



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<i>ABCA3</i>	Surfactant metabolism dysfunction, pulmonary 3	AR	General Population	1 in 116	99%	1 in 11,501	1 in 5,336,464
<i>ABCB11</i>	Progressive familial intrahepatic cholestasis	AR	General Population	1 in 112	98%	1 in 5,551	1 in 2,486,848
<i>ABCC8</i>	Familial hyperinsulinism	AR	General Population	1 in 112	98%	1 in 5,551	1 in 2,486,848
			Ashkenazi Jewish Population	1 in 44	98%	1 in 2,151	1 in 378,576
			Finnish Population	1 in 25	98%	1 in 1,201	1 in 120,100
			Middle-Eastern Population	1 in 25	98%	1 in 1,201	1 in 120,100
<i>ABCD1</i>	Adrenoleukodystrophy, X-linked	XL	General Population	1 in 21,000	99%	1 in 2,099,901	1 in 8,399,804
<i>ABCD4</i>	Methylmalonic aciduria and homocystinuria, cblJ type	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>ACAD9</i>	Acyl-CoA dehydrogenase-9 (ACAD9) deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>ACADM</i>	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	AR	General Population	1 in 69	98%	1 in 3,401	1 in 938,676
			Caucasian / European Population	1 in 52	99%	1 in 5,101	1 in 1,061,008
			East Asian Population	1 in 198	99%	1 in 19,701	<1 in 10 million
			Native American Population	1 in 43	96%	1 in 1,051	1 in 180,772
<i>ACADS</i>	Short-chain acyl-coA dehydrogenase (SCAD) deficiency	AR	General Population	1 in 85	99%	1 in 8,401	1 in 2,856,340
			African/African American Population	1 in 52	99%	1 in 5,101	1 in 1,061,008
			Caucasian / European Population	1 in 76	99%	1 in 7,501	1 in 2,280,304
			Middle-Eastern Population	1 in 52	99%	1 in 5,101	1 in 1,061,008
			South Asian/Indian Population	1 in 51	99%	1 in 5,001	1 in 1,020,204
<i>ACADSB</i>	Short branched chain acyl-CoA dehydrogenase (SBCAD) deficiency	AR	General Population	1 in 368	99%	1 in 36,701	<1 in 10 million
			Hmong Population	1 in 6	99%	1 in 501	<1 in 10 million
<i>ACADVL</i>	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	AR	General Population	1 in 118	93%	1 in 1,672	1 in 789,184
			Middle-Eastern Population	1 in 74	93%	1 in 1,044	1 in 309,024
			Native American Population	1 in 61	93%	1 in 858	1 in 209,352
			South Asian/Indian Population	1 in 73	93%	1 in 1,030	1 in 300,760
<i>ACAT1</i>	3-ketothiolase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>ACOX1</i>	Peroxisomal acyl-CoA oxidase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>ACSF3</i>	Combined malonic and methylmalonic aciduria	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>ADA</i>	Adenosine deaminase deficiency	AR	General Population	1 in 224	93%	1 in 3,187	1 in 2,855,552
<i>ADAMTS2</i>	Ehlers-Danlos syndrome, dermatosparaxis type	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Ashkenazi Jewish Population	1 in 248	98%	1 in 12,351	<1 in 10 million
<i>ADGRG1</i>	Bilateral frontoparietal polymicrogyria	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>ADK</i>	Hypermethioninemia due to adenosine kinase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>AFF2</i>	Fragile XE syndrome	XL	General Population	<1 in 50,000	98%	1 in 2,499,951	1 in 9,999,804
<i>AGA</i>	Aspartylglucosaminuria	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Finnish Population	1 in 71	98%	1 in 3,501	1 in 994,284
<i>AGL</i>	Glycogen storage disease type III	AR	General Population	1 in 158	95%	1 in 3,141	1 in 1,985,112
			Faroese Population	1 in 28	95%	1 in 541	1 in 60,592
			Inuit Population	1 in 25	95%	1 in 481	1 in 48,100
			North African Jewish Population	1 in 37	95%	1 in 721	1 in 106,708
<i>AGPS</i>	Rhizomelic chondrodysplasia punctata, type 3	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>AGXT</i>	Primary hyperoxaluria type 1	AR	General Population	1 in 120	99%	1 in 11,901	1 in 5,712,480
			Caucasian / European Population	1 in 173	99%	1 in 17,201	<1 in 10 million
<i>AHCY</i>	Hypermethioninemia due to deficiency of S-adenosylhomocysteine hydrolase	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>AHI1</i>	Joubert syndrome, AHI1-related	AR	General Population	1 in 448	99%	1 in 44,701	<1 in 10 million
<i>AIP1</i>	Childhood-onset severe retinal dystrophy, AIP1-related	AR	General Population	1 in 409	99%	1 in 40,801	<1 in 10 million
<i>AIRE</i>	Autoimmune polyendocrinopathy syndrome type I	AR	General Population	1 in 150	98%	1 in 7,451	1 in 4,470,600
			Finnish Population	1 in 79	98%	1 in 3,901	1 in 1,232,716
<i>ALDH3A2</i>	Sjögren-Larsson syndrome	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
<i>ALDH4A1</i>	Hyperprolinemia type II	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>ALDOB</i>	Hereditary fructose intolerance	AR	General Population	1 in 122	99%	1 in 12,101	1 in 5,905,288
			African/African American Population	1 in 250	99%	1 in 24,901	<1 in 10 million
			Caucasian / European Population	1 in 67	99%	1 in 6,601	1 in 1,769,068
			Middle-Eastern Population	1 in 97	99%	1 in 9,601	1 in 3,725,188
<i>ALG6</i>	Congenital disorder of glycosylation type Ic	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>ALMS1</i>	Alstrom syndrome	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
ALPL	Hypophosphatasia	AR	General Population	1 in 158	95%	1 in 3,141	1 in 1,985,112
			Caucasian / European Population	1 in 274	95%	1 in 5,461	1 in 5,985,256
			Mennonite Population	1 in 25	95%	1 in 481	1 in 48,100
AMT	Glycine encephalopathy	AR	General Population	1 in 373	98%	1 in 18,601	<1 in 10 million
			Finnish Population	1 in 117	98%	1 in 5,801	1 in 2,714,868
ANO10	Spinocerebellar ataxia 10	AR	General Population	1 in 93	99%	1 in 9,201	1 in 3,422,772
AP1S2	X-linked Intellectual disability, AP1S2-related	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
AQP2	Nephrogenic diabetes insipidus	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
			Finnish Population	1 in 169	95%	1 in 3,361	1 in 2,272,036
ARG1	Arginase deficiency	AR	General Population	1 in 296	98%	1 in 14,751	<1 in 10 million
ARL13B	Joubert syndrome, ARL13B-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
ARSA	Metachromatic leukodystrophy	AR	General Population	1 in 100	99%	1 in 9,901	1 in 3,960,400
			Caucasian / European Population	1 in 78	99%	1 in 7,701	1 in 2,402,712
			Yemenite Jewish Population	1 in 75	99%	1 in 7,401	1 in 2,220,300
ARSB	Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome)	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
			Western Australian Population	1 in 283	98%	1 in 14,101	<1 in 10 million
ARSE	Chondrodysplasia punctata type 1, X-linked	XL	General Population	1 in 250,000	98%	1 in 12,499,951	<1 in 10 million
ARX	X-linked intellectual disability, ARX-related	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
ASL	Argininosuccinate lyase deficiency	AR	General Population	1 in 132	90%	1 in 1,311	1 in 692,208
ASNS	Asparagine synthetase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Iranian Jewish Population	1 in 80	99%	1 in 7,901	1 in 2,528,320
ASPA	Canavan disease	AR	General Population	1 in 300	97%	1 in 9,968	<1 in 10 million
			Ashkenazi Jewish Population	1 in 55	96%	1 in 1,351	1 in 297,220
ASS1	Citrullinemia	AR	General Population	1 in 119	96%	1 in 2,951	1 in 1,404,676
			East Asian Population	1 in 132	96%	1 in 3,276	1 in 1,729,728
ATM	Ataxia-telangiectasia	AR	General Population	1 in 100	92%	1 in 1,239	1 in 495,600
ATP6V1B1	Renal tubular acidosis with deafness	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
ATP7A	Menkes disease	XL	General Population	1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
ATP7B	Wilson disease	AR	General Population	1 in 87	98%	1 in 4,301	1 in 1,496,748
			Caucasian / European Population	1 in 42	98%	1 in 2,051	1 in 344,568
			Ashkenazi Jewish Population	1 in 70	98%	1 in 3,451	1 in 966,280
ATRX	Alpha thalassemia X-linked intellectual disability syndrome	XL	General Population	<1 in 250,000	99%	1 in 24,999,901	<1 in 10 million
BBS1	Bardet-Biedl syndrome type 1	AR	General Population	1 in 367	99%	1 in 36,601	<1 in 10 million
BBS10	Bardet-Biedl syndrome type 10	AR	General Population	1 in 395	99%	1 in 39,401	<1 in 10 million
BBS12	Bardet-Biedl syndrome type 12	AR	General Population	1 in 791	99%	1 in 79,001	<1 in 10 million
BBS2	Bardet-Biedl syndrome 2	AR	General Population	1 in 621	99%	1 in 62,001	<1 in 10 million
			Ashkenazi Jewish Population	1 in 107	99%	1 in 10,601	1 in 4,537,228
BBS2	Retinitis Pigmentosa 74	AR	General Population	1 in 621	99%	1 in 62,001	<1 in 10 million
			Ashkenazi Jewish Population	1 in 107	99%	1 in 10,601	1 in 4,537,228
BCHE	Butyrylcholinesterase deficiency	AR	General Population	1 in 28	99%	1 in 2,701	1 in 302,512
BCKDHA	Maple syrup urine disease type Ia	AR	General Population	1 in 321	98%	1 in 16,001	<1 in 10 million
			Mennonite Population	1 in 10	98%	1 in 451	1 in 18,040
BCKDHB	Maple syrup urine disease type Ib	AR	General Population	1 in 364	98%	1 in 18,151	<1 in 10 million
			Ashkenazi Jewish Population	1 in 97	98%	1 in 4,801	1 in 1,862,788
BCS1L	Björnstad syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
BCS1L	GRACILE syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
BCS1L	Mitochondrial complex III deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
BLM	Bloom syndrome	AR	General Population	1 in 800	87%	1 in 6,147	<1 in 10 million
			Ashkenazi Jewish Population	1 in 134	99%	1 in 13,301	1 in 7,129,336
BRWD3	X-linked intellectual disability, BRWD3-related	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
BSND	Bartter syndrome	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
BTD	Biotinidase deficiency	AR	General Population	1 in 124	99%	1 in 12,301	1 in 6,101,296
			Caucasian / European Population	1 in 71	99%	1 in 7,001	1 in 1,988,284
			Latino Population	1 in 136	99%	1 in 13,501	1 in 7,344,544
			Middle-Eastern Population	1 in 55	99%	1 in 5,401	1 in 1,188,220
CAPN3	Limb-girdle muscular dystrophy type 2A	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Caucasian / European Population	1 in 103	98%	1 in 5,101	1 in 2,101,612
CASQ2	Catecholaminergic polymorphic ventricular tachycardia	AR	General Population	1 in 224	99%	1 in 22,301	<1 in 10 million



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
CBS	Homocystinuria due to cystathionine beta-synthase deficiency	AR	General Population	1 in 224	99%	1 in 22,301	<1 in 10 million
			Caucasian / European Population	1 in 86	99%	1 in 8,501	1 in 2,924,344
			Middle-Eastern Population	1 in 21	99%	1 in 2,001	1 in 168,084
CC2D2A	Joubert syndrome 9	AR	General Population	1 in 201	99%	1 in 20,001	1 in 16,080,804
CCDC103	Primary ciliary dyskinesia, type 17	AR	General Population	1 in 316	98%	1 in 15,751	<1 in 10 million
CCDC151	Primary ciliary dyskinesia, type 30	AR	General Population	1 in 365	98%	1 in 18,201	<1 in 10 million
CCDC39	Primary ciliary dyskinesia, type 14	AR	General Population	1 in 211	98%	1 in 10,501	1 in 8,862,844
CCDC88C	Congenital hydrocephalus 1	AR	General Population	1 in 137	99%	1 in 13,601	1 in 7,453,348
CD40LG	Hyper IgM syndrome, X-linked	XL	General Population	1 in 50,000	98%	1 in 2,499,951	1 in 9,999,904
CDH23	Usher syndrome, type 1D	AR	General Population	1 in 285	90%	1 in 2,841	1 in 11,364
CEP290	Joubert syndrome 5	AR	General Population	1 in 190	98%	1 in 9,451	1 in 7,182,760
CEP290	Leber congenital amaurosis 10	AR	General Population	1 in 190	98%	1 in 9,451	1 in 7,182,760
CEP290	Bardet-Biedl syndrome 14	AR	General Population	1 in 190	98%	1 in 9,451	1 in 7,182,760
CEP290	CEP290-related disorders	AR	General Population	1 in 190	98%	1 in 9,451	1 in 7,182,760
CEP290	Senior-Løken syndrome 6	AR	General Population	1 in 190	98%	1 in 9,451	1 in 7,182,760
CEP290	Meckel syndrome 4	AR	General Population	1 in 190	98%	1 in 9,451	1 in 7,182,760
CERKL	Retinitis pigmentosa 26	AR	General Population	1 in 148	98%	1 in 7,351	1 in 4,351,792
CFTR	Cystic Fibrosis	AR	General Population	1 in 32	99%	1 in 3,101	1 in 396,928
			African/African American Population	1 in 61	99%	1 in 6,001	1 in 1,464,244
			Ashkenazi Jewish Population	1 in 24	99%	1 in 2,301	1 in 220,896
			Caucasian / European Population	1 in 25	99%	1 in 2,401	1 in 240,100
			East Asian Population	1 in 94	99%	1 in 9,301	1 in 3,497,176
			Latino Population	1 in 58	99%	1 in 5,701	1 in 1,322,632
CHM	Choroideremia	XL	General Population	1 in 25,000	95%	1 in 499,981	1 in 1,999,964
CHRNE	Congenital myasthenic syndrome	AR	General Population	1 in 408	99%	1 in 40,701	<1 in 10 million
CHRNA3	Multiple pterygium syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
CHST6	Macular corneal dystrophy, CHST6-related	AR	General Population	1 in 79	99%	1 in 7,801	1 in 2,465,116
CIITA	Bare lymphocyte syndrome, type II	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
CLCN1	Autosomal recessive congenital myotonia	AR	General Population	1 in 176	99%	1 in 17,501	1 in 12,320,704
CLN3	Neuronal ceroid lipofuscinosis	AR	General Population	1 in 230	98%	1 in 11,451	<1 in 10 million
			Finnish Population	1 in 72	98%	1 in 3,551	1 in 1,022,688
CLN5	Neuronal ceroid lipofuscinosis 5	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
			Finnish Population	1 in 115	95%	1 in 2,281	1 in 1,049,260
CLN6	Neuronal ceroid lipofuscinosis, CLN6-related	AR	General Population	<1 in 500	92%	1 in 6,239	<1 in 10 million
CLN8	Neuronal ceroid lipofuscinosis, CLN8-related	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
			Finnish Population	1 in 135	95%	1 in 2,681	1 in 1,447,740
CLRN1	Usher syndrome, type 3A	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
			Ashkenazi Jewish Population	1 in 120	98%	1 in 5,951	1 in 2,856,480
			Finnish Population	1 in 70	98%	1 in 3,451	1 in 966,280
CNGA1	Retinitis Pigmentosa, CNGA1-related	AR	General Population	1 in 210	99%	1 in 20,901	<1 in 10 million
CNGB1	Retinitis Pigmentosa, CNGB1-related	AR	General Population	1 in 296	99%	1 in 29,501	<1 in 10 million
CNGB3	Achromatopsia	AR	General Population	1 in 87	99%	1 in 8,601	1 in 2,993,148
			Micronesian Population	1 in 2	99%	1 in 101	1 in 808
COL27A1	Steel syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
COL4A3	Alport syndrome, COL4A3-related	AR	General Population	1 in 267	98%	1 in 13,301	<1 in 10 million
			Ashkenazi Jewish Population	1 in 188	98%	1 in 9,351	1 in 7,031,952
COL4A4	Alport syndrome, COL4A4-related	AR	General Population	1 in 267	98%	1 in 13,301	<1 in 10 million
COL4A5	Alport syndrome, COL4A5-related	XL	General Population	1 in 139	98%	1 in 6,901	1 in 27,604
COL7A1	Dystrophic epidermolysis bullosa	AR	General Population	1 in 196	97%	1 in 6,501	1 in 5,096,784
CPS1	Carbamoylphosphate synthetase I deficiency	AR	General Population	1 in 570	98%	1 in 28,451	<1 in 10 million
CPT1A	Carnitine palmitoyltransferase IA deficiency	AR	General Population	1 in 354	90%	1 in 3,531	1 in 4,999,896
			Hutterite Population	1 in 16	90%	1 in 151	1 in 9,664
CPT2	Carnitine palmitoyltransferase II deficiency	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
			Ashkenazi Jewish Population	1 in 51	95%	1 in 1,001	1 in 204,204
CRB1	Leber congenital amaurosis 8	AR	General Population	1 in 104	98%	1 in 5,151	1 in 2,142,816
CRB1	Retinitis pigmentosa 12	AR	General Population	1 in 104	98%	1 in 5,151	1 in 2,142,816
CRYL1	GJB6-CRYL1 related nonsyndromic hearing loss	UK	General Population	1 in 423	99%	1 in 42,201	<1 in 10 million
CTNS	Cystinosis	AR	General Population	1 in 158	99%	1 in 15,701	1 in 9,923,032
			British Population	1 in 81	99%	1 in 8,001	1 in 2,592,324
			Moroccan Jewish Population	1 in 100	99%	1 in 9,901	1 in 3,960,400



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<i>CTSK</i>	Pycnodysostosis	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>CUL4B</i>	X-linked intellectual disability, CUL4B-related	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
<i>CYBA</i>	Chronic granulomatous disease	AR	General Population	1 in 224	99%	1 in 22,301	<1 in 10 million
<i>CYBB</i>	Chronic granulomatous disease, X-linked	XL	General Population	1 in 149,254	99%	1 in 14,925,301	<1 in 10 million
<i>CYP11A1</i>	Congenital adrenal insufficiency	AR	General Population	1 in 114	99%	1 in 11,301	1 in 5,153,256
<i>CYP11B1</i>	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	AR	General Population Moroccan Jewish Population	1 in 158 1 in 35	98% 98%	1 in 7,851 1 in 1,701	1 in 4,961,832 1 in 238,140
<i>CYP11B2</i>	Corticosterone methyl oxidase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>CYP17A1</i>	Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
<i>CYP19A1</i>	Aromatase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>CYP1B1</i>	Primary congenital glaucoma	AR	General Population	1 in 50	99%	1 in 4,901	1 in 980,200
<i>CYP21A2</i>	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	AR	General Population Inuit Population Middle-Eastern Population	1 in 61 1 in 9 1 in 35	99% 99% 99%	1 in 6,001 1 in 801 1 in 3,401	1 in 1,464,244 1 in 28,836 1 in 476,140
<i>CYP27A1</i>	Cerebrotendinous xanthomatosis	AR	General Population Moroccan Jewish Population	1 in 500 1 in 5	98% 98%	1 in 24,951 1 in 201	<1 in 10 million 1 in 4,020
<i>CYP27B1</i>	Vitamin D-dependent rickets, type 1	AR	General Population	1 in 181	99%	1 in 18,001	1 in 13,032,724
<i>DBT</i>	Maple syrup urine disease, type II	AR	General Population	1 in 481	98%	1 in 24,001	<1 in 10 million
<i>DCLRE1C</i>	Severe combined immunodeficiency with sensitivity to ionizing radiation	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>DCX</i>	Lissencephaly, X-linked	XL	General Population	1 in 42,500	98%	1 in 2,124,951	1 in 8,499,904
<i>DHCR7</i>	Smith-Lemli-Opitz syndrome	AR	General Population African/African American Population Ashkenazi Jewish Population	1 in 30 1 in 138 1 in 36	96% 96% 96%	1 in 726 1 in 3,426 1 in 876	1 in 87,120 1 in 1,891,152 1 in 126,144
<i>DHDDS</i>	Retinitis pigmentosa 59	AR	General Population Ashkenazi Jewish Population	1 in 296 1 in 118	98% 98%	1 in 14,751 1 in 5,851	<1 in 10 million 1 in 2,761,672
<i>DLD</i>	Dihydrolipoamide dehydrogenase deficiency	AR	General Population Ashkenazi Jewish Population	1 in 500 1 in 107	98% 98%	1 in 24,951 1 in 5,301	<1 in 10 million 1 in 2,268,828
<i>DLG3</i>	X-linked intellectual disability, DLG3-related	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
<i>DMD</i>	Duchenne Muscular Dystrophy	XL	General Population	1 in 2,350	93%	1 in 33,558	1 in 134,260
<i>DMD</i>	Becker Muscular Dystrophy	XL	General Population	1 in 2,350	93%	1 in 33,558	1 in 134,260
<i>DNAH5</i>	Primary ciliary dyskinesia, DNAH5-related	AR	General Population Ashkenazi Jewish Population	1 in 142 1 in 113	98% 99%	1 in 7,051 1 in 11,201	1 in 4,004,968 1 in 5,062,852
<i>DNAI1</i>	Primary ciliary dyskinesia, DNAI1-related	AR	General Population	1 in 230	98%	1 in 11,451	<1 in 10 million
<i>DNAI2</i>	Primary ciliary dyskinesia, DNAI2-related	AR	General Population	1 in 447	98%	1 in 22,301	<1 in 10 million
<i>DNAL1</i>	Primary ciliary dyskinesia, DNAL1-related	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>DPYD</i>	Dihydropyrimidine dehydrogenase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>DUOX2</i>	Congenital hypothyroidism, DUOX2-related	AR	General Population	1 in 366	91%	1 in 4,057	1 in 5,938,797
<i>DUOX2</i>	Congenital hypothyroidism, DUOX2-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>DYNC2H1</i>	Short-rib thoracic dysplasia 3 with or without polydactyly	AR	General Population	1 in 68	98%	1 in 3,351	1 in 924,876
<i>DYSF</i>	Limb-girdle muscular dystrophy type 2B	AR	General Population Japanese Population Libyan Jewish Population	<1 in 500 1 in 332 1 in 18	95% 95% 95%	1 in 9,981 1 in 6,621 1 in 341	<1 in 10 million 1 in 8,792,688 1 in 24,552
<i>EDA</i>	Hypohidrotic ectodermal dysplasia	XL	General Population	1 in 14,167	99%	1 in 1,416,601	1 in 5,666,472
<i>EIF2AK3</i>	Wolcott-Rallison Syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>EIF2B5</i>	Leukoencephalopathy with vanishing white matter	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>ELP1</i>	Familial Dysautonomia	AR	General Population Ashkenazi Jewish Population	1 in 300 1 in 31	99% 99%	1 in 29,901 1 in 3,001	<1 in 10 million 1 in 372,124
<i>EMD</i>	Emery-Dreifuss muscular dystrophy	XL	General Population	1 in 81,967	99%	1 in 8,196,601	<1 in 10 million
<i>ERCC2</i>	Xeroderma pigmentosum, group D	AR	General Population	1 in 65	99%	1 in 6,401	1 in 1,664,260
<i>ERCC2</i>	Photosensitive trichothiodystrophy 1	AR	General Population	1 in 65	99%	1 in 6,401	1 in 1,664,260
<i>ERCC2</i>	Cerebrooculofacioskeletal syndrome 2	AR	General Population	1 in 65	99%	1 in 6,401	1 in 1,664,260
<i>ERCC6</i>	De Sanctis-Cacchione syndrome	AR	General Population Japanese Population	1 in 500 1 in 74	99% 99%	1 in 49,901 1 in 7,301	<1 in 10 million 1 in 2,161,096
<i>ERCC6</i>	Cockayne syndrome type B	AR	General Population Japanese Population	1 in 500 1 in 74	99% 99%	1 in 49,901 1 in 7,301	<1 in 10 million 1 in 2,161,096
<i>ERCC8</i>	Cockayne syndrome type A	AR	General Population	1 in 822	98%	1 in 41,051	<1 in 10 million
<i>ESCO2</i>	Roberts syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<i>ETFA</i>	Glutaric aciduria IIA	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
<i>ETFB</i>	Glutaric aciduria IIB	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
<i>ETFDH</i>	Glutaric aciduria IIC	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
			East Asian Population	1 in 74	98%	1 in 3,651	1 in 1,080,696
<i>ETHE1</i>	Ethylmalonic encephalopathy	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>EVC</i>	Weyers acrofacial dysostosis, EVC-related	AR	General Population	1 in 142	98%	1 in 7,051	1 in 4,004,968
			Amish Population	1 in 7	98%	1 in 301	1 in 8,428
<i>EVC</i>	Ellis-van Creveld syndrome, EVC-related	AR	General Population	1 in 142	98%	1 in 7,051	1 in 4,004,968
			Amish Population	1 in 7	98%	1 in 301	1 in 8,428
<i>EVC2</i>	Weyers acrofacial dysostosis, EVC2-related	AR	General Population	1 in 240	98%	1 in 11,951	<1 in 10 million
			Amish Population	1 in 7	98%	1 in 301	1 in 8,428
<i>EVC2</i>	Ellis-van Creveld syndrome, EVC2-related	AR	General Population	1 in 240	98%	1 in 11,951	<1 in 10 million
			Amish Population	1 in 7	98%	1 in 301	1 in 8,428
<i>EXOSC3</i>	Pontocerebellar hypoplasia type 1B	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>EYS</i>	Retinitis pigmentosa 25	AR	General Population	1 in 66	98%	1 in 3,251	1 in 858,264
<i>F11</i>	Factor XI deficiency	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
			Ashkenazi Jewish Population	1 in 11	98%	1 in 501	1 in 22,044
<i>F2</i>	Prothrombin-related conditions	AR	General Population	1 in 33	99%	1 in 3,201	1 in 422,532
			Caucasian / European Population	1 in 4	99%	1 in 301	1 in 4,816
<i>F5</i>	Factor V deficiency	AR	General Population	1 in 36	99%	1 in 3,501	1 in 504,144
			Caucasian / European Population	1 in 19	99%	1 in 1,801	1 in 136,876
			Latino Population	1 in 45	99%	1 in 4,401	1 in 792,180
			African/African American Population	1 in 83	99%	1 in 8,201	1 in 2,722,732
			East Asian Population	1 in 222	99%	1 in 22,101	<1 in 10 million
			Native American Population	1 in 80	99%	1 in 7,901	1 in 2,528,320
<i>F8</i>	Hemophilia A	XL	General Population	1 in 3,250	48%	1 in 6,249	1 in 25,000
<i>F9</i>	Hemophilia B	XL	General Population	1 in 15,000	99%	1 in 1,499,901	1 in 5,999,804
<i>FAH</i>	Tyrosinemia, type 1	AR	General Population	1 in 99	95%	1 in 1,961	1 in 776,556
			Ashkenazi Jewish Population	1 in 150	95%	1 in 2,981	1 in 1,788,600
			Finnish Population	1 in 122	95%	1 in 2,421	1 in 1,181,448
			French Canadian Population	1 in 66	95%	1 in 1,301	1 in 343,464
			South Asian/Indian Population	1 in 172	95%	1 in 3,421	1 in 2,353,648
<i>FAM161A</i>	Retinitis pigmentosa 28	AR	General Population	1 in 296	98%	1 in 14,751	<1 in 10 million
<i>FANCA</i>	Fanconi anemia group A	AR	General Population	1 in 239	99%	1 in 23,801	<1 in 10 million
			Moroccan Jewish	1 in 100	99%	1 in 9,901	1 in 3,960,400
			Indian Jewish Population	1 in 27	99%	1 in 2,601	1 in 280,908
<i>FANCC</i>	Fanconi anemia group C	AR	General Population	1 in 535	99%	1 in 53,401	<1 in 10 million
			Ashkenazi Jewish Population	1 in 99	99%	1 in 9,801	1 in 3,881,196
<i>FANCG</i>	Fanconi anemia group G	AR	General Population	1 in 632	90%	1 in 6,311	<1 in 10 million
<i>FGD1</i>	X-linked Aarskog-Scott syndrome	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
<i>FH</i>	Fumarate deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Ashkenazi Jewish Population	1 in 99	99%	1 in 9,801	1 in 3,881,196
<i>FKRP</i>	Muscular dystrophy-dystroglycanopathy, FKRP-related	AR	General Population	1 in 158	98%	1 in 7,851	1 in 4,961,832
<i>FKRP</i>	Walker-Warburg syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>FKTN</i>	Muscular dystrophy-dystroglycanopathy, FKTN-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Ashkenazi Jewish Population	1 in 150	99%	1 in 14,901	1 in 8,940,600
			Japanese Population	1 in 82	99%	1 in 8,101	1 in 2,657,128
<i>FKTN</i>	Fukuyama congenital muscular dystrophy	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Ashkenazi Jewish Population	1 in 150	99%	1 in 14,901	1 in 8,940,600
			Japanese Population	1 in 82	99%	1 in 8,101	1 in 2,657,128
<i>FKTN</i>	Walker-Warburg syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>FMO3</i>	Trimethylaminuria	AR	General Population	1 in 139	99%	1 in 13,801	1 in 7,763,356
<i>FMR1</i>	Fragile X Syndrome Intermediate Allele	XL	General Population	1 in 259	99%	1 in 25,801	1 in 103,204
			Ashkenazi Jewish Population	1 in 115	99%	1 in 11,401	1 in 45,604
<i>FMR1</i>	Fragile X Syndrome Premutation	XL	General Population	1 in 259	99%	1 in 25,801	1 in 103,204
			Ashkenazi Jewish Population	1 in 115	99%	1 in 11,401	1 in 45,604
<i>FMR1</i>	Fragile X Syndrome Full Mutation	XL	General Population	1 in 11,111	99%	1 in 1,111,001	1 in 4,444,004
<i>FTCD</i>	Glutamate formiminotransferase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>FTSJ1</i>	X-linked intellectual disability, FTSJ1-related	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
<i>FXN</i>	Friedreich ataxia	AR	General Population	1 in 80	4%	1 in 83	1 in 26,653
			Caucasian / European Population	1 in 80	98%	1 in 3951	1 in 1,264,320



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<i>G6PC</i>	Glycogen storage disease, type 1a	AR	General Population	1 in 177	95%	1 in 3,521	1 in 2,492,868
			Ashkenazi Jewish Population	1 in 64	95%	1 in 1,261	1 in 322,816
<i>G6PD</i>	Glucose-6-phosphate dehydrogenase deficiency	XL	General Population	1 in 7	98%	1 in 301	1 in 1,204
<i>GAA</i>	Pompe disease	AR	General Population	1 in 100	98%	1 in 4,951	1 in 1,980,400
			African/African American Population	1 in 60	98%	1 in 2,951	1 in 708,240
			East Asian Population	1 in 112	98%	1 in 5,551	1 in 2,486,848
			Ashkenazi Jewish Population	1 in 76	99%	1 in 7,501	1 in 2,280,304
<i>GALC</i>	Krabbe disease	AR	General Population	1 in 158	99%	1 in 15,701	1 in 9,923,032
			Israeli Druze Population	1 in 6	99%	1 in 501	1 in 12,024
<i>GALE</i>	Galactose epimerase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>GALK1</i>	Galactokinase deficiency	AR	General Population	1 in 110	95%	1 in 2,181	1 in 959,640
			Irish Population	1 in 64	95%	1 in 1,261	1 in 322,816
<i>GALNS</i>	Mucopolysaccharidosis IVA (Morquio syndrome A)	AR	General Population	1 in 224	97%	1 in 7,434	1 in 6,660,864
<i>GALT</i>	Galactosemia	AR	General Population	1 in 110	99%	1 in 10,901	1 in 4,796,440
			African/African American Population	1 in 94	99%	1 in 9,301	1 in 3,497,176
			Ashkenazi Jewish Population	1 in 127	99%	1 in 12,601	1 in 6,401,308
<i>GAMT</i>	Guanidinoacetate methyltransferase deficiency	AR	General Population	1 in 371	99%	1 in 37,001	<1 in 10 million
<i>GBA</i>	Gaucher disease	AR	General Population	1 in 77	99%	1 in 7,601	1 in 2,341,108
			African/African American Population	1 in 35	99%	1 in 3,401	1 in 476,140
			Ashkenazi Jewish Population	1 in 15	99%	1 in 1,401	1 in 84,060
<i>GBE1</i>	Glycogen storage disease IV	AR	General Population	1 in 387	99%	1 in 38,601	<1 in 10 million
<i>GCDH</i>	Glutaric aciduria, type I	AR	General Population	1 in 87	98%	1 in 4,301	1 in 1,496,748
			Amish Population	1 in 9	98%	1 in 401	1 in 14,436
<i>GDAP1</i>	Charcot-Marie-Tooth disease, GDAP1-related	AR	General Population	1 in 152	99%	1 in 15,101	1 in 9,181,408
<i>GFM1</i>	Combined oxidative phosphorylation deficiency, GFM1-related	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>GJB1</i>	Charcot-Marie-Tooth disease, X-linked type 1	XL	General Population	1 in 667	90%	1 in 6,661	1 in 26,644
<i>GJB2</i>	Nonsyndromic hearing loss 1A	AR	General Population	1 in 42	99%	1 in 4,101	1 in 688,968
			African/African American Population	1 in 25	99%	1 in 2,401	1 in 240,100
			Ashkenazi Jewish Population	1 in 21	99%	1 in 2,001	1 in 168,084
			Caucasian / European Population	1 in 33	99%	1 in 3,201	1 in 422,532
			Latino Population	1 in 100	99%	1 in 9,901	1 in 3,960,400
			Middle-Eastern Population	1 in 83	99%	1 in 8,201	1 in 2,722,732
<i>GJB6</i>	GJB6-CRYL1 related nonsyndromic hearing loss	AR	General Population	1 in 423	99%	1 in 42,201	<1 in 10 million
<i>GLA</i>	Fabry disease	XL	General Population	1 in 25,000	99%	1 in 2,499,901	1 in 9,999,804
<i>GLB1</i>	GM1-gangliosidosis	AR	General Population	1 in 134	99%	1 in 13,301	1 in 7,129,336
			Maltese Population	1 in 30	99%	1 in 2,901	1 in 348,120
			Roma Population	1 in 50	99%	1 in 4,901	1 in 980,200
<i>GLB1</i>	Mucopolysaccharidosis type IVB (Morquio syndrome B)	AR	General Population	1 in 134	99%	1 in 13,301	1 in 7,129,336
			Maltese Population	1 in 30	99%	1 in 2,901	1 in 348,120
			Roma Population	1 in 50	99%	1 in 4,901	1 in 980,200
<i>GLDC</i>	Glycine encephalopathy, GLDC-related	AR	General Population	1 in 193	98%	1 in 9,601	1 in 7,411,972
			British Columbia Canadian Population	1 in 125	99%	1 in 12,401	1 in 6,200,500
			Finnish Population	1 in 117	99%	1 in 11,601	1 in 5,429,268
<i>GLE1</i>	Lethal congenital contracture syndrome 1	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Finnish Population	1 in 80	98%	1 in 3,951	1 in 1,264,320
<i>GNE</i>	Inclusion body myopathy type 2 (Nonaka myopathy)	AR	General Population	<1 in 500	99%	1 in 49,901	1 in 99,802,000
<i>GNPTAB</i>	Mucopolysaccharidosis II alpha/beta	AR	General Population	1 in 11	99%	1 in 1,001	1 in 44,044
			Iranian Jewish Population				
<i>GNPTAB</i>	Mucopolysaccharidosis III alpha/beta	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
<i>GNPTG</i>	Mucopolysaccharidosis III gamma	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
<i>GNRHR</i>	Hypogonadotropic hypogonadism, GNRHR-related	AR	General Population	1 in 347	99%	1 in 34,601	<1 in 10 million
<i>GNS</i>	Mucopolysaccharidosis IIID (Sanfilippo syndrome D)	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
<i>GP1BA</i>	Bernard-Soulier syndrome type A1	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
<i>GP9</i>	Bernard-Soulier syndrome type C	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
<i>GPR143</i>	X-linked Ocular albinism, GPR143-related	XL	General Population	1 in 25,000	99%	1 in 2,499,901	<1 in 10 million



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<i>GRHPR</i>	Primary hyperoxaluria type II	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>GRIP1</i>	Fraser syndrome	AR	General Population	1 in 84	99%	1 in 8,301	1 in 2,789,136
<i>GUSB</i>	Mucopolysaccharidosis type VII	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
<i>HADHA</i>	Trifunctional protein deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Finnish Population	1 in 124	98%	1 in 6,151	1 in 3,050,896
<i>HADHA</i>	Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Finnish Population	1 in 124	98%	1 in 6,151	1 in 3,050,896
<i>HAX1</i>	Severe congenital neutropenia, HAX1-related	AR	General Population	1 in 224	98%	1 in 11,151	1 in 9,991,296
<i>HBA1</i>	Alpha thalassemia	AR	General Population	1 in 1000	98%	1 in 860	1 in 3,440,364
			General Population†	1 in 18	98%	1 in 860	1 in 3,440,364
			Southeast Asian Population	≤1 in 7	98%	≤1 in 305	≤1 in 17,228
			Southeast Asian Population†	≤1 in 14	98%	≤1 in 305	≤1 in 17,228
			Mediterranean Population	≤1 in 6	98%	≤1 in 229	≤1 in 457,556
			Mediterranean Population†	1 in 500	98%	≤1 in 229	≤1 in 457,556
			African/African American Population	1 in 30	98%	1 in 1,451	1 in 5,804,000
<i>HBA2</i>	Alpha thalassemia	AR	General Population	1 in 1000	98%	1 in 860	1 in 3,440,364
			General Population†	1 in 18	98%	1 in 860	1 in 3,440,364
			Southeast Asian Population	≤1 in 7	98%	≤1 in 305	≤1 in 17,228
			Southeast Asian Population†	≤1 in 14	98%	≤1 in 305	≤1 in 17,228
			Mediterranean Population	≤1 in 6	98%	≤1 in 229	≤1 in 457,556
			Mediterranean Population†	1 in 500	98%	≤1 in 229	≤1 in 457,556
			African/African American Population	1 in 30	98%	1 in 1,451	1 in 5,804,000
<i>HBB</i>	Sickle cell disease	AR	General Population	1 in 158	95%	1 in 3,141	1 in 1,985,112
			African/African American Population	1 in 10	95%	1 in 181	1 in 7,240
			East Asian Population	1 in 50	95%	1 in 981	1 in 196,200
			Latino Population	1 in 128	95%	1 in 2,541	1 in 1,300,992
			Mediterranean Population	1 in 3	95%	1 in 41	1 in 492
			South Asian/Indian Population	1 in 25	95%	1 in 481	1 in 48,100
			<i>HBB</i>	Hemoglobin C disease	AR	General Population	1 in 158
African/African American Population	1 in 10	95%				1 in 181	1 in 7,240
East Asian Population	1 in 50	95%				1 in 981	1 in 196,200
Latino Population	1 in 128	95%				1 in 2,541	1 in 1,300,992
Mediterranean Population	1 in 3	95%				1 in 41	1 in 492
South Asian/Indian Population	1 in 25	95%				1 in 481	1 in 48,100
<i>HBB</i>	Beta thalassemia	AR				General Population	1 in 158
			African/African American Population	1 in 10	99%	1 in 901	1 in 36,040
			East Asian Population	1 in 50	99%	1 in 4,901	1 in 980,200
			Latino Population	1 in 128	99%	1 in 12,701	1 in 6,502,912
			Mediterranean Population	1 in 3	99%	1 in 201	1 in 2,412
			South Asian/Indian Population	1 in 25	99%	1 in 2,401	1 in 240,100
			<i>HEXA</i>	Tay-Sachs disease	AR	General Population	1 in 300
Ashkenazi Jewish Population	1 in 27	99%				1 in 2,601	1 in 280,908
Moroccan Jewish Population	1 in 110	99%				1 in 10,901	1 in 4,796,440
<i>HEXB</i>	Sandhoff disease	AR	General Population	1 in 600	98%	1 in 29,951	<1 in 10 million
<i>HFE</i>	Hereditary Hemochromatosis	AR	General Population	1 in 10	99%	1 in 901	1 in 36,040
			African/African American Population	1 in 17	99%	1 in 1,601	1 in 108,868
			Caucasian / European Population	1 in 3	99%	1 in 201	1 in 2,412
			East Asian Population	1 in 12	99%	1 in 1,101	1 in 52,848
			Latino Population	1 in 6	99%	1 in 501	1 in 12,024
<i>HGD</i>	Alkaptonuria	AR	General Population	1 in 250	90%	1 in 2,491	1 in 2,491,000
<i>HGSNAT</i>	Mucopolysaccharidosis type IIIC (Sanfilippo syndrome C)	AR	General Population	1 in 434	98%	1 in 21,651	<1 in 10 million
			Caucasian / European Population	1 in 345	98%	1 in 17,201	<1 in 10 million
<i>HJV</i>	Hemochromatosis, type 2A	AR	General Population	1 in 500	99%	1 in 49,901	<1 in 10 million
<i>HLCS</i>	Holocarboxylase synthetase deficiency	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
<i>HMGCL</i>	3-hydroxy-3-methylglutaryl-CoA lyase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>HOGA1</i>	Primary hyperoxaluria type III	AR	General Population	1 in 184	99%	1 in 18,301	<1 in 10 million
<i>HPS1</i>	Hermansky-Pudlak syndrome 1	AR	General Population	1 in 354	98%	1 in 17,651	<1 in 10 million
			Puerto Rican Population	1 in 21	98%	1 in 1,001	1 in 84,084
<i>HPS3</i>	Hermansky-Pudlak syndrome 3	AR	General Population	1 in 354	98%	1 in 17,651	<1 in 10 million
<i>HSD17B4</i>	D-bifunctional protein deficiency	AR	General Population	1 in 158	98%	1 in 7,851	1 in 4,961,832
<i>HSD3B2</i>	Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>HYAL1</i>	Mucopolysaccharidosis type IX	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<i>HYLS1</i>	Hydrolethalus syndrome	AR	General Population Finnish Population	<1 in 500 1 in 50	98% 98%	1 in 24,951 1 in 2,451	<1 in 10 million 1 in 490,200
<i>IDH3B</i>	Retinitis pigmentosa, IDH3B-related	AR	General Population	1 in 296	99%	1 in 29,501	<1 in 10 million
<i>IDS</i>	Mucopolysaccharidosis type II (Hunter syndrome)	XL	General Population	1 in 50,000	91%	1 in 555,545	1 in 2,222,204
<i>IDUA</i>	Mucopolysaccharidosis, type I (Hurler syndrome)	AR	General Population Caucasian / European Population	<1 in 500 1 in 153	95% 95%	1 in 9,981 1 in 3,041	<1 in 10 million 1 in 1,861,092
<i>IL1RAPL1</i>	X-linked intellectual disability, IL1RAPL1-related	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
<i>IL2RG</i>	Severe combined immunodeficiency, X-linked	XL	General Population	1 in 25,000	99%	1 in 2,499,901	1 in 9,999,804
<i>IVD</i>	Isovaleric Acidemia	AR	General Population African/African American Population Caucasian / European Population East Asian Population	1 in 167 1 in 100 1 in 115 1 in 407	90% 90% 90% 90%	1 in 1,661 1 in 991 1 in 1,141 1 in 4,061	1 in 1,109,548 1 in 396,400 1 in 524,860 1 in 6,611,308
<i>IYD</i>	Thyroid dysmorphogenesis, IYD-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>JAK3</i>	Severe combined immunodeficiency, JAK3-related	AR	General Population	1 in 299	99%	1 in 29,801	<1 in 10 million
<i>KCNJ11</i>	Congenital hyperinsulinism	AR	General Population Caucasian / European Population	1 in 423 1 in 232	99% 99%	1 in 42,201 1 in 23,101	<1 in 10 million <1 in 10 million
<i>KCNJ11</i>	Permanent neonatal diabetes mellitus	AR	General Population Caucasian / European Population	1 in 423 1 in 232	99% 99%	1 in 42,201 1 in 23,101	<1 in 10 million <1 in 10 million
<i>KDM5C</i>	X-linked intellectual disability, KDM5C-related	XL	General Population	<1 in 50,000	98%	1 in 2,499,951	<1 in 10 million
<i>L1CAM</i>	L1 syndrome	XL	General Population	1 in 15,000	99%	1 in 1,499,901	1 in 5,999,804
<i>LAMA2</i>	Muscular dystrophy, LAMA2-related	AR	General Population Caucasian / European Population	<1 in 500 1 in 125	99% 99%	1 in 49,901 1 in 12,401	<1 in 10 million 1 in 6,200,500
<i>LAMA3</i>	Junctional epidermolysis bullosa, LAMA3-related	AR	General Population	1 in 781	98%	1 in 39,001	<1 in 10 million
<i>LAMA3</i>	Laryngo-onycho-cutaneous syndrome	AR	General Population	1 in 781	98%	1 in 39,001	<1 in 10 million
<i>LAMB3</i>	Junctional epidermolysis bullosa, LAMB3-related	AR	General Population	1 in 781	98%	1 in 39,001	<1 in 10 million
<i>LAMC2</i>	Junctional epidermolysis bullosa, LAMC2-related	AR	General Population	1 in 781	98%	1 in 39,001	<1 in 10 million
<i>LCA5</i>	Leber congenital amaurosis 5	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
<i>LDLRAP1</i>	Familial Hypercholesterolemia	AR	General Population Amish Population Caucasian / European Population French Canadian Population	1 in 8 1 in 2 1 in 7 1 in 8	99% 99% 99% 99%	1 in 701 1 in 101 1 in 601 1 in 701	1 in 22,432 1 in 808 1 in 16,828 1 in 22,432
<i>LHX3</i>	Combined pituitary hormone deficiency 3	AR	General Population	1 in 45	98%	1 in 2,201	1 in 396,180
<i>LIFR</i>	Stuve-Wiedemann syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>LIPA</i>	Lysosomal acid lipase deficiency	AR	General Population Caucasian / European Population Iranian Jewish Population	<1 in 500 1 in 112 1 in 26	99% 99% 99%	1 in 49,901 1 in 11,101 1 in 2,501	<1 in 10 million 1 in 4,973,248 1 in 260,104
<i>LMBRD1</i>	Methylmalonic aciduria and homocystinuria, cbIF type	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>LOXHD1</i>	Nonsyndromic hearing loss 77	AR	General Population Ashkenazi Jewish Population	1 in 500 1 in 180	98% 98%	1 in 24,951 1 in 8,951	<1 in 10 million 1 in 6,444,720
<i>LPL</i>	Familial lipoprotein lipase deficiency	AR	General Population French Canadian Population	1 in 500 1 in 46	99% 99%	1 in 49,901 1 in 4,501	<1 in 10 million 1 in 828,184
<i>LRP2</i>	Donnai-Barrow syndrome	AR	General Population	1 in 214	99%	1 in 10,651	1 in 9,117,256
<i>LRPPRC</i>	Leigh syndrome with Complex IV deficiency	AR	General Population Faroese Population French Canadian Population	1 in 447 1 in 21 1 in 22	98% 98% 98%	1 in 22,301 1 in 1,001 1 in 1,051	<1 in 10 million 1 in 84,084 1 in 92,488
<i>LYST</i>	Chediak-Higashi syndrome	AR	General Population	<1 in 500	90%	1 in 4,991	1 in 9,982,000
<i>MAN2B1</i>	Alpha-Mannosidosis	AR	General Population Caucasian / European Population	1 in 354 1 in 274	99% 99%	1 in 35,301 1 in 27,301	<1 in 10 million <1 in 10 million
<i>MCCC1</i>	3-Methylcrotonyl-CoA carboxylase 1 deficiency (3-MCC deficiency)	AR	General Population	1 in 95	98%	1 in 4,701	1 in 1,786,380
<i>MCCC2</i>	3-Methylcrotonyl-CoA carboxylase 2 deficiency (3-MCC deficiency)	AR	General Population	1 in 95	98%	1 in 4,701	1 in 1,786,380
<i>MCEE</i>	Methylmalonyl-CoA epimerase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>MCOLN1</i>	Mucopolipidosis IV	AR	General Population Ashkenazi Jewish Population	1 in 300 1 in 100	99% 99%	1 in 29,901 1 in 9,901	<1 in 10 million 1 in 3,960,400



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<i>MCPH1</i>	Primary microcephaly 1, recessive	AR	General Population	1 in 147	99%	1 in 14,601	1 in 8,585,388
<i>MED17</i>	Postnatal Progressive Microcephaly with Seizures and Brain Atrophy	AR	General Population Bukharan/Kurdish Jewish Population	<1 in 500 1 in 20	99% 99%	1 in 49,901 1 in 1,901	<1 in 10 million 1 in 152,080
<i>MEFV</i>	Familial Mediterranean fever	AR	General Population Mediterranean Population	1 in 20 1 in 7	99% 90%	1 in 1,901 1 in 61	1 in 152,080 1 in 1,708
<i>MESP2</i>	Spondylocostal dysostosis	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>MFSD8</i>	Neuronal ceroid lipofuscinosis, MFSD8-related	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
<i>MID1</i>	Opitz GBBB syndrome, type I	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
<i>MKS1</i>	Bardet-Biedl syndrome 13	AR	General Population Finnish Population	1 in 260 1 in 47	98% 98%	1 in 12,951 1 in 2,301	<1 in 10 million 1 in 432,588
<i>MKS1</i>	Joubert syndrome 28	AR	General Population Finnish Population	1 in 260 1 in 47	98% 98%	1 in 12,951 1 in 2,301	<1 in 10 million 1 in 432,588
<i>MKS1</i>	Meckel syndrome 1	AR	General Population Finnish Population	1 in 260 1 in 47	98% 98%	1 in 12,951 1 in 2,301	<1 in 10 million 1 in 432,588
<i>MLC1</i>	Megalencephalic leukoencephalopathy with subcortical cysts	AR	General Population Libyan Jewish Population	<1 in 500 1 in 40	99% 99%	1 in 49,901 1 in 3,901	<1 in 10 million 1 in 624,160
<i>MMAA</i>	Methylmalonic aciduria, cblA type	AR	General Population	1 in 301	97%	1 in 10,001	<1 in 10 million
<i>MMAB</i>	Methylmalonic aciduria, cblB type	AR	General Population	1 in 435	98%	1 in 21,701	<1 in 10 million
<i>MMACHC</i>	Methylmalonic aciduria and homocystinuria, cblC type	AR	General Population	1 in 134	90%	1 in 1,331	1 in 713,416
<i>MMADHC</i>	Methylmalonic aciduria and homocystinuria, cblD type	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>MPI</i>	Congenital disorder of glycosylation type Ib	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>MPL</i>	Congenital amegakaryocytic thrombocytopenia	AR	General Population Ashkenazi Jewish Population	1 in 102 1 in 55	98% 98%	1 in 5,051 1 in 2,701	1 in 2,060,808 1 in 594,220
<i>MPV17</i>	Hepatocerebral mitochondrial DNA depletion syndrome, MPV17-related	AR	General Population Native American Population	<1 in 500 1 in 20	96% 96%	1 in 12,476 1 in 476	<1 in 10 million 1 in 38,080
<i>MTHFR</i>	Homocystinuria, MTHFR-related	AR	General Population	1 in 224	98%	1 in 11,151	1 in 9,991,296
<i>MTM1</i>	Myotubular myopathy, X-linked	XL	General Population	1 in 25,000	98%	1 in 1,249,951	1 in 4,999,904
<i>MTMR2</i>	Charcot-Marie-Tooth disease, type 4B1	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>MTRR</i>	Homocystinuria-megaloblastic anemia, cobalamin E type	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>MTTP</i>	Abetalipoproteinemia	AR	General Population Ashkenazi Jewish Population	<1 in 500 1 in 180	98% 98%	1 in 24,951 1 in 8,951	<1 in 10 million 1 in 6,444,720
<i>MUT</i>	Methylmalonic aciduria-methylmalonyl-CoA mutase deficiency	AR	General Population	1 in 100	99%	1 in 9,901	1 in 3,960,400
<i>MVK</i>	Hyperimmunoglobulinemia D syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>MVK</i>	Mevalonate kinase deficiency	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>MYO7A</i>	Usher syndrome, type 1B	AR	General Population East Asian Population	1 in 206 1 in 62	98% 98%	1 in 10,251 1 in 3,051	1 in 8,446,824 1 in 756,648
<i>MYO7A</i>	Non-syndromic hearing loss, MYO7A-related	AR	General Population East Asian Population	1 in 206 1 in 62	98% 98%	1 in 10,251 1 in 3,051	1 in 8,446,824 1 in 756,648
<i>NAGA</i>	Schindler disease types 1 and 3	AR	General Population	1 in 94	99%	1 in 9,301	1 in 3,497,176
<i>NAGLU</i>	Mucopolysaccharidosis type IIIB (Sanfilippo syndrome B)	AR	General Population Caucasian / European Population East Asian Population	<1 in 500 1 in 346 1 in 298	99% 99% 99%	1 in 49,901 1 in 34,501 1 in 29,701	<1 in 10 million <1 in 10 million <1 in 10 million
<i>NAGS</i>	N-acetylglutamate synthase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>NBN</i>	Nijmegen breakage syndrome	AR	General Population	1 in 158	99%	1 in 15,701	1 in 9,923,032
<i>NDP</i>	Norrie disease	XL	General Population	<1 in 50,000	98%	1 in 2,499,951	<1 in 10 million
<i>NDRG1</i>	Charcot-Marie-Tooth disease, type 4D	AR	General Population	1 in 22	98%	1 in 1,051	1 in 92,488
<i>NDUFAF5</i>	Mitochondrial complex I deficiency (Leigh syndrome)	AR	General Population Ashkenazi Jewish Population	1 in 447 1 in 290	98% 98%	1 in 22,301 1 in 14,451	<1 in 10 million <1 in 10 million
<i>NDUFS6</i>	Mitochondrial complex I deficiency (Leigh syndrome)	AR	General Population Bukharan/Kurdish Jewish Population	<1 in 500 1 in 24	99% 99%	1 in 49,901 1 in 2,301	<1 in 10 million 1 in 220,896
<i>NEB</i>	Nemaline myopathy	AR	General Population Amish Population Ashkenazi Jewish Population Finnish Population	1 in 112 1 in 11 1 in 108 1 in 112	98% 98% 98% 98%	1 in 5,551 1 in 501 1 in 5,351 1 in 5,551	1 in 2,486,848 1 in 22,044 1 in 2,311,632 1 in 2,486,848
<i>NPC1</i>	Niemann-Pick disease, type C1	AR	General Population	1 in 194	90%	1 in 1,931	1 in 1,498,456
<i>NPC2</i>	Niemann-Pick disease, type C2	AR	General Population	1 in 194	99%	1 in 19,301	<1 in 10 million
<i>NPHP1</i>	Joubert syndrome 4	AR	General Population Finnish Population	1 in 480 1 in 124	98% 98%	1 in 23,951 1 in 6,151	<1 in 10 million 1 in 3,050,896



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
NPHP1	Nephronophthisis	AR	General Population	1 in 480	98%	1 in 23,951	<1 in 10 million
			Finnish Population	1 in 124	98%	1 in 6,151	1 in 3,050,896
NPHP1	NPHP1-related disorders	AR	General Population	1 in 480	98%	1 in 23,951	<1 in 10 million
			Finnish Population	1 in 124	98%	1 in 6,151	1 in 3,050,896
NPHP1	Senior-Løken syndrome 1	AR	General Population	1 in 480	98%	1 in 23,951	<1 in 10 million
			Finnish Population	1 in 124	98%	1 in 6,151	1 in 3,050,896
NPHS1	Congenital nephrotic syndrome, type 1	AR	General Population	1 in 289	98%	1 in 14,401	<1 in 10 million
			Finnish Population	1 in 50	98%	1 in 2,451	1 in 490,200
NPHS2	Congenital nephrotic syndrome, type 2	AR	General Population	1 in 289	98%	1 in 14,401	<1 in 10 million
			Finnish Population	1 in 50	98%	1 in 2,451	1 in 490,200
NR0B1	Congenital adrenal hypoplasia, X-linked	XL	General Population	1 in 6,250	99%	1 in 624,901	1 in 2,499,804
NR2E3	Retinitis pigmentosa 37	AR	General Population	1 in 209	98%	1 in 10,401	1 in 8,695,236
NR2E3	Enhanced S-cone syndrome	AR	General Population	1 in 209	98%	1 in 10,401	1 in 8,695,236
NTRK1	Congenital insensitivity to pain with anhidrosis	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
OAT	Gyrate atrophy of choroid and retina	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
OCA2	Oculocutaneous albinism type II	AR	General Population	1 in 76	99%	1 in 7,501	1 in 2,280,304
OCRL	Lowe syndrome	XL	General Population	1 in 250,000	95%	1 in 4,999,981	<1 in 10 million
OCRL	Dent disease 2	XL	General Population	1 in 250,000	95%	1 in 4,999,981	<1 in 10 million
OPA3	Costeff syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Iraqi Jewish Population	1 in 50	98%	1 in 2,451	1 in 490,200
OPHN1	X-linked intellectual disability with cerebellar hypoplasia and distinctive facial appearance	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
OTC	Ornithine transcarbamylase deficiency	XL	General Population	1 in 7,000	90%	1 in 69,991	1 in 279,984
OTOF	Nonsyndromic hearing loss, OTOF-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Spanish Population	1 in 106	99%	1 in 10,501	1 in 4,452,424
P3H1	Osteogenesis imperfecta, type VIII	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			West African Population	1 in 67	99%	1 in 6,601	1 in 1,769,068
			African American Population	1 in 250	99%	1 in 24,901	<1 in 10,000,000
PAH	Phenylalanine Hydroxylase deficiency (Phenylketonuria)	AR	General Population	1 in 93	99%	1 in 9,201	1 in 3,422,772
			Caucasian / European Population	1 in 63	99%	1 in 6,201	1 in 1,562,652
			Middle-Eastern Population	1 in 74	99%	1 in 7,301	1 in 2,161,096
			South East Asian	1 in 59	99%	1 in 5,801	1 in 1,369,036
PAK3	X-linked intellectual disability, PAK3-related	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
PANK2	Pantothenate kinase-associated neurodegeneration	AR	General Population	1 in 289	99%	1 in 28,801	<1 in 10 million
PC	Pyruvate carboxylase deficiency	AR	General Population	1 in 250	95%	1 in 4,981	1 in 4,981,000
PCBD1	Tetrahydrobiopterin deficiency, PCBD1-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PCCA	Propionic acidemia, PCCA-related	AR	General Population	1 in 224	96%	1 in 5,576	1 in 4,996,096
			Native American Population	1 in 85	96%	1 in 2,101	1 in 714,340
PCCB	Propionic acidemia, PCCB-related	AR	General Population	1 in 224	99%	1 in 22,301	<1 in 10 million
			Native American Population	1 in 85	99%	1 in 8,401	1 in 2,856,340
PCDH15	Non-syndromic hearing loss, PCDH15-related	AR	General Population	1 in 395	98%	1 in 19,701	1 in 78,804
			Ashkenazi Jewish Population	1 in 72	98%	1 in 3,551	1 in 14,204
PCDH15	Usher syndrome, type 1F	AR	General Population	1 in 395	98%	1 in 19,701	1 in 78,804
			Ashkenazi Jewish Population	1 in 72	98%	1 in 3,551	1 in 14,204
PDE6A	Retinitis pigmentosa, PDE6A-related	AR	General Population	1 in 133	99%	1 in 13,201	1 in 7,022,932
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency	XL	General Population	<1 in 250,000	98%	1 in 12,499,951	<1 in 10 million
PDHB	Pyruvate dehydrogenase E1-beta deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
PEX1	Zellweger syndrome, PEX1-related	AR	General Population	1 in 147	95%	1 in 2,921	1 in 1,717,548
PEX10	Zellweger syndrome, PEX10-related	AR	General Population	1 in 500	95%	1 in 9,981	<1 in 10 million
			Japanese Population	1 in 354	95%	1 in 7,061	1 in 9,998,376
PEX12	Zellweger syndrome, PEX12-related	AR	General Population	1 in 373	95%	1 in 7,441	<1 in 10 million
PEX2	Zellweger syndrome, PEX2-related	AR	General Population	1 in 500	95%	1 in 9,981	<1 in 10 million
			Ashkenazi Jewish Population	1 in 123	95%	1 in 2,441	1 in 1,200,972
PEX6	Zellweger syndrome, PEX6-related	AR	General Population	1 in 280	99%	1 in 27,901	<1 in 10 million
			Yemenite Jewish Population	1 in 18	99%	1 in 1,701	1 in 122,472
PEX7	Rhizomelic chondrodysplasia punctata, type 1	AR	General Population	1 in 158	99%	1 in 15,701	1 in 9,923,032
PFKM	Glycogen storage disease VII	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Ashkenazi Jewish Population	1 in 120	99%	1 in 11,901	1 in 5,712,480
PGK1	Phosphoglycerate kinase 1 deficiency	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
PHF8	X-linked intellectual disability, Siderius type	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
PHGDH	Phosphoglycerate dehydrogenase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Ashkenazi Jewish Population	1 in 280	98%	1 in 13,951	<1 in 10 million
PKHD1	Polycystic kidney disease, PKHD1-related	AR	General Population	1 in 70	98%	1 in 3,451	1 in 966,280
			Ashkenazi Jewish Population	1 in 107	98%	1 in 5,301	1 in 2,268,828
PLA2G6	Infantile neuroaxonal dystrophy	AR	General Population	1 in 500	97%	1 in 16,634	<1 in 10 million
PLOD1	Ehlers-Danlos syndrome with kyphoscoliosis, PLOD1-related	AR	General Population	1 in 159	99%	1 in 15,801	<1 in 10 million
PLP1	Spastic paraplegia type 2	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
PLP1	Pelizaeus-Merzbacher disease	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
PMM2	Congenital disorder of glycosylation type 1a	AR	General Population	1 in 63	99%	1 in 6,201	1 in 1,562,652
			Ashkenazi Jewish Population	1 in 57	99%	1 in 5,601	1 in 1,277,028
			Caucasian / European Population	1 in 71	99%	1 in 7,001	1 in 1,988,284
POLG	Ataxia neuropathy spectrum	AR	General Population	1 in 113	95%	1 in 2,241	1 in 1,012,932
POLG	Progressive external ophthalmoplegia	AR	General Population	1 in 113	95%	1 in 2,241	1 in 1,012,932
POLG	Mycocerebrohepatopathy syndrome	AR	General Population	1 in 113	95%	1 in 2,241	1 in 1,012,932
POLG	POLG-related disorders	AR	General Population	1 in 113	95%	1 in 2,241	1 in 1,012,932
POLG	Alpers-Huttenlocher syndrome	AR	General Population	1 in 113	95%	1 in 2,241	1 in 1,012,932
POLR1C	Hypomyelinating Leukodystrophy, POLR1C-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
POLR1C	Treacher Collins syndrome, POLR1C-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
POMGNT1	Muscular dystrophy-dystroglycanopathy	AR	General Population	1 in 462	98%	1 in 23,051	<1 in 10 million
			Finnish Population	1 in 111	98%	1 in 5,501	1 in 2,442,444
POMGNT1	Retinitis pigmentosa 76	AR	General Population	1 in 462	98%	1 in 23,051	<1 in 10 million
POMGNT1	Walker-Warburg syndrome	AR	General Population	1 in 111	98%	1 in 5,501	1 in 2,442,444
POMGNT1	Walker-Warburg syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
POMT1	Muscular dystrophy-dystroglycanopathy, POMT1-related	AR	General Population	1 in 290	99%	1 in 28,901	<1 in 10 million
POMT1	Walker-Warburg syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
POMT2	Muscular dystrophy-dystroglycanopathy, POMT2-related	AR	General Population	1 in 371	99%	1 in 37,001	<1 in 10 million
POMT2	Walker-Warburg syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
POU3F4	X-linked hearing loss, POU3F4-related	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
PPT1	Neuronal ceroid lipofuscinosis, PPT1-related	AR	General Population	1 in 368	98%	1 in 18,351	<1 in 10 million
			Caucasian / European Population	1 in 488	98%	1 in 24,351	<1 in 10 million
			Finnish Population	1 in 75	98%	1 in 3,701	1 in 1,110,300
PQBP1	Renpenning syndrome	XL	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PRF1	Hemophagocytic lymphohistiocytosis, familial, 2	AR	General Population	1 in 149	99%	1 in 14,801	1 in 8,821,396
PROP1	Combined pituitary hormone deficiency 2	AR	General Population	1 in 45	98%	1 in 2,201	1 in 396,180
PRPS1	Rosenberg-Chutorian syndrome	XL	General Population	<1 in 250,000	98%	1 in 12,499,951	<1 in 10 million
PRPS1	Arts syndrome	XL	General Population	<1 in 250,000	98%	1 in 12,499,951	<1 in 10 million
PRPS1	Non-syndromic hearing loss, PRPS1-related	XL	General Population	<1 in 250,000	98%	1 in 12,499,951	<1 in 10 million
PRPS1	Phosphoribosylpyrophosphate synthetase superactivity	XL	General Population	<1 in 250,000	98%	1 in 12,499,951	<1 in 10 million
PSAP	Metachromatic leukodystrophy due to saposin-b deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
PTS	Tetrahydrobiopterin deficiency	AR	General Population	1 in 354	96%	1 in 8,826	<1 in 10 million
PUS1	Mitochondrial myopathy and sideroblastic anemia 1	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
PYGM	Glycogen storage disease type V	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Caucasian / European Population	1 in 206	99%	1 in 20,501	<1 in 10 million
QDPR	Tetrahydrobiopterin deficiency, QDPR-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
RAB23	Carpenter syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
RAG1	Omenn syndrome, RAG1-related	AR	General Population	1 in 290	98%	1 in 14,451	1 in 16,763,160
RAG2	Omenn syndrome, RAG2-related	AR	General Population	1 in 137	98%	1 in 6,801	1 in 3,726,948
RAPSN	Congenital myasthenic syndrome, RAPSN-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
RAPSN	Fetal akinesia deformation sequence	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
RARS2	Pontocerebellar hypoplasia type 6	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
RAX	Microphthalmia, isolated 3	AR	General Population	1 in 289	99%	1 in 28,801	<1 in 10 million



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<i>RDH12</i>	Leber congenital amaurosis type 13	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Caucasian / European Population	1 in 456	98%	1 in 22,751	<1 in 10 million
<i>RMRP</i>	Metaphyseal dysplasia without hypotrichosis	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Amish Population	1 in 16	99%	1 in 1,501	1 in 96,064
			Finnish Population	1 in 76	99%	1 in 7,501	1 in 2,280,304
<i>RMRP</i>	Cartilage-Hair Hypoplasia Anauxetic Dysplasia Spectrum Disorder	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Amish Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Finnish Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>RMRP</i>	Anauxetic dysplasia	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Amish Population	1 in 16	99%	1 in 1,501	1 in 96,064
			Finnish Population	1 in 76	99%	1 in 7,501	1 in 2,280,304
<i>RMRP</i>	Cartilage-hair hypoplasia	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Amish Population	1 in 16	99%	1 in 1,501	1 in 96,064
			Finnish Population	1 in 76	99%	1 in 7,501	1 in 2,280,304
<i>RNASEH2B</i>	Aicardi Goutieres syndrome 2	AR	General Population	1 in 217	99%	1 in 10,801	1 in 9,375,268
<i>RP2</i>	X-linked Retinitis pigmentosa, RP2-related	XL	General Population	1 in 4,000	99%	1 in 399,901	1 in 1,600,000
<i>RPE65</i>	Retinitis pigmentosa 20	AR	General Population	1 in 228	98%	1 in 11,351	<1 in 10 million
<i>RPE65</i>	Leber congenital amaurosis 2	AR	General Population	1 in 228	98%	1 in 11,351	<1 in 10 million
<i>RPGR</i>	X-linked Retinitis pigmentosa, RPGR-related	XL	General Population	1 in 3,000	75%	1 in 11,997	1 in 48,000
<i>RPGRIP1L</i>	COACH syndrome	AR	General Population	1 in 259	98%	1 in 12,901	<1 in 10 million
<i>RPGRIP1L</i>	Joubert syndrome 7	AR	General Population	1 in 259	98%	1 in 12,901	<1 in 10 million
<i>RPGRIP1L</i>	Meckel syndrome 5	AR	General Population	1 in 259	98%	1 in 12,901	<1 in 10 million
<i>RS1</i>	Juvenile retinoschisis, X-linked	XL	General Population	1 in 2,500	96%	1 in 62,476	1 in 249,956
<i>RTEL1</i>	Dyskeratosis congenita type 5	AR	General Population	1 in 500	99%	1 in 49,901	<1 in 10 million
			Ashkenazi Jewish Population	1 in 203	99%	1 in 20,201	<1 in 10 million
<i>SACS</i>	Autosomal recessive spastic ataxia of Charlevoix-Saguenay	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
			French Canadian Population	1 in 19	95%	1 in 361	1 in 27,436
<i>SAMHD1</i>	Aicardi-Goutieres syndrome	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
<i>SCO2</i>	Mitochondrial complex IV deficiency	AR	General Population	1 in 150	99%	1 in 14,901	1 in 8,940,600
<i>SEPSECS</i>	Pontocerebellar hypoplasia type 2D	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Moroccan/Iraqi Jewish Population	1 in 44	99%	1 in 4,301	1 in 756,976
<i>SERPINA1</i>	Alpha-1 antitrypsin deficiency	AR	General Population	1 in 33	95%	1 in 641	1 in 84,612
			Caucasian / European Population	1 in 19	95%	1 in 361	1 in 27,436
<i>SGCA</i>	Limb-girdle muscular dystrophy, type 2D	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Caucasian / European Population	1 in 288	98%	1 in 14,351	<1 in 10 million
			Finnish Population	1 in 150	98%	1 in 7,451	1 in 4,470,600
<i>SGCB</i>	Limb-girdle muscular dystrophy, type 2E	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
			Caucasian / European Population	1 in 406	98%	1 in 20,251	<1 in 10 million
<i>SGCD</i>	Limb-girdle muscular dystrophy, type 2F	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>SGCG</i>	Limb-girdle muscular dystrophy, type 2C	AR	General Population	1 in 381	98%	1 in 19,001	<1 in 10 million
			Moroccan Population	1 in 250	98%	1 in 12,451	<1 in 10 million
			Roma / Gypsy Population	1 in 96	98%	1 in 4,751	1 in 1,824,384
<i>SGSH</i>	Mucopolysaccharidosis IIIA (Sanfilippo syndrome A)	AR	General Population	1 in 454	98%	1 in 22,651	<1 in 10 million
			Caucasian / European Population	1 in 253	98%	1 in 12,601	<1 in 10 million
<i>SH3TC2</i>	Charcot-Marie-Tooth disease, SH3TC2-related	AR	General Population	1 in 69	99%	1 in 6,801	1 in 1,877,076
<i>SLC12A3</i>	Gitelman syndrome	AR	General Population	1 in 100	98%	1 in 4,951	1 in 1,980,400
<i>SLC12A6</i>	Andermann syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			French Canadian Population	1 in 23	99%	1 in 2,201	1 in 202,492
<i>SLC16A2</i>	Allan-Herndon-Dudley syndrome	XL	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
<i>SLC17A5</i>	Sialic acid storage disorder	AR	General Population	<1 in 500	91%	1 in 5,545	<1 in 10 million
			Finnish Population	1 in 100	91%	1 in 1,101	1 in 440,400
<i>SLC19A3</i>	Biotin-responsive basal ganglia disease	AR	General Population	1 in 109	99%	1 in 5,401	1 in 2,354,836
<i>SLC22A5</i>	Systemic primary carnitine deficiency	AR	General Population	1 in 129	99%	1 in 12,801	1 in 6,605,316
			African/African American Population	1 in 86	99%	1 in 8,501	1 in 2,924,344
			East Asian Population	1 in 77	99%	1 in 7,601	1 in 2,341,108
			Faroese Population	1 in 9	99%	1 in 801	1 in 28,836
			Pacific Islander Population	1 in 37	99%	1 in 3,601	1 in 532,948
			South Asian/Indian Population	1 in 51	99%	1 in 5,001	1 in 1,020,204
<i>SLC25A13</i>	Citrin deficiency	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
			East Asian Population	1 in 65	95%	1 in 1,281	1 in 333,060



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome (Triple H syndrome)	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			French Canadian Population	1 in 37	99%	1 in 3,601	1 in 532,948
SLC25A20	Carnitine-acylcarnitine translocase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
SLC26A2	Diastrophic dysplasia	AR	General Population	1 in 158	90%	1 in 1,571	1 in 992,872
			Finnish Population	1 in 50	90%	1 in 491	1 in 98,200
SLC26A2	Achondrogenesis, type IB	AR	General Population	1 in 158	90%	1 in 1,571	1 in 992,872
			Finnish Population	1 in 50	90%	1 in 491	1 in 98,200
SLC26A2	Multiple epiphyseal dysplasia	AR	General Population	1 in 158	90%	1 in 1,571	1 in 992,872
			Finnish Population	1 in 50	90%	1 in 491	1 in 98,200
SLC26A2	Atelosteogenesis II	AR	General Population	1 in 158	90%	1 in 1,571	1 in 992,872
			Finnish Population	1 in 50	90%	1 in 491	1 in 98,200
SLC26A3	Congenital secretory chloride diarrhea	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Middle-Eastern Population	1 in 57	98%	1 in 2,801	1 in 638,628
SLC26A4	Pendred syndrome	AR	General Population	1 in 80	98%	1 in 3,951	1 in 1,264,320
			African/African American Population	1 in 76	98%	1 in 3,751	1 in 1,140,304
			Caucasian / European Population	1 in 88	98%	1 in 4,351	1 in 1,531,552
			East Asian Population	1 in 74	98%	1 in 3,651	1 in 1,080,696
SLC35A3	Arthrogyriposis, intellectual disability, and seizures	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Ashkenazi Jewish Population	1 in 453	98%	1 in 22,601	<1 in 10 million
SLC37A4	Glycogen storage disease, type Ib	AR	General Population	1 in 158	95%	1 in 3,141	1 in 1,985,112
			Ashkenazi Jewish Population	1 in 71	95%	1 in 1,401	1 in 397,884
SLC39A4	Acrodermatitis enteropathica	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
SLC46A1	Hereditary folate malabsorption	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Puerto Rican Population	1 in 500	99%	1 in 49,901	<1 in 10 million
SLC4A11	Corneal endothelial dystrophy	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
SLC5A5	Thyroid dysmorphogenesis, SLC5A5-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SLC6A19	Hartnup disorder	AR	General Population	1 in 87	99%	1 in 8,601	1 in 2,993,148
SLC6A8	Creatine deficiency syndrome	XL	General Population	1 in 3,434	98%	1 in 171,651	1 in 686,716
SLC7A7	Lysinuric protein intolerance	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
			Finnish Population	1 in 122	95%	1 in 2,421	1 in 1,181,448
			Japanese Population	1 in 119	95%	1 in 2,361	1 in 1,123,836
SMARCAL1	Schimke immunosseous dysplasia	AR	General Population	1 in 500	90%	1 in 4,991	1 in 9,982,000
SMN1	Spinal muscular atrophy	AR	General Population	1 in 54	91%	1 in 590	1 in 127,440
			African/African American Population	1 in 72	71%	1 in 246	1 in 70,848
			Ashkenazi Jewish Population	1 in 67	91%	1 in 734	1 in 196,712
			Caucasian / European Population	1 in 47	95%	1 in 921	1 in 173,148
			East Asian Population	1 in 59	93%	1 in 830	1 in 195,880
			Latino Population	1 in 68	90%	1 in 671	1 in 182,512
			Sephardic Jewish Population	1 in 34	96%	1 in 826	1 in 112,336
SMN1	Spinal muscular atrophy silent carrier	AR	General Population	1 in 54	91%	1 in 590	1 in 127,440
SMPD1	Niemann-Pick disease, type A/B	AR	General Population	1 in 250	95%	1 in 4,981	1 in 4,981,000
			Ashkenazi Jewish Population	1 in 115	95%	1 in 2,281	1 in 1,049,260
			Latino Population	1 in 106	95%	1 in 2,101	1 in 890,824
SPG11	SPG11-related Neuromuscular Disorders	AR	General Population	1 in 159	99%	1 in 15,801	<1 in 10 million
SPG7	Spastic paraplegia type 7	AR	General Population	1 in 159	99%	1 in 15,801	<1 in 10 million
STAR	Lipoid congenital adrenal hyperplasia	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
SUMF1	Multiple sulfatase deficiency	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
			Ashkenazi Jewish Population	1 in 320	98%	1 in 15,951	<1 in 10 million
SURF1	Charcot-Marie-Tooth disease, SURF1-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SURF1	Leigh syndrome, SURF1-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
SYN1	X-linked epilepsy with variable learning disabilities	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
TAT	Tyrosinemia, type II	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
TCIRG1	Osteopetrosis 1	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
TECPR2	Spastic paraplegia 49	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
TF	Atransferrinemia	AR	General Population	1 in 116	99%	1 in 11,501	1 in 5,336,464
TFR2	Hemochromatosis, type 3	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
TG	Thyroid dysmorphogenesis, TG-related	AR	General Population	1 in 241	99%	1 in 24,001	<1 in 10 million
TGM1	Congenital ichthyosis	AR	General Population	1 in 224	95%	1 in 4,461	1 in 3,997,056
TH	Segawa syndrome	AR	General Population	1 in 224	98%	1 in 11,151	1 in 9,991,296
THOC2	X-linked Intellectual disability, THOC2-related	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
TMEM216	Joubert syndrome 2	AR	General Population	1 in 141	98%	1 in 7,001	1 in 3,948,564
			Ashkenazi Jewish Population	1 in 92	98%	1 in 4,551	1 in 1,674,768
TMEM216	Meckel syndrome 2	AR	General Population	1 in 141	98%	1 in 7,001	1 in 3,948,564
			Ashkenazi Jewish Population	1 in 92	98%	1 in 4,551	1 in 1,674,768
TNXB	Ehlers–Danlos-like syndrome due to tenascin-X deficiency	AR	General Population	1 in 28	99%	1 in 2,701	1 in 302,512
TPO	Thyroid dysmorphogenesis, TPO-related	AR	General Population	1 in 373	99%	1 in 37,201	<1 in 10 million
TPP1	Neuronal ceroid lipofuscinosis, TPP1-related	AR	General Population	1 in 252	97%	1 in 8,368	1 in 8,434,944
			French Canadian Population	1 in 53	97%	1 in 1,734	1 in 367,608
TRDN	Catecholaminergic polymorphic ventricular tachycardia	AR	General Population	1 in 354	98%	1 in 17,651	<1 in 10 million
TRIM32	Limb-girdle muscular dystrophy, type 2H	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Hutterite Population	1 in 12	98%	1 in 551	1 in 26,448
TRIM32	Bardet-Biedl syndrome 11	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Hutterite Population	1 in 12	98%	1 in 551	1 in 26,448
TRMU	Liver failure, acute infantile	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Yemeni Jewish Population	1 in 34	98%	1 in 1,651	1 in 224,536
TSFM	Combined oxidative phosphorylation deficiency, TSFM-related	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Finnish Population	1 in 80	98%	1 in 3,951	1 in 1,264,320
TSHB	Congenital hypothyroidism, TSHB-related	AR	General Population	1 in 500	99%	1 in 49,901	<1 in 10 million
TTC37	Trichohepatoenteric syndrome	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
TPPA	Ataxia with isolated vitamin E deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Caucasian / European Population	1 in 267	90%	1 in 2,661	1 in 2,841,948
TYMP	Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
TYR	Oculocutaneous albinism types 1A and 1B	AR	General Population	1 in 20	99%	1 in 1,901	1 in 152,080
UGT1A1	Crigler-Najjar syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
UPF3B	Lujan-Fryns syndrome, UPF3B-related	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
USH1C	Usher syndrome, type IC	AR	General Population	1 in 353	90%	1 in 3,521	1 in 4,971,652
			French Canadian Population	1 in 227	90%	1 in 2,261	1 in 2,052,988
USH1C	Non-syndromic hearing loss, USH1C-related	AR	General Population	1 in 353	90%	1 in 3,521	1 in 4,971,652
			French Canadian Population	1 in 227	90%	1 in 2,261	1 in 2,052,988
USH1G	Usher syndrome type IG	AR	General Population	1 in 434	99%	1 in 43,301	<1 in 10 million
USH2A	Usher syndrome, type 2A	AR	General Population	1 in 126	96%	1 in 3,126	1 in 1,575,504
			Caucasian / European Population	1 in 73	96%	1 in 1,801	1 in 525,892
			Ashkenazi Jewish Population	1 in 35	99%	1 in 3,401	1 in 476,140
			Iranian Jewish Population	1 in 60	99%	1 in 5,901	1 in 1,416,240
VPS13A	Choreoacanthocytosis	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
VPS13B	Cohen syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
VPS45	Severe congenital neutropenia, VPS45-related	AR	General Population	1 in 224	98%	1 in 11,151	1 in 9,991,296
VRK1	Pontocerebellar hypoplasia type 1A	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
VSX2	Microphthalmia with or without coloboma	AR	General Population	1 in 91	98%	1 in 4,501	1 in 1,638,364
WAS	Thrombocytopenia, X-linked	XL	General Population	1 in 125,000	99%	1 in 12,499,901	<1 in 10 million
WAS	Severe congenital neutropenia, WAS-related	XL	General Population	1 in 125,000	99%	1 in 12,499,901	<1 in 10 million
WAS	Wiskott-Aldrich syndrome	XL	General Population	1 in 125,000	99%	1 in 12,499,901	<1 in 10 million
WHRN	Usher syndrome type 2D	AR	General Population	1 in 282	99%	1 in 28,101	<1 in 10 million
WNT10A	Schopf-Schulz-Passarge syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
WNT10A	Odontonychia dysplasia	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
XPA	Xeroderma pigmentosum, group A	AR	General Population	1 in 500	99%	1 in 49,901	<1 in 10 million
			Japanese Population	1 in 74	99%	1 in 7,301	1 in 2,161,096
XPC	Xeroderma pigmentosum, group C	AR	General Population	1 in 500	99%	1 in 49,901	<1 in 10 million
ZDHHC9	X-linked intellectual disability, ZDHHC9-related	XL	General Population	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
ZFYVE26	Spastic paraplegia 15	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
ZNF711	X-linked intellectual disability, ZNF711-related	XL	General Population	<1 in 50,000	93%	1 in 714,272	1 in 2,857,143

* For genes that have tested negative

† The carrier frequency for heterozygous alpha thalassemia carriers ($\alpha\alpha/\alpha-$) is described in rows marked with a dagger symbol. The carrier frequency for alpha thalassemia trait cis ($\alpha\alpha/-$) is 1 in 1000.

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