Monash IVF

Expanded Carrier Screen

Gene list (by gene)

Please Note: If there is a specific genetic condition that you are wanting to be screened for, this general screening test may not be the best option for you. This list of genes is our standard screening panel and there is the option of customising the genes screened if clinically indicated.

Please contact **carrierscreening@monashivf.com** or **1800 684 198** if the above applies to you, or if you have any additional questions.

Gene	Disorder	Inheritance	Carrier Frequency*
CFTR	Cystic Fibrosis	AR	1 in 32
FMR1	Fragile X Syndrome Intermediate Allele/Premutation/Full Mutation	XL	1 in 259
SMN1	Spinal muscular atrophy	AR	1 in 54
HBA1/HBA2	Alpha thalassemia trait cis (αα/- -)	AR	1 in 1000
	Heterozygous alpha thalassemia carriers (αα/α-)	AR	1 in 18
HBB	Sickle cell disease, Hemoglobin C disease, Beta thalassemia, Hemoglobin E disease	AR	1 in 158
ABCA12	Congenital ichthyosis, ABCA12-related	AR	<1 in 500
ABCA3	Surfactant metabolism dysfunction, pulmonary 3	AR	1 in 116
ABCA4	Stargardt disease	AR	1 in 51
ABCB11	Progressive familial intrahepatic cholestasis	AR	1 in 112
ABCC8	Familial hyperinsulinism	AR	1 in 112
ABCD1	X-linked Adrenoleukodystrophy	XL	1 in 21,000
ACAD9	Acyl-CoA dehydrogenase-9 (ACAD9) deficiency	AR	<1 in 500
ACADM	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	AR	1 in 69
ACADVL	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	AR	1 in 118
ACAT1	3-ketothiolase deficiency	AR	<1 in 500
ACOX1	Peroxisomal acyl-CoA oxidase deficiency	AR	<1 in 500
ACSF3	Combined malonic and methylmalonic aciduria	AR	<1 in 500
ADA	Adenosine deaminase deficiency	AR	1 in 224
ADAMTS2	Ehlers-Danlos syndrome, dermatosparaxis type	AR	<1 in 500
ADGRG1	Bilateral frontoparietal polymicrogyria	AR	<1 in 500
ADK	Hypermethioninemia due to adenosine kinase deficiency	AR	<1 in 500
AGA	Aspartylglucosaminuria	AR	<1 in 500
AGL	Glycogen storage disease type III	AR	1 in 158
AGPS	Rhizomelic chondrodysplasia punctata, type 3	AR	<1 in 500
AGXT	Primary hyperoxaluria type 1	AR	1 in 120
AHI1	Joubert syndrome, AHI1-related	AR	1 in 448
AIPL1	Childhood-onset severe retinal dystrophy, AIPL1-related	AR	1 in 409
ALDH3A2	Sjögren-Larsson syndrome	AR	1 in 250
ALDOB	Hereditary fructose intolerance	AR	1 in 122
ALG6	Congenital disorder of glycosylation type Ic	AR	<1 in 500
ALMS1	Alstrom syndrome	AR	1 in 500
ALPL	Hypophosphatasia	AR	1 in 158
AMT	Glycine encephalopathy	AR	1 in 373
AP1S2	X-linked Intellectual disability, AP1S2-related	XL	<1 in 50,000
AQP2	Nephrogenic diabetes insipidus	AR	<1 in 500
ARG1	Arginase deficiency	AR	1 in 296
ARL13B	Joubert syndrome, ARL13B-related	AR	<1 in 500
ARSA	Metachromatic leukodystrophy	AR	1 in 100

*Carrier Frequency for the general population
Abbreviations: AR, autosomal recessive; XL, X-linked

ARSB	Mucopolysaccharidosis type VI (Maroteaux- Lamy syndrome)	AR	1 in 250
ARSE	Chondrodysplasia punctata type 1, X-linked	XL	1 in 250,000
ARX	X-linked intellectual disability, ARX-related	XL	<1 in 50,000
ASL	Argininosuccinate lyase deficiency	AR	1 in 132
ASNS	Asparagine synthetase deficiency	AR	<1 in 500
ASPA	Canavan disease	AR	1 in 300
ASS1	Citrullinemia	AR	1 in 119
ATM	Ataxia-telangiectasia	AR	1 in 100
ATP6V1B1	Renal tubular acidosis with deafness	AR	<1 in 500
ATP7A	Menkes disease	XL	1 in 50,000
ATP7B	Wilson disease	AR	1 in 87
ATRX	Alpha thalassemia X-linked intellectual disability syndrome	XL	<1 in 250,000
BBS1	Bardet-Biedl syndrome type 1	AR	1 in 367
BBS10	Bardet-Biedl syndrome type 10	AR	1 in 395
BBS12	Bardet-Biedl syndrome type 12	AR	1 in 791
BBS2	BBS2-related ciliopathies	AR	1 in 621
BCKDHA	Maple syrup urine disease type Ia	AR	1 in 321
BCKDHB	Maple syrup urine disease type Ib	AR	1 in 364
BCS1L	Mitochondrial complex III deficiency	AR	<1 in 500
BLM	Bloom syndrome	AR	1 in 800
BRWD3	X-linked intellectual disability, BRWD3-related	XL	<1 in 50,000
BSND	Bartter syndrome type 4a	AR	<1 in 500
CAPN3	Limb-girdle muscular dystrophy type 2A	AR	<1 in 500
CASQ2	Catecholaminergic polymorphic ventricular tachycardia	AR	1 in 224
CBS	Homocystinuria due to cystathionine beta-synthase deficiency	AR	1 in 224
CC2D2A	Joubert syndrome 9	AR	1 in 201
CCDC103	Primary ciliary dyskinesia, type 17	AR	1 in 316
CCDC39	Primary ciliary dyskinesia, type 14	AR	1 in 211
CCDC88C	Congenital hydrocephalus 1	AR	1 in 137
CD40LG	Hyper IgM syndrome, X-linked	XL	1 in 50,000
CDH23	Usher syndrome, type 1D	AR	1 in 285
CEP290	CEP290-related Ciliopathies	AR	1 in 190
CHM	Choroideremia	XL	1 in 25,000
CHRNE	Congenital myasthenic syndrome	AR	1 in 408
CHRNG	Multiple pterygium syndrome	AR	<1 in 500
CHST6	Macular corneal dystrophy, CHST6-related	AR	1 in 79
CIITA	Bare lymphocyte syndrome, type II	AR	<1 in 500
CLN3	Neuronal ceroid lipofuscinosis	AR	1 in 230
CLN5	Neuronal ceroid lipofuscinosis 5	AR	<1 in 500
CLN6	Neuronal ceroid lipofuscinosis, CLN6-related	AR	<1 in 500
CLN8	Neuronal ceroid lipofuscinosis, CLN8-related	AR	<1 in 500
CLRN1	Usher syndrome, type 3A	AR	1 in 500
CNGB3	Achromatopsia	AR	1 in 87
COL27A1	Steel syndrome	AR	<1 in 500
COL4A3	Alport syndrome, COL4A3-related	AR	1 in 267
COL4A4	Alport syndrome, COL4A4-related	AR	1 in 267
COL4A5	Alport syndrome, COL4A5-related	XL	1 in 139

*Carrier Frequency for the general population
Abbreviations: AR, autosomal recessive; XL, X-linked

COL7A1	Dystrophic epidermolysis bullosa	AR	1 in 196
COX15	Mitochondrial complex IV deficiency	AR	<1 in 500
CPS1	Carbamoylphosphate synthetase I deficiency	AR	1 in 570
CPT1A	Carnitine palmitoyltransferase IA deficiency	AR	1 in 354
CPT2	Carnitine palmitoyltransferase II deficiency	AR	<1 in 500
CRB1	CRB1-related retinopathy	AR	1 in 104
CRYL1	GJB6-CRYL1 related nonsyndromic hearing loss	UK	1 in 423
CTNS	Cystinosis	AR	1 in 158
CTSA	Galactosialidosis	AR	<1 in 500
CTSC	Papillon-Lefevre syndrome	AR	<1 in 500
CTSD	Neuronal ceroid lipofuscinosis, CTSD-related	AR	<1 in 500
CTSK	Pycnodysostosis	AR	<1 in 500
CUL4B	X-linked intellectual disability, CUL4B-related	XL	<1 in 50,000
CYBA	Chronic granulomatous disease	AR	1 in 224
CYBB	Chronic granulomatous disease, X-linked	XL	1 in 149,254
CYP11A1	Congenital adrenal insufficiency	AR	1 in 114
CYP11B1	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	AR	1 in 158
CYP11B2	Corticosterone methyloxidase deficiency	AR	<1 in 500
CYP17A1	Congenital adrenal hyperplasia due to 17- alpha-hydroxylase deficiency	AR	1 in 500
CYP1B1	Primary congenital glaucoma	AR	1 in 50
CYP21A2	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	AR	1 in 61
CYP27A1	Cerebrotendinous xanthomatosis	AR	1 in 500
DBT	Maple syrup urine disease, type II	AR	1 in 481
DCLRE1C	Severe combined immunodeficiency with sensitivity to ionizing radiation	AR	<1 in 500
DCX	Lissencephaly, X-linked	XL	1 in 42,500
DDX11	Warsaw breakage syndrome	AR	<1 in 500
DHCR7	Smith-Lemli-Opitz syndrome	AR	1 in 30
DHDDS	Retinitis pigmentosa 59	AR	1 in 296
DLD	Dihydrolipoamide dehydrogenase deficiency	AR	1 in 500
DLG3	X-linked intellectual disability, DLG3-related	XL	<1 in 50,000
DMD	Dystrophinopathies	XL	1 in 2,350
DNAH5	Primary ciliary dyskinesia, DNAH5-related	AR	1 in 142
DNAI1	Primary ciliary dyskinesia, DNAI1-related	AR	1 in 230
DNAI2	Primary ciliary dyskinesia, DNAI2-related	AR	1 in 447
DUOX2	Congenital hypothyroidism, DUOX2-related	AR	1 in 56
DUOXA2	Congenital hypothyroidism, DUOXA2-related	AR	<1 in 500
DYNC2H1	Short-rib thoracic dysplasia 3 with or without polydactyly	AR	1 in 68
DYSF	Limb-girdle muscular dystrophy type 2B	AR	<1 in 500
EDA	Hypohidrotic ectodermal dysplasia	XL	1 in 14,167
EIF2AK3	Wolcott-Rallison Syndrome	AR	<1 in 500
EIF2B5	Leukoencephalopathy with vanishing white matter	AR	<1 in 500
ELP1	Familial Dysautonomia	AR	1 in 300
EMD	Emery-Dreifuss muscular dystrophy	XL	1 in 81,967
ERCC2	ERCC2-related disorders	AR	1 in 65
ERCC5	Xeroderma Pigmentosa, group G	AR	<1 in 500
ERCC6	ERCC6-related disorders	AR	1 in 500

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ERCC8	Cockayne syndrome type A	AR	1 in 822
ESCO2	Roberts syndrome	AR	<1 in 500
ETFA	Glutaric aciduria IIA	AR	1 in 500
ETFB	Glutaric aciduria IIB	AR	1 in 500
ETFDH	Glutaric aciduria IIC	AR	1 in 250
ETHE1	Ethylmalonic encephalopathy	AR	<1 in 500
EVC	EVC-related bone growth disorders	AR	1 in 142
EVC2	EVC2-related bone growth disorders	AR	1 in 240
EXOSC3	Pontocerebellar hypoplasia type 1B	AR	<1 in 500
F2	Prothrombin-related conditions	AR	1 in 33
F5	Factor V deficiency	AR	1 in 36
F8	Hemophilia A	XL	1 in 3,250
F9	Hemophilia B	XL	1 in 15,000
FAH	Tyrosinemia, type 1	AR	1 in 99
FAM126A	Hypomyelinating leukodystropy type 5	AR	<1 in 500
FAM161A	Retinitis pigmentosa 28	AR	1 in 296
FANCA	Fanconi anemia group A	AR	1 in 239
FANCC	Fanconi anemia group C	AR	1 in 535
FANCG	Fanconi anemia group G	AR	1 in 632
FH	Fumarase deficiency	AR	<1 in 500
FKRP	FKRP Alpha-dystroglycanopathies	AR	1 in 158
FKTN	FKTN Alpha-dystroglycanopathies	AR	1 in 500
FOXRED1	Mitochondrial complex I deficiency	AR	<1 in 500
FTCD	Glutamate formiminotransferase deficiency	AR	<1 in 500
FTSJ1	X-linked intellectual disability, FTSJ1-related	XL	<1 in 50,000
FUCA1	Fucosidosis	AR	<1 in 500
G6PC	Glycogen storage disease, type 1a	AR	1 in 177
GAA	Pompe disease	AR	1 in 100
GALC	Krabbe disease	AR	1 in 158
GALNS	Mucopolysaccharidosis IVA (Morquio syndrome A)	AR	1 in 224
GALT	Galactosemia	AR	1 in 110
GAMT	Guanidinoacetate methyltransferase deficiency	AR	1 in 371
GBA	Gaucher disease	AR	1 in 77
GBE1	Glycogen storage disease IV	AR	1 in 387
GCDH	Glutaric aciduria, type I	AR	1 in 87
GDAP1	Charcot-Marie-Tooth disease, GDAP1-related	AR	1 in 152
GDF5	Du Pan Syndrome	AR	<1 in 500
GFM1	Combined oxidative phosphorylation deficiency, GFM1-related	AR	<1 in 500
GJB2	Nonsyndromic hearing loss 1A	AR	1 in 42
GJB6	GJB6-CRYL1 related nonsyndromic hearing loss	AR	1 in 423
GLA	Fabry disease	XL	1 in 25,000
GLB1	GLB1-related disorders	AR	1 in 134
GLDC	Glycine encephalopathy, GLDC-related	AR	1 in 193
GLE1	Lethal congenital contracture syndrome 1	AR	<1 in 500
GNE	Inclusion body myopathy type 2 (Nonaka myopathy)	AR	<1 in 500
GNPTAB	Mucolipidosis II & III	AR	<1 in 500
GNPTG	Mucolipidosis III gamma	AR	<1 in 500

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GNS	Mucopolysaccharidosis IIID (Sanfilippo syndrome D)	AR	1 in 500
GPR143	X-linked Ocular albinism, GPR143-related	XL	1 in 25,000
GSS	Glutathione synthetase deficiency	AR	<1 in 500
GUCY2D	Leber congenital amaurosis 1	AR	<1 in 500
GUSB	Mucopolysaccharidosis type VII	AR	1 in 250
HADHA	Trifunctional protein deficiency	AR	<1 in 500
HADHB	Trifunctional protein deficiency	AR	<1 in 500
HAX1	Severe congenital neutropenia, HAX1-related	AR	1 in 224
HEXA	Tay-Sachs disease	AR	1 in 300
HEXB	Sandhoff disease	AR	1 in 600
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo syndrome C)	AR	1 in 434
HJV	Hemochromatosis, type 2A	AR	1 in 500
HLCS	Holocarboxylase synthetase deficiency	AR	1 in 500
HMGCL	3-hydroxy-3-methylglutaryl-CoA lyase deficiency	AR	<1 in 500
HOGA1	Primary hyperoxaluria type III	AR	1 in 184
HPS1	Hermansky-Pudlak syndrome 1	AR	1 in 354
HPS3	Hermansky-Pudlak syndrome 3	AR	1 in 354
HPS4	Hermansky-Pudlak syndrome 4	AR	<1 in 500
HSD17B4	D-bifunctional protein deficiency	AR	1 in 158
HSD3B2	Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	AR	<1 in 500
HYLS1	Hydrolethalus syndrome	AR	<1 in 500
IDS	Mucopolysaccharidosis type II (Hunter syndrome)	XL	1 in 50,000
IDUA	Mucopolysaccharidosis, type I (Hurler syndrome)	AR	<1 in 500
IL1RAPL1	X-linked intellectual disability, IL1RAPL1- related	XL	<1 in 50,000
IL2RG	X-linked severe combined immunodeficiency	XL	1 in 25,000
IVD	Isovaleric Acidemia	AR	1 in 167
IYD	Thyroid dyshormonogenesis, IYD-related	AR	<1 in 500
JAK3	Severe combined immunodeficiency, JAK3- related	AR	1 in 299
KCNJ11	KCNJ11-related hyperinsulinism	AR	1 in 423
KDM5C	X-linked intellectual disability, KDM5C-related	XL	<1 in 50,000
L1CAM	L1 syndrome	XL	1 in 15,000
LAMA2	Muscular dystrophy, LAMA2-related	AR	<1 in 500
LAMA3	Junctional epidermolysis bullosa 2	AR	1 in 781
LAMB3	Junctional epidermolysis bullosa, LAMB3- related	AR	1 in 781
LAMC2	Junctional epidermolysis bullosa, LAMC2- related	AR	1 in 781
LCA5	Leber congenital amaurosis 5	AR	1 in 500
LDLRAP1	Familial Hypercholesterolemia	AR	1 in 8
LHX3	Combined pituitary hormone deficiency 3	AR	1 in 45
LIFR	Stuve-Wiedemann syndrome	AR	<1 in 500
LIPA	Lysosomal acid lipase deficiency	AR	1 in 211
LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type	AR	<1 in 500
LOXHD1	Nonsyndromic hearing loss 77	AR	1 in 500
LPL	Familial lipoprotein lipase deficiency	AR	1 in 500
LRP2	Donnai–Barrow syndrome	AR	1 in 214
LRPPRC	Leigh syndrome with Complex IV deficiency	AR	1 in 447
LYST	Chediak-Higashi syndrome	AR	<1 in 500
MAN2B1	Alpha-Mannosidosis	AR	1 in 354

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Abbreviations: AR, autosomal recessive; XL, X-linked

MANBA	Beta-Mannosidosis	AR	<1 in 500
MCOLN1	Mucolipidosis IV	AR	1 in 300
MCPH1	Primary microcephaly 1, recessive	AR	1 in 147
MED17	Postnatal Progressive Microcephaly with Seizures and Brain Atrophy	AR	<1 in 500
MESP2	Spondylocostal dysostosis	AR	<1 in 500
MFSD8	Neuronal ceroid lipofuscinosis, MFSD8-related	AR	<1 in 500
MID1	Opitz GBBB syndrome, type I	XL	<1 in 50,000
MKS1	MKS1-related ciliopathies	AR	1 in 260
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts	AR	<1 in 500
MLYCD	Malonyl-CoA decarboxylase deficiency	AR	<1 in 500
MMAA	Methylmalonic aciduria, cblA type	AR	1 in 301
MMAB	Methylmalonic aciduria, cblB type	AR	1 in 435
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type	AR	1 in 134
MMADHC	Methylmalonic aciduria and homocystinuria, cblD type	AR	<1 in 500
MPI	Congenital disorder of glycosylation type Ib	AR	<1 in 500
MPL	Congenital amegakaryocytic thrombocytopenia	AR	1 in 102
MPV17	Hepatocerebral mitochondrial DNA depletion syndrome, MPV17-related	AR	<1 in 500
MTHFR	Homocystinuria, MTHFR-related	AR	1 in 224
MTM1	Myotubular myopathy, X-linked	XL	1 in 25,000
MTMR2	Charcot-Marie-Tooth disease, type 4B1	AR	<1 in 500
MTRR	Homocystinuria-megaloblastic anemia, cobalamin E type	AR	<1 in 500
MTTP	Abetalipoproteinemia	AR	<1 in 500
MUT	Methylmalonic aciduria–methylmalonyl–CoA mutase deficiency	AR	1 in 100
MVK	Mevalonate kinase deficiency	AR	<1 in 500
MYO7A	MYO7A-related disorders	AR	1 in 206
NAGA	Schindler disease types 1 and 3	AR	1 in 94
NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo syndrome B)	AR	<1 in 500
NAGS	N-acetylglutamate synthase deficiency	AR	<1 in 500
NBN	Nijmegen breakage syndrome	AR	1 in 158
NDP	Norrie disease	XL	<1 in 50,000
NDRG1	Charcot-Marie-Tooth disease, type 4D	AR	1 in 22
NDUFAF2	Mitochondrial complex I deficiency	AR	<1 in 500
NDUFAF5	Mitochondrial complex I deficiency (Leigh syndrome)	AR	1 in 447
NDUFS4	Mitochondrial complex I deficiency	AR	<1 in 500
NDUFS6	Mitochondrial complex I deficiency (Leigh syndrome)	AR	<1 in 500
NDUFS7	Mitochondrial complex I deficiency	AR	<1 in 500
NDUFV1	Mitochondrial complex I deficiency, nuclear type 4	AR	<1 in 500
NEB	Nemaline myopathy	AR	1 in 112
NEU1	Sialidosis, type I and II	AR	<1 in 500
NPC1	Niemann-Pick disease, type C1	AR	1 in 194
NPC2	Niemann-Pick disease, type C2	AR	1 in 194
NPHP1	NPHP1-related ciliopathies	AR	1 in 480
NPHS1	Congenital nephrotic syndrome, type 1	AR	1 in 289
NPHS2	Congenital nephrotic syndrome, type 2	AR	1 in 289
NR0B1	Congenital adrenal hypoplasia, X-linked	XL	1 in 6,250
NTRK1	Congenital insensitivity to pain with anhidrosis	AR	<1 in 500

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Abbreviations: AR, autosomal recessive; XL, X-linked

OAT	Gyrate atrophy of choroid and retina	AR	<1 in 500
OCA2	Oculocutaneous albinism type II	AR	1 in 76
OCRL	OCRL-related disorders	XL	1 in 250,000
OPA3	Costeff syndrome	AR	<1 in 500
OPHN1	X-linked intellectual disability-cerebellar hypoplasia syndrome	XL	<1 in 50,000
OTC	Ornithine transcarbamylase deficiency	XL	1 in 7,000
OTOF	Nonsyndromic hearing loss, OTOF-related	AR	<1 in 500
P3H1	Osteogenesis imperfecta, type VIII	AR	<1 in 500
PAH	Phenylalanine Hydroxylase deficiency (Phenylketonuria)	AR	1 in 93
PAK3	X-linked intellectual disability, PAK3-related	XL	<1 in 50,000
PANK2	Pantothenate kinase-associated neurodegeneration	AR	1 in 289
PC	Pyruvate carboxylase deficiency	AR	1 in 250
PCCA	Propionic acidemia, PCCA-related	AR	1 in 224
PCCB	Propionic acidemia, PCCB-related	AR	1 in 224
PCDH15	PCDH15-related sensory loss	AR	1 in 395
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II	AR	<1 in 500
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency	XL	<1 in 250,000
PDHB	Pyruvate dehydrogenase E1-beta deficiency	AR	<1 in 500
PEX1	Zellweger syndrome, PEX1-related	AR	1 in 147
PEX10	Zellweger syndrome, PEX10-related	AR	1 in 500
PEX12	Zellweger syndrome, PEX12-related	AR	1 in 373
PEX2	Zellweger syndrome, PEX2-related	AR	1 in 500
PEX26	Zellweger syndrome	AR	<1 in 500
PEX6	Zellweger syndrome, PEX6-related	AR	1 in 280
PEX7	Rhizomelic chondrodysplasia punctata, type 1	AR	1 in 158
PFKM	Glycogen storage disease VII	AR	<1 in 500
PGK1	Phosphoglycerate kinase 1 deficiency	XL	<1 in 50,000
PHF8	X-linked intellectual disability, Siderius type	XL	<1 in 50,000
PHGDH	Phosphoglycerate dehydrogenase deficiency	AR	<1 in 500
PHYH	Refsum disease	AR	<1 in 500
PKHD1	Polycystic kidney disease, PKHD1-related	AR	1 in 70
PLA2G6	Infantile neuroaxonal dystrophy	AR	1 in 500
PLOD1	Ehlers-Danlos syndrome with kyphoscoliosis, PLOD1-related	AR	1 in 159
PLP1	PLP1-related disorders	XL	<1 in 50,000
PMM2	PMM2-glycosylation disorders	AR	1 in 63
POLG	POLG-related disorders	AR	1 in 113
POLR1C	POLR1C-related disorders	AR	<1 in 500
POMGNT1	POMGNT1 Alpha-dystroglycanopathies	AR	1 in 462
POMT1	POMT1 Alpha-dystroglycanopathies	AR	1 in 290
POMT2	POMT2 Alpha-dystroglycanopathies	AR	1 in 371
POR	Antley-Bixler syndrome	AR	1 in 159
POU3F4	X-linked hearing loss, POU3F4-related	XL	<1 in 50,000
PPT1	Neuronal ceroid lipofuscinosis, PPT1-related	AR	1 in 368
PQBP1	Renpenning syndrome	XL	<1 in 500
PRF1	Hemophagocytic lymphohistiocytosis, familial, 2	AR	1 in 149
PROP1	Combined pituitary hormone deficiency 2	AR	1 in 45
PRPS1	PRPS1-related disorders	XL	<1 in 250,000

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Abbreviations: AR, autosomal recessive; XL, X-linked

PSAP	Metachromatic leukodystrophy due to saposin-b deficiency	AR	<1 in 500
PTS	Tetrahydrobiopterin deficiency	AR	1 in 354
PUS1	Mitochondrial myopathy and sideroblastic anemia 1	AR	<1 in 500
QDPR	Tetrahydrobiopterin deficiency, QDPR-related	AR	<1 in 500
RAB23	Carpenter syndrome	AR	<1 in 500
RAG1	Omenn syndrome, RAG1-related	AR	1 in 290
RAG2	Omenn syndrome, RAG2-related	AR	1 in 137
RAPSN	RAPSN-associated acetylcholine receptor deficiency	AR	<1 in 500
RARS2	Pontocerebellar hypoplasia type 6	AR	<1 in 500
RAX	Microphthalmia, isolated 3	AR	1 in 289
RDH12	Leber congenital amaurosis type 13	AR	<1 in 500
RMRP	Cartilage-Hair Hypoplasia Anauxetic Dysplasia Spectrum Disorder	AR	<1 in 500
RNASEH2B	Aicardi Goutieres syndrome 2	AR	1 in 217
RP2	X-linked Retinitis pigmentosa, RP2-related	XL	1 in 4,000
RPE65	RPE65-related retinopathy	AR	1 in 228
RPGR	X-linked Retinitis pigmentosa, RPGR-related	XL	1 in 3,000
RPGRIP1L	RPGRIP1L-related ciliopathies	AR	1 in 259
RTEL1	Dyskeratosis congenita type 5	AR	1 in 500
SACS	Autosomal recessive spastic ataxia of Charlevoix-Saguenay	AR	<1 in 500
SAMD9	Normophosphatemic Familial Tumoral Calcinosis	AR	<1 in 500
SAMHD1	Aicardi-Goutieres syndrome	AR	<1 in 500
SCO2	Mitochondrial complex IV deficiency	AR	1 in 150
SEPSECS	Pontocerebellar hypoplasia type 2D	AR	<1 in 500
SERPINA1	Alpha-1 antitrypsin deficiency	AR	1 in 33
SGCA	Limb-girdle muscular dystrophy, type 2D	AR	<1 in 500
SGCB	Limb-girdle muscular dystrophy, type 2E	AR	1 in 500
SGCD	Limb-girdle muscular dystrophy, type 2F	AR	<1 in 500
SGCG	Limb-girdle muscular dystrophy, type 2C	AR	1 in 381
SGSH	Mucopolysaccharidosis IIIA (Sanfilippo syndrome A)	AR	1 in 454
SH3TC2	Charcot-Marie-Tooth disease, SH3TC2- related	AR	1 in 69
SLC12A6	Andermann syndrome	AR	<1 in 500
SLC16A2	Allan-Herndon-Dudley syndrome	XL	<1 in 500
SLC17A5	Sialic acid storage disorder	AR	<1 in 500
SLC19A3	Biotin-responsive basal ganglia disease	AR	1 in 109
SLC1A4	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly syndrome	AR	<1 in 500
SLC22A5	Systemic primary carnitine deficiency	AR	1 in 129
SLC25A13	Citrin deficiency	AR	<1 in 500
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome (Triple H syndrome)	AR	<1 in 500
SLC26A2	SLC26A2-related disorders	AR	1 in 158
SLC26A3	Congenital secretory chloride diarrhea	AR	<1 in 500
SLC35A3	Arthrogryposis, intellectual disability, and seizures	AR	<1 in 500
SLC37A4	Glycogen storage disease, type Ib	AR	1 in 158
SLC39A4	Acrodermatitis enteropathica	AR	<1 in 500
SLC45A2	Oculocutaneous albinism, type IV	AR	1 in 159
SLC46A1	Hereditary folate malabsorption	AR	<1 in 500
SLC5A5	Thyroid dyshormonogenesis, SLC5A5-related	AR	<1 in 500

*Carrier Frequency for the general population
Abbreviations: AR, autosomal recessive; XL, X-linked

SLC6A8	Creatine deficiency syndrome	XL	1 in 3,434
SLC7A7	Lysinuric protein intolerance	AR	<1 in 500
SMARCAL1	Schimke immunoosseous dysplasia	AR	1 in 500
SMPD1	Niemann-Pick disease, type A/B	AR	1 in 250
SPG11	SPG11-related Neuromuscular Disorders	AR	1 in 159
SPINK5	Netherton syndrome	AR	1 in 224
STAR	Lipoid congenital adrenal hyperplasia	AR	<1 in 500
SUMF1	Multiple sulfatase deficiency	AR	1 in 500
SURF1	Charcot-Marie-Tooth disease, SURF1-related	AR	<1 in 500
SURF1	Leigh syndrome, SURF1-related	AR	<1 in 500
SYN1	X-linked epilepsy with variable learning disabilities	XL	<1 in 50,000
TCIRG1	Osteopetrosis 1	AR	1 in 250
TCTN2	TCTN2-related ciliopathies	AR	<1 in 500
TECPR2	Spastic paraplegia 49	AR	<1 in 500
TF	Atransferrinemia	AR	1 in 116
TG	Thyroid dyshormonogenesis, TG-related	AR	1 in 241
TGM1	Congenital ichthyosis	AR	1 in 224
TH	Segawa syndrome	AR	<1 in 500
THOC2	X-linked Intellectual disability, THOC2-related	XL	<1 in 50,000
TMEM216	TMEM216-related ciliopathies	AR	1 in 141
TPO	Thyroid dyshormonogenesis, TPO-related	AR	1 in 373
TPP1	Neuronal ceroid lipofuscinosis, TPP1-related	AR	1 in 252
TRDN	Catecholaminergic polymorphic ventricular tachycardia	AR	1 in 354
TRIM32	TRIM32-related disorders	AR	<1 in 500
TRMU	Liver failure, acute infantile	AR	<1 in 500
TSEN54	Pontocerebellar hypoplasia type 2A	AR	1 in 250
TSFM	Combined oxidative phosphorylation deficiency, TSFM-related	AR	<1 in 500
TSHB	Congenital hypothyroidism, TSHB-related	AR	1 in 500
TTC37	Trichohepatoenteric syndrome	AR	1 in 500
TTPA	Ataxia with isolated vitamin E deficiency	AR	<1 in 500
TYMP	Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease	AR	<1 in 500
TYR	Oculocutaneous albinism types 1A and 1B	AR	1 in 20
TYRP1	Oculocutaneous albinism, type III	AR	<1 in 500
UGT1A1	Crigler-Najjar syndrome	AR	<1 in 500
UPF3B	Lujan-Fryns syndrome, UPF3B-related	XL	<1 in 50,000
USH1C	USH1C-related disorders	AR	1 in 353
USH1G	Usher syndrome type IG	AR	1 in 434
USH2A	Usher syndrome, type 2A	AR	1 in 126
VPS13A	Choreoacanthocytosis	AR	<1 in 500
VPS13B	Cohen syndrome	AR	<1 in 500
VPS45	Severe congenital neutropenia, VPS45-related	AR	1 in 224
VPS53	Pontocerebellar hypoplasia type 2E	AR	<1 in 500
VRK1	Pontocerebellar hypoplasia type 1A	AR	<1 in 500
VSX2	Microphthalmia with or without coloboma	AR	1 in 91
WAS	WAS-related hematopoietic disorder	XL	1 in 125,000
WHRN	Usher syndrome type 2D	AR	1 in 282
WRN	Werner syndrome	AR	1 in 308

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Abbreviations: AR, autosomal recessive; XL, X-linked

XPA	Xeroderma pigmentosum, group A	AR	1 in 500
XPC	Xeroderma pigmentosum, group C	AR	1 in 500
ZDHHC9	X-linked intellectual disability, ZDHHC9- related	XL	<1 in 50,000
ZFYVE26	Spastic paraplegia 15	AR	<1 in 500
ZNF711	X-linked intellectual disability, ZNF711-related	XL	<1 in 50,000

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Abbreviations: AR, autosomal recessive; XL, X-linked