disability syndrome 34	XL
PAK3	X-linked intellectual disability, PAK3- related	XL
PHF8	X-linked intellectual disability, Siderius type	XL
PHKA1	Glycogen storage disease type IXd	XL
PHKA2	Glycogen storage disease type IXa	XL
ZNF711	X-linked intellectual disability, ZNF711-related	XL
	TOTAL X-linked genes	73
	TOTAL GENES	**550**