Myriad Foresight® Residual Risk Table

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Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
6-pyruvoyl-tetrahydropterin Synthase Deficiency (*PTS*) NM_000317:1-6 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 240 Middle East: 1 in 61 Southeast Asia: 1 in 330 Other Populations: < 1 in 500	Eastern Asia: 99% Middle East: 99% Southeast Asia: 99% Other Populations: 99%	Eastern Asia: < 1 in 24,000 Middle East: < 1 in 6,000 Southeast Asia: < 1 in 33,000 Other Populations: < 1 in 50,000
* **ATP7A-related Disorders (*ATP7A*)** NM_000052:2-23 Inheritance: X-linked Recessive	Eastern Asia: 1 in 100,000 Northwestern Europe: 1 in 64,000 Other Populations: 1 in 38,000	Eastern Asia: 90% Northwestern Europe: 90% Other Populations: 90%	Eastern Asia: < 1 in 1,000,000 Northwestern Europe: 1 in 660,000 Other Populations: 1 in 400,000
Adenosine Deaminase Deficiency (*ADA*) NM_000022:1-12 Inheritance: Autosomal Recessive	Worldwide: 1 in 390	Worldwide: 98%	Worldwide: < 1 in 22,000
Alpha-mannosidosis (*MAN2B1*) NM_000528:1-23 Inheritance: Autosomal Recessive	Worldwide: 1 in 350	Worldwide: 99%	Worldwide: < 1 in 35,000
Alpha-sarcoglycanopathy (*SGCA*) NM_000023:1-9 Inheritance: Autosomal Recessive	Finland: 1 in 150 French Canadian/Cajun: 1 in 23 Northwestern Europe: < 1 in 500 Southern Europe: 1 in 290 Other Populations: 1 in 340	Finland: 99% French Canadian/Cajun: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Finland: < 1 in 15,000 French Canadian/Cajun: < 1 in 2,200 Northwestern Europe: < 1 in 50,000 Southern Europe: < 1 in 29,000 Other Populations: < 1 in 34,000
Alstrom Syndrome (*ALMS1*) NM_015120:1-23 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
Andermann Syndrome (*SLC12A6*) NM_133647:1-25 Inheritance: Autosomal Recessive	French Canadian/Cajun: 1 in 24 Other Populations: < 1 in 500	French Canadian/Cajun: 99% Other Populations: 99%	French Canadian/Cajun: < 1 in 2,300 Other Populations: < 1 in 50,000
Argininemia (*ARG1*) NM_000045:1-8 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 170 Finland: < 1 in 500 Other Populations: 1 in 330	Eastern Asia: 97% Finland: 97% Other Populations: 97%	Eastern Asia: < 1 in 6,300 Finland: < 1 in 18,000 Other Populations: < 1 in 12,000
Argininosuccinic Aciduria (*ASL*) NM_001024943:1-16 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 450 Finland: 1 in 190 Hispanic: 1 in 290	Eastern Asia: 99% Finland: 99% Hispanic: 99%	Eastern Asia: < 1 in 45,000 Finland: < 1 in 19,000 Hispanic: < 1 in 29,000

* For X-linked diseases, female carrier frequencies are presented.
Detection rates are determined by using literature to estimate the fraction of disease alleles, weighted by frequency, that the methodology is unable to detect.
Other Populations denotes default ethnicity grouping based on identical risk and detection rates.

Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
	Northwestern Europe: 1 in 160 Other Populations: 1 in 130	Northwestern Europe: 99% Other Populations: 99%	Northwestern Europe: < 1 in 15,000 Other Populations: < 1 in 13,000
Aspartylglucosaminuria (*AGA*) NM_000027:1-9 Inheritance: Autosomal Recessive	Finland: 1 in 71 Other Populations: < 1 in 500	Finland: 99% Other Populations: 99%	Finland: < 1 in 7,000 Other Populations: < 1 in 50,000
Ataxia with Vitamin E Deficiency (*TTPA*) NM_000370:1-5 Inheritance: Autosomal Recessive	Middle East: 1 in 160 Other Populations: < 1 in 500	Middle East: 99% Other Populations: 99%	Middle East: < 1 in 16,000 Other Populations: < 1 in 50,000
Ataxia-telangiectasia (*ATM*) NM_000051:2-63 Inheritance: Autosomal Recessive	Worldwide: 1 in 160	Worldwide: 96%	Worldwide: < 1 in 4,200
Autoimmune Polyglandular Syndrome Type 1 (*AIRE*) NM_000383:1-14 Inheritance: Autosomal Recessive	Finland: 1 in 80 Northwestern Europe: 1 in 150 Other Populations: 1 in 180	Finland: 99% Northwestern Europe: 99% Other Populations: 99%	Finland: < 1 in 7,900 Northwestern Europe: < 1 in 15,000 Other Populations: < 1 in 18,000
Autosomal Recessive Osteopetrosis Type 1 (*TCIRG1*) NM_006019:2-20 Inheritance: Autosomal Recessive	Middle East: 1 in 110 Other Populations: 1 in 350	Middle East: 96% Other Populations: 96%	Middle East: < 1 in 2,800 Other Populations: < 1 in 8,900
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related (*PKHD1*) NM_138694:2-67 Inheritance: Autosomal Recessive	Finland: 1 in 52 Other Populations: 1 in 82	Finland: 99% Other Populations: 99%	Finland: < 1 in 5,100 Other Populations: < 1 in 8,100
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (*SACS*) NM_014363:2-10 Inheritance: Autosomal Recessive	French Canadian/Cajun: 1 in 22 Other Populations: < 1 in 500	French Canadian/Cajun: 99% Other Populations: 99%	French Canadian/Cajun: < 1 in 1,800 Other Populations: < 1 in 44,000
BCS1L-related Disorders (*BCS1L*) NM_004328:3-9 Inheritance: Autosomal Recessive	Finland: 1 in 120 Other Populations: < 1 in 500	Finland: 99% Other Populations: 99%	Finland: < 1 in 12,000 Other Populations: < 1 in 50,000
Bardet-Biedl Syndrome, BBS1-related (*BBS1*) NM_024649:1-17 Inheritance: Autosomal Recessive	African American: 1 in 410 Eastern Asia: < 1 in 500 Middle East: 1 in 200 Northwestern Europe: 1 in 330 South Asia: < 1 in 500 Southern Europe: < 1 in 500 Other Populations: 1 in 390	African American: 99% Eastern Asia: 99% Middle East: 99% Northwestern Europe: 99% South Asia: 99% Southern Europe: 99% Other Populations: 99%	African American: < 1 in 41,000 Eastern Asia: < 1 in 50,000 Middle East: < 1 in 20,000 Northwestern Europe: < 1 in 32,000 South Asia: < 1 in 50,000 Southern Europe: < 1 in 50,000 Other Populations: < 1 in 39,000
Bardet-Biedl Syndrome, BBS10-related (*BBS10*) NM_024685:1-2 Inheritance: Autosomal Recessive	African American: 1 in 440 Eastern Asia: < 1 in 500 French Canadian/Cajun: 1 in 150 Middle East: 1 in 110	African American: 99% Eastern Asia: 99% French Canadian/Cajun: 99% Middle East: 99%	African American: < 1 in 44,000 Eastern Asia: < 1 in 50,000 French Canadian/Cajun: < 1 in 15,000 Middle East: < 1 in 11,000

* For X-linked diseases, female carrier frequencies are presented.

Detection rates are determined by using literature to estimate the fraction of disease alleles, weighted by frequency, that the methodology is unable to detect.

Other Populations denotes default ethnicity grouping based on identical risk and detection rates.

Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
	Northwestern Europe: 1 in 420 South Asia: < 1 in 500 Southern Europe: < 1 in 500 Other Populations: 1 in 420	Northwestern Europe: 99% South Asia: 99% Southern Europe: 99% Other Populations: 99%	Northwestern Europe: < 1 in 42,000 South Asia: < 1 in 50,000 Southern Europe: < 1 in 50,000 Other Populations: < 1 in 42,000
Bardet-Biedl Syndrome, BBS12-related (*BBS12*) NM_152618:2 Inheritance: Autosomal Recessive	Middle East: 1 in 210 Other Populations: < 1 in 500	Middle East: 99% Other Populations: 99%	Middle East: < 1 in 20,000 Other Populations: < 1 in 50,000
Bardet-Biedl Syndrome, BBS2-related (*BBS2*) NM_031885:1-17 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 140 Middle East: 1 in 180 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Middle East: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 14,000 Middle East: < 1 in 18,000 Other Populations: < 1 in 50,000
Beta-sarcoglycanopathy (*SGCB*) NM_000232:1-6 Inheritance: Autosomal Recessive	Worldwide: 1 in 400	Worldwide: 99%	Worldwide: < 1 in 39,000
Biotinidase Deficiency (*BTD*) NM_000060:1-4 Inheritance: Autosomal Recessive	African American: 1 in 310 Ashkenazi Jewish: 1 in 440 Eastern Asia: 1 in 460 Hispanic: 1 in 160 Northwestern Europe: 1 in 130 Southeast Asia: 1 in 160 Other Populations: 1 in 160	African American: 99% Ashkenazi Jewish: 99% Eastern Asia: 99% Hispanic: 99% Northwestern Europe: 99% Southeast Asia: 99% Other Populations: 99%	African American: < 1 in 38,000 Ashkenazi Jewish: < 1 in 60,000 Eastern Asia: < 1 in 67,000 Hispanic: < 1 in 17,000 Northwestern Europe: < 1 in 13,000 Southeast Asia: < 1 in 18,000 Other Populations: < 1 in 17,000
Bloom Syndrome (*BLM*) NM_000057:2-22 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 110 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 11,000 Other Populations: < 1 in 50,000
CLN3-related Neuronal Ceroid Lipofuscinosis (*CLN3*) NM_001042432:2-16 Inheritance: Autosomal Recessive	Finland: 1 in 71 Northwestern Europe: 1 in 87 Southern Europe: 1 in 280 Other Populations: 1 in 130	Finland: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Finland: < 1 in 7,000 Northwestern Europe: < 1 in 8,600 Southern Europe: < 1 in 28,000 Other Populations: < 1 in 13,000
CLN5-related Neuronal Ceroid Lipofuscinosis (*CLN5*) NM_006493:1-4 Inheritance: Autosomal Recessive	Finland: 1 in 110 Other Populations: < 1 in 500	Finland: 99% Other Populations: 99%	Finland: < 1 in 11,000 Other Populations: < 1 in 50,000
CLN8-related Neuronal Ceroid Lipofuscinosis (*CLN8*) NM_018941:2-3 Inheritance: Autosomal Recessive	Finland: 1 in 140 Other Populations: < 1 in 500	Finland: 99% Other Populations: 99%	Finland: < 1 in 13,000 Other Populations: < 1 in 50,000
COL4A3-related Alport Syndrome (*COL4A3*) NM_000091:1-52 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 190 Finland: 1 in 370 Northwestern Europe: 1 in 210 Southern Europe: 1 in 210 Other Populations: 1 in 350	Ashkenazi Jewish: 94% Finland: 94% Northwestern Europe: 94% Southern Europe: 94% Other Populations: 94%	Ashkenazi Jewish: < 1 in 3,100 Finland: < 1 in 6,000 Northwestern Europe: < 1 in 3,400 Southern Europe: < 1 in 3,400 Other Populations: < 1 in 5,800

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Other Populations denotes default ethnicity grouping based on identical risk and detection rates.

Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
COL4A4-related Alport Syndrome (*COL4A4*) NM_000092:2-48 Inheritance: Autosomal Recessive	Finland: 1 in 370 Southern Europe: 1 in 210 Other Populations: 1 in 350	Finland: 99% Southern Europe: 99% Other Populations: 99%	Finland: < 1 in 36,000 Southern Europe: < 1 in 21,000 Other Populations: < 1 in 35,000
Calpainopathy (*CAPN3*) NM_000070:1-24 Inheritance: Autosomal Recessive	African American: 1 in 97 Ashkenazi Jewish: < 1 in 500 Finland: < 1 in 500 Northwestern Europe: 1 in 160 Southern Europe: 1 in 120 Other Populations: 1 in 140	African American: 99% Ashkenazi Jewish: 99% Finland: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	African American: < 1 in 7,400 Ashkenazi Jewish: < 1 in 38,000 Finland: < 1 in 38,000 Northwestern Europe: < 1 in 13,000 Southern Europe: < 1 in 9,400 Other Populations: < 1 in 11,000
Canavan Disease (*ASPA*) NM_000049:1-6 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 55 Other Populations: 1 in 160	Ashkenazi Jewish: 98% Other Populations: 98%	Ashkenazi Jewish: < 1 in 3,300 Other Populations: < 1 in 9,700
Carbamoylphosphate Synthetase I Deficiency (*CPS1*) NM_001875:1-38 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 450 Finland: 1 in 370 Other Populations: < 1 in 570	Eastern Asia: 99% Finland: 99% Other Populations: 99%	Eastern Asia: < 1 in 45,000 Finland: < 1 in 37,000 Other Populations: < 1 in 57,000
Carnitine Palmitoyltransferase IA Deficiency (*CPT1A*) NM_001876:2-19 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
Carnitine Palmitoyltransferase II Deficiency (*CPT2*) NM_000098:1-5 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 47 Eastern Asia: 1 in 320 Middle East: 1 in 110 Northwestern Europe: 1 in 250 Southern Europe: 1 in 200 Other Populations: 1 in 180	Ashkenazi Jewish: 99% Eastern Asia: 99% Middle East: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 4,600 Eastern Asia: < 1 in 31,000 Middle East: < 1 in 11,000 Northwestern Europe: < 1 in 25,000 Southern Europe: < 1 in 20,000 Other Populations: < 1 in 18,000
Cartilage-hair Hypoplasia (*RMRP*) NR_003051:1 Inheritance: Autosomal Recessive	Finland: 1 in 76 Other Populations: < 1 in 500	Finland: 99% Other Populations: 99%	Finland: < 1 in 7,500 Other Populations: < 1 in 50,000
Cerebrotendinous Xanthomatosis (*CYP27A1*) NM_000784:1-9 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 130 Other Populations: 1 in 110	Eastern Asia: 99% Other Populations: 99%	Eastern Asia: < 1 in 13,000 Other Populations: < 1 in 11,000
Citrullinemia Type 1 (*ASS1*) NM_000050:3-16 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 97 Northwestern Europe: 1 in 140 Other Populations: 1 in 120	Eastern Asia: 86% Northwestern Europe: 99% Other Populations: 99%	Eastern Asia: < 1 in 700 Northwestern Europe: < 1 in 14,000 Other Populations: < 1 in 12,000
Cohen Syndrome (*VPS13B*) NM_017890:2-62 Inheritance: Autosomal Recessive	Finland: 1 in 160 Other Populations: < 1 in 500	Finland: 97% Other Populations: 97%	Finland: < 1 in 4,800 Other Populations: < 1 in 15,000

* For X-linked diseases, female carrier frequencies are presented.

Detection rates are determined by using literature to estimate the fraction of disease alleles, weighted by frequency, that the methodology is unable to detect.

Other Populations denotes default ethnicity grouping based on identical risk and detection rates.

Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
Combined Pituitary Hormone Deficiency, PROP1-related (*PROP1*) NM_006261:1-3 Inheritance: Autosomal Recessive	Worldwide: 1 in 62	Worldwide: 99%	Worldwide: < 1 in 6,100
Congenital Adrenal Hyperplasia, CYP11B1-related (*CYP11B1*) NM_000497:1-9 Inheritance: Autosomal Recessive	Worldwide: 1 in 220	Worldwide: 97%	Worldwide: < 1 in 8,400
Congenital Adrenal Hyperplasia, CYP21A2-related (*CYP21A2*) I173N, V282L, R357W, P31L, c.293-13C>G, G111VfsX21, Q319*, L308FfsX6, CYP21A2 deletion, CYP21A2 duplication, Q319*+CYP21A2dup, [I237N;V238E;M240K], CYP21A2 triplication Inheritance: Autosomal Recessive	African American: 1 in 140 Ashkenazi Jewish: 1 in 62 Eastern Asia: 1 in 67 Finland: 1 in 62 French Canadian/Cajun: 1 in 62 Hispanic: 1 in 62 Middle East: 1 in 43 Native American: 1 in 62 Northwestern Europe: 1 in 55 South Asia: 1 in 62 Southeast Asia: 1 in 50 Southern Europe: 1 in 68	African American: 92% Ashkenazi Jewish: 99% Eastern Asia: 88% Finland: 89% French Canadian/Cajun: 96% Hispanic: 95% Middle East: 97% Native American: 90% Northwestern Europe: 96% South Asia: 89% Southeast Asia: 88% Southern Europe: 96%	African American: < 1 in 1,700 Ashkenazi Jewish: < 1 in 6,100 Eastern Asia: < 1 in 550 Finland: < 1 in 560 French Canadian/Cajun: < 1 in 1,400 Hispanic: < 1 in 1,200 Middle East: < 1 in 1,300 Native American: < 1 in 610 Northwestern Europe: < 1 in 1,300 South Asia: < 1 in 530 Southeast Asia: 1 in 410 Southern Europe: < 1 in 1,600
Congenital Disorder of Glycosylation Type Ia (*PMM2*) NM_000303:1-8 Inheritance: Autosomal Recessive	Worldwide: 1 in 160	Worldwide: 99%	Worldwide: < 1 in 16,000
Congenital Disorder of Glycosylation Type Ic (*ALG6*) NM_013339:2-15 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
Congenital Disorder of Glycosylation, MPI-related (*MPI*) NM_002435:1-8 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
Costeff Optic Atrophy Syndrome (*OPA3*) NM_025136:1-2 Inheritance: Autosomal Recessive	Middle East: 1 in 51 Other Populations: < 1 in 500	Middle East: 99% Other Populations: 99%	Middle East: < 1 in 5,000 Other Populations: < 1 in 50,000
Cystic Fibrosis (*CFTR*) NM_000492:1-27 Inheritance: Autosomal Recessive	African American: 1 in 62 Ashkenazi Jewish: 1 in 24 Eastern Asia: 1 in 91 Finland: 1 in 80 French Canadian/Cajun: 1 in 16 Hispanic: 1 in 53 Middle East: 1 in 30 Native American: 1 in 53 Northwestern Europe: 1 in 31 South Asia: 1 in 100 Southeast Asia: 1 in 91 Southern Europe: 1 in 28	African American: 99% Ashkenazi Jewish: 99% Eastern Asia: 99% Finland: 99% French Canadian/Cajun: 99% Hispanic: 99% Middle East: 99% Native American: 99% Northwestern Europe: 99% South Asia: 99% Southeast Asia: 99% Southern Europe: 99%	African American: < 1 in 6,100 Ashkenazi Jewish: < 1 in 2,300 Eastern Asia: < 1 in 9,000 Finland: < 1 in 7,900 French Canadian/Cajun: < 1 in 1,500 Hispanic: < 1 in 5,200 Middle East: < 1 in 2,900 Native American: < 1 in 5,200 Northwestern Europe: < 1 in 3,000 South Asia: < 1 in 10,000 Southeast Asia: < 1 in 9,000 Southern Europe: < 1 in 2,700

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Detection rates are determined by using literature to estimate the fraction of disease alleles, weighted by frequency, that the methodology is unable to detect.

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Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
Cystinosis (*CTNS*) NM_004937:3-12 Inheritance: Autosomal Recessive	Worldwide: 1 in 220	Worldwide: 99%	Worldwide: < 1 in 22,000
D-bifunctional Protein Deficiency (*HSD17B4*) NM_000414:1-24 Inheritance: Autosomal Recessive	Worldwide: 1 in 160	Worldwide: 98%	Worldwide: < 1 in 9,000
Delta-sarcoglycanopathy (*SGCD*) NM_000337:2-9 Inheritance: Autosomal Recessive	Hispanic: 1 in 320 Middle East: 1 in 350 Other Populations: < 1 in 500	Hispanic: 96% Middle East: 96% Other Populations: 96%	Hispanic: < 1 in 8,400 Middle East: < 1 in 9,300 Other Populations: < 1 in 13,000
Dihydrolipoamide Dehydrogenase Deficiency (*DLD*) NM_000108:1-14 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 94 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 9,300 Other Populations: < 1 in 50,000
Dysferlinopathy (*DYSF*) NM_003494:1-55 Inheritance: Autosomal Recessive	Worldwide: 1 in 190	Worldwide: 98%	Worldwide: < 1 in 11,000
ERCC6-related Disorders (*ERCC6*) NM_000124:2-21 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 370 Northwestern Europe: 1 in 380 Southern Europe: 1 in 380 Other Populations: 1 in 380	Eastern Asia: 96% Northwestern Europe: 96% Southern Europe: 96% Other Populations: 96%	Eastern Asia: < 1 in 8,400 Northwestern Europe: < 1 in 8,500 Southern Europe: < 1 in 8,500 Other Populations: < 1 in 8,400
ERCC8-related Disorders (*ERCC8*) NM_000082:1-12 Inheritance: Autosomal Recessive	Eastern Asia: < 1 in 500 Northwestern Europe: < 1 in 500 Southern Europe: < 1 in 500 Other Populations: 1 in 380	Eastern Asia: 78% Northwestern Europe: 97% Southern Europe: 97% Other Populations: 97%	Eastern Asia: < 1 in 2,300 Northwestern Europe: < 1 in 16,000 Southern Europe: < 1 in 16,000 Other Populations: < 1 in 12,000
EVC-related Ellis-van Creveld Syndrome (*EVC*) NM_153717:1-21 Inheritance: Autosomal Recessive	Worldwide: 1 in 280	Worldwide: 97%	Worldwide: < 1 in 7,800
EVC2-related Ellis-van Creveld Syndrome (*EVC2*) NM_147127:1-22 Inheritance: Autosomal Recessive	Worldwide: 1 in 250	Worldwide: 98%	Worldwide: < 1 in 9,800
FKRP-related Disorders (*FKRP*) NM_024301:4 Inheritance: Autosomal Recessive	African American: 1 in 420 Ashkenazi Jewish: < 1 in 500 Eastern Asia: 1 in 320 Finland: 1 in 180 French Canadian/Cajun: 1 in 220 Hispanic: 1 in 380 Middle East: 1 in 240 Native American: 1 in 220 Northwestern Europe: 1 in 160 South Asia: 1 in 320	African American: 99% Ashkenazi Jewish: 99% Eastern Asia: 99% Finland: 99% French Canadian/Cajun: 99% Hispanic: 99% Middle East: 99% Native American: 99% Northwestern Europe: 99% South Asia: 99%	African American: < 1 in 42,000 Ashkenazi Jewish: < 1 in 50,000 Eastern Asia: < 1 in 32,000 Finland: < 1 in 18,000 French Canadian/Cajun: < 1 in 21,000 Hispanic: < 1 in 38,000 Middle East: < 1 in 24,000 Native American: < 1 in 21,000 Northwestern Europe: < 1 in 16,000 South Asia: < 1 in 32,000

* For X-linked diseases, female carrier frequencies are presented.

Detection rates are determined by using literature to estimate the fraction of disease alleles, weighted by frequency, that the methodology is unable to detect.

Other Populations denotes default ethnicity grouping based on identical risk and detection rates.

Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
	Southeast Asia: 1 in 320 Southern Europe: 1 in 280	Southeast Asia: 99% Southern Europe: 99%	Southeast Asia: < 1 in 32,000 Southern Europe: < 1 in 28,000
FKTN-related Disorders (*FKTN*) NM_001079802:3-11 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 64 Eastern Asia: 1 in 95 Middle East: 1 in 460 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Eastern Asia: 10% Middle East: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 6,300 Eastern Asia: 1 in 110 Middle East: < 1 in 46,000 Other Populations: < 1 in 50,000
* **Fabry Disease (*GLA*)** NM_000169:1-7 Inheritance: X-linked Recessive	Northwestern Europe: 1 in 55,000 Other Populations: 1 in 20,000	Northwestern Europe: 99% Other Populations: 99%	Northwestern Europe: < 1 in 1,000,000 Other Populations: < 1 in 1,000,000
Familial Dysautonomia (*ELP1*) NM_003640:2-37 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 31 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 3,000 Other Populations: < 1 in 50,000
Familial Hyperinsulinism, ABCC8-related (*ABCC8*) NM_000352:1-39 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 45 Eastern Asia: 1 in 140 Finland: 1 in 150 Middle East: 1 in 67 Northwestern Europe: 1 in 170 Other Populations: 1 in 170	Ashkenazi Jewish: 99% Eastern Asia: 99% Finland: 99% Middle East: 99% Northwestern Europe: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 4,400 Eastern Asia: < 1 in 14,000 Finland: < 1 in 15,000 Middle East: < 1 in 6,600 Northwestern Europe: < 1 in 17,000 Other Populations: < 1 in 17,000
Familial Hyperinsulinism, KCNJ11-related (*KCNJ11*) NM_000525:1 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 230 Eastern Asia: 1 in 420 Finland: 1 in 450 Middle East: 1 in 410 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Eastern Asia: 99% Finland: 99% Middle East: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 23,000 Eastern Asia: < 1 in 42,000 Finland: < 1 in 45,000 Middle East: < 1 in 40,000 Other Populations: < 1 in 50,000
Familial Mediterranean Fever (*MEFV*) NM_000243:1-10 Inheritance: Autosomal Recessive	African American: 1 in 6 Ashkenazi Jewish: 1 in 98 Eastern Asia: < 1 in 500 Middle East: 1 in 6 Northwestern Europe: 1 in 110 South Asia: < 1 in 500 Southeast Asia: < 1 in 500 Southern Europe: 1 in 24 Other Populations: 1 in 29	African American: 99% Ashkenazi Jewish: 99% Eastern Asia: 99% Middle East: 99% Northwestern Europe: 99% South Asia: 99% Southeast Asia: 99% Southern Europe: 99% Other Populations: 99%	African American: < 1 in 530 Ashkenazi Jewish: < 1 in 9,700 Eastern Asia: < 1 in 50,000 Middle East: 1 in 460 Northwestern Europe: < 1 in 11,000 South Asia: < 1 in 50,000 Southeast Asia: < 1 in 50,000 Southern Europe: < 1 in 2,300 Other Populations: < 1 in 2,800
Fanconi Anemia Complementation Group A (*FANCA*) NM_000135:1-43 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 260 Hispanic: 1 in 250 Northwestern Europe: 1 in 240 Southern Europe: 1 in 240 Other Populations: 1 in 260	Eastern Asia: 92% Hispanic: 92% Northwestern Europe: 92% Southern Europe: 92% Other Populations: 92%	Eastern Asia: < 1 in 3,100 Hispanic: < 1 in 2,900 Northwestern Europe: < 1 in 2,800 Southern Europe: < 1 in 2,800 Other Populations: < 1 in 3,100

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Detection rates are determined by using literature to estimate the fraction of disease alleles, weighted by frequency, that the methodology is unable to detect.

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Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
Fanconi Anemia, FANCC-related (*FANCC*) NM_000136:2-15 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 94 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 9,300 Other Populations: < 1 in 50,000
* **Fragile X Syndrome (*FMR1*)** FMR1 CGG repeat number Inheritance: X-linked	Not Calculated	Worldwide: 99%	Not Calculated
Free Sialic Acid Storage Disorders (*SLC17A5*) NM_012434:1-11 Inheritance: Autosomal Recessive	Finland: 1 in 51 Other Populations: < 1 in 500	Finland: 99% Other Populations: 98%	Finland: < 1 in 5,000 Other Populations: < 1 in 30,000
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness (*GJB2*) NM_004004:1-2 Inheritance: Autosomal Recessive	African American: 1 in 42 Ashkenazi Jewish: 1 in 21 Eastern Asia: 1 in 34 Hispanic: 1 in 33 Middle East: 1 in 33 Native American: 1 in 33 South Asia: 1 in 41 Southeast Asia: 1 in 34 Other Populations: 1 in 26	African American: 99% Ashkenazi Jewish: 99% Eastern Asia: 99% Hispanic: 99% Middle East: 99% Native American: 99% South Asia: 99% Southeast Asia: 99% Other Populations: 99%	African American: < 1 in 4,100 Ashkenazi Jewish: < 1 in 2,000 Eastern Asia: < 1 in 3,300 Hispanic: < 1 in 3,200 Middle East: < 1 in 3,200 Native American: < 1 in 3,200 South Asia: < 1 in 4,000 Southeast Asia: < 1 in 3,300 Other Populations: < 1 in 2,500
GLB1-related Disorders (*GLB1*) NM_000404:1-16 Inheritance: Autosomal Recessive	Worldwide: 1 in 170	Worldwide: 99%	Worldwide: < 1 in 17,000
GNE Myopathy (*GNE*) NM_001128227:1-12 Inheritance: Autosomal Recessive	Northwestern Europe: 1 in 230 Other Populations: < 1 in 500	Northwestern Europe: 99% Other Populations: 99%	Northwestern Europe: < 1 in 23,000 Other Populations: < 1 in 50,000
GNPTAB-related Disorders (*GNPTAB*) NM_024312:1-21 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 200 French Canadian/Cajun: 1 in 40 Middle East: 1 in 140 Other Populations: 1 in 200	Eastern Asia: 92% French Canadian/Cajun: 99% Middle East: 99% Other Populations: 99%	Eastern Asia: < 1 in 2,600 French Canadian/Cajun: < 1 in 3,900 Middle East: < 1 in 14,000 Other Populations: < 1 in 20,000
Galactokinase Deficiency (*GALK1*) NM_000154:1-8 Inheritance: Autosomal Recessive	Eastern Asia: < 1 in 500 Northwestern Europe: 1 in 370 Southern Europe: 1 in 310 Other Populations: 1 in 440	Eastern Asia: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Eastern Asia: < 1 in 50,000 Northwestern Europe: < 1 in 37,000 Southern Europe: < 1 in 30,000 Other Populations: < 1 in 44,000
Galactosemia (*GALT*) NM_000155:1-11 Inheritance: Autosomal Recessive	African American: 1 in 71 Ashkenazi Jewish: 1 in 160 Eastern Asia: 1 in 320 Northwestern Europe: 1 in 87 Other Populations: 1 in 110	African American: 99% Ashkenazi Jewish: 99% Eastern Asia: 99% Northwestern Europe: 99% Other Populations: 99%	African American: < 1 in 7,000 Ashkenazi Jewish: < 1 in 16,000 Eastern Asia: < 1 in 32,000 Northwestern Europe: < 1 in 8,600 Other Populations: < 1 in 11,000

* For X-linked diseases, female carrier frequencies are presented.
Detection rates are determined by using literature to estimate the fraction of disease alleles, weighted by frequency, that the methodology is unable to detect.
Other Populations denotes default ethnicity grouping based on identical risk and detection rates.

Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
Gamma-sarcoglycanopathy (*SGCG*) NM_000231:2-8 Inheritance: Autosomal Recessive	Eastern Asia: < 1 in 500 Hispanic: 1 in 430 Middle East: 1 in 95 Northwestern Europe: 1 in 440 South Asia: < 1 in 500 Southern Europe: 1 in 260 Other Populations: 1 in 340	Eastern Asia: 87% Hispanic: 87% Middle East: 87% Northwestern Europe: 87% South Asia: 87% Southern Europe: 87% Other Populations: 87%	Eastern Asia: < 1 in 3,800 Hispanic: < 1 in 3,300 Middle East: < 1 in 710 Northwestern Europe: < 1 in 3,300 South Asia: < 1 in 3,800 Southern Europe: < 1 in 2,000 Other Populations: < 1 in 2,600
Gaucher Disease (*GBA*) N409S, V433L, D448H, D448V, L483P, R502C, R502H, R535H, c.84dupG, c.115+1G>A Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 14 Eastern Asia: 1 in 180 Finland: 1 in 110 French Canadian/Cajun: 1 in 110 Northwestern Europe: 1 in 110 Southern Europe: 1 in 110 Other Populations: 1 in 120	Ashkenazi Jewish: 95% Eastern Asia: 60% Finland: 60% French Canadian/Cajun: 60% Northwestern Europe: 60% Southern Europe: 60% Other Populations: 60%	Ashkenazi Jewish: 1 in 250 Eastern Asia: 1 in 450 Finland: 1 in 260 French Canadian/Cajun: 1 in 260 Northwestern Europe: 1 in 260 Southern Europe: 1 in 260 Other Populations: 1 in 310
Glutaric Acidemia, GCDH-related (*GCDH*) NM_000159:2-12 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 140 French Canadian/Cajun: 1 in 160 Middle East: 1 in 66 Northwestern Europe: 1 in 160 Southern Europe: 1 in 140 Other Populations: 1 in 160	Eastern Asia: 99% French Canadian/Cajun: 99% Middle East: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Eastern Asia: < 1 in 13,000 French Canadian/Cajun: < 1 in 16,000 Middle East: < 1 in 6,500 Northwestern Europe: < 1 in 16,000 Southern Europe: < 1 in 14,000 Other Populations: < 1 in 16,000
Glycine Encephalopathy, AMT-related (*AMT*) NM_000481:1-9 Inheritance: Autosomal Recessive	Finland: 1 in 260 Other Populations: 1 in 260	Finland: 99% Other Populations: 99%	Finland: < 1 in 26,000 Other Populations: < 1 in 26,000
Glycine Encephalopathy, GLDC-related (*GLDC*) NM_000170:1-25 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 260 Finland: 1 in 130 Middle East: 1 in 97 Southeast Asia: 1 in 250 Southern Europe: 1 in 130 Other Populations: 1 in 160	Eastern Asia: 94% Finland: 94% Middle East: 94% Southeast Asia: 94% Southern Europe: 94% Other Populations: 94%	Eastern Asia: < 1 in 4,200 Finland: < 1 in 2,100 Middle East: < 1 in 1,600 Southeast Asia: < 1 in 4,100 Southern Europe: < 1 in 2,100 Other Populations: < 1 in 2,500
Glycogen Storage Disease Type III (*AGL*) NM_000642:2-34 Inheritance: Autosomal Recessive	Worldwide: 1 in 160	Worldwide: 99%	Worldwide: < 1 in 16,000
Glycogen Storage Disease Type Ia (*G6PC1*) NM_000151:1-5 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 68 Other Populations: 1 in 180	Ashkenazi Jewish: 99% Other Populations: 98%	Ashkenazi Jewish: < 1 in 6,700 Other Populations: < 1 in 8,700
Glycogen Storage Disease Type Ib (*SLC37A4*) NM_001164277:3-11 Inheritance: Autosomal Recessive	Worldwide: 1 in 350	Worldwide: 99%	Worldwide: < 1 in 35,000

* For X-linked diseases, female carrier frequencies are presented.

Detection rates are determined by using literature to estimate the fraction of disease alleles, weighted by frequency, that the methodology is unable to detect.

Other Populations denotes default ethnicity grouping based on identical risk and detection rates.

Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
HADHA-related Disorders (*HADHA*) NM_000182:1-20 Inheritance: Autosomal Recessive	Finland: 1 in 130 Northwestern Europe: 1 in 200 Other Populations: 1 in 250	Finland: 99% Northwestern Europe: 99% Other Populations: 99%	Finland: < 1 in 12,000 Northwestern Europe: < 1 in 20,000 Other Populations: < 1 in 25,000
HMG-CoA Lyase Deficiency (*HMGCL*) NM_000191:1-9 Inheritance: Autosomal Recessive	Middle East: 1 in 180 Southern Europe: 1 in 180 Other Populations: < 1 in 500	Middle East: 99% Southern Europe: 99% Other Populations: 99%	Middle East: < 1 in 18,000 Southern Europe: < 1 in 18,000 Other Populations: < 1 in 50,000
Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) (*HBB*) NM_000518:1-3 Inheritance: Autosomal Recessive	African American: 1 in 8 Ashkenazi Jewish: 1 in 24 Eastern Asia: 1 in 10 Finland: 1 in 80 French Canadian/Cajun: 1 in 24 Hispanic: 1 in 26 Middle East: 1 in 27 Native American: 1 in 24 Northwestern Europe: 1 in 38 South Asia: 1 in 25 Southeast Asia: 1 in 18 Southern Europe: 1 in 8	African American: 99% Ashkenazi Jewish: 99% Eastern Asia: 99% Finland: 99% French Canadian/Cajun: 99% Hispanic: 99% Middle East: 99% Native American: 99% Northwestern Europe: 99% South Asia: 99% Southeast Asia: 99% Southern Europe: 99%	African American: < 1 in 700 Ashkenazi Jewish: < 1 in 2,300 Eastern Asia: < 1 in 900 Finland: < 1 in 7,900 French Canadian/Cajun: < 1 in 2,300 Hispanic: < 1 in 2,500 Middle East: < 1 in 2,600 Native American: < 1 in 2,300 Northwestern Europe: < 1 in 3,700 South Asia: < 1 in 2,400 Southeast Asia: < 1 in 1,700 Southern Europe: < 1 in 710
Hereditary Fructose Intolerance (*ALDOB*) NM_000035:2-9 Inheritance: Autosomal Recessive	African American: 1 in 230 French Canadian/Cajun: 1 in 81 Middle East: 1 in 98 Other Populations: 1 in 80	African American: 99% French Canadian/Cajun: 99% Middle East: 99% Other Populations: 99%	African American: < 1 in 23,000 French Canadian/Cajun: < 1 in 8,000 Middle East: < 1 in 9,700 Other Populations: < 1 in 7,900
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) (*HEXA*) NM_000520:1-14 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 31 French Canadian/Cajun: 1 in 51 Other Populations: 1 in 300	Ashkenazi Jewish: 99% French Canadian/Cajun: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 3,000 French Canadian/Cajun: < 1 in 5,000 Other Populations: < 1 in 30,000
Holocarboxylase Synthetase Deficiency (*HLCS*) NM_000411:4-12 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 160 Other Populations: 1 in 150	Eastern Asia: 99% Other Populations: 99%	Eastern Asia: < 1 in 16,000 Other Populations: < 1 in 15,000
Homocystinuria, CBS-related (*CBS*) NM_000071:3-17 Inheritance: Autosomal Recessive	Eastern Asia: < 1 in 500 Hispanic: 1 in 100 Middle East: 1 in 41 Northwestern Europe: 1 in 95 Southern Europe: 1 in 180 Other Populations: 1 in 270	Eastern Asia: 99% Hispanic: 99% Middle East: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Eastern Asia: < 1 in 50,000 Hispanic: < 1 in 10,000 Middle East: < 1 in 4,000 Northwestern Europe: < 1 in 9,400 Southern Europe: < 1 in 17,000 Other Populations: < 1 in 27,000
Hydrolethalus Syndrome (*HYLS1*) NM_145014:4 Inheritance: Autosomal Recessive	Finland: 1 in 51 Other Populations: < 1 in 500	Finland: 99% Other Populations: 99%	Finland: < 1 in 5,000 Other Populations: < 1 in 50,000

* For X-linked diseases, female carrier frequencies are presented.

Detection rates are determined by using literature to estimate the fraction of disease alleles, weighted by frequency, that the methodology is unable to detect.

Other Populations denotes default ethnicity grouping based on identical risk and detection rates.

Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
Hypophosphatasia (*ALPL*) NM_000478:2-12 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 240 Northwestern Europe: 1 in 300 Southern Europe: 1 in 270 Other Populations: 1 in 230	Eastern Asia: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Eastern Asia: < 1 in 24,000 Northwestern Europe: < 1 in 30,000 Southern Europe: < 1 in 27,000 Other Populations: < 1 in 23,000
Isovaleric Acidemia (*IVD*) NM_002225:1-12 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 390 French Canadian/Cajun: 1 in 310 Middle East: 1 in 84 Northwestern Europe: 1 in 330 Southern Europe: 1 in 220 Other Populations: 1 in 260	Eastern Asia: 99% French Canadian/Cajun: 99% Middle East: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Eastern Asia: < 1 in 39,000 French Canadian/Cajun: < 1 in 31,000 Middle East: < 1 in 8,300 Northwestern Europe: < 1 in 32,000 Southern Europe: < 1 in 22,000 Other Populations: < 1 in 26,000
Joubert Syndrome 2 (*TMEM216*) NM_001173990:1-5 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 97 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 9,600 Other Populations: < 1 in 50,000
Junctional Epidermolysis Bullosa, LAMA3-related (*LAMA3*) NM_000227:1-38 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
Junctional Epidermolysis Bullosa, LAMB3-related (*LAMB3*) NM_000228:2-23 Inheritance: Autosomal Recessive	Northwestern Europe: 1 in 320 Southern Europe: 1 in 310 Other Populations: 1 in 320	Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Northwestern Europe: < 1 in 32,000 Southern Europe: < 1 in 31,000 Other Populations: < 1 in 31,000
Junctional Epidermolysis Bullosa, LAMC2-related (*LAMC2*) NM_005562:1-23 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
Krabbe Disease (*GALC*) NM_000153:1-17 Inheritance: Autosomal Recessive	Middle East: 1 in 160 Northwestern Europe: 1 in 140 Southern Europe: 1 in 150 Other Populations: 1 in 180	Middle East: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Middle East: < 1 in 16,000 Northwestern Europe: < 1 in 14,000 Southern Europe: < 1 in 14,000 Other Populations: < 1 in 17,000
Leigh Syndrome, French-Canadian Type (*LRPPRC*) NM_133259:1-38 Inheritance: Autosomal Recessive	French Canadian/Cajun: 1 in 23 Other Populations: < 1 in 500	French Canadian/Cajun: 99% Other Populations: 99%	French Canadian/Cajun: < 1 in 2,200 Other Populations: < 1 in 50,000
Lipoid Congenital Adrenal Hyperplasia (*STAR*) NM_000349:1-7 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 400 Other Populations: < 1 in 500	Eastern Asia: 99% Other Populations: 99%	Eastern Asia: < 1 in 40,000 Other Populations: < 1 in 50,000
Lysosomal Acid Lipase Deficiency (*LIPA*) NM_000235:2-10 Inheritance: Autosomal Recessive	African American: 1 in 240 Ashkenazi Jewish: 1 in 470 French Canadian/Cajun: 1 in 150 Hispanic: 1 in 160 Middle East: 1 in 33 Native American: 1 in 150	African American: 99% Ashkenazi Jewish: 99% French Canadian/Cajun: 99% Hispanic: 99% Middle East: 99% Native American: 99%	African American: < 1 in 17,000 Ashkenazi Jewish: < 1 in 32,000 French Canadian/Cajun: < 1 in 10,000 Hispanic: < 1 in 11,000 Middle East: < 1 in 2,200 Native American: < 1 in 10,000

* For X-linked diseases, female carrier frequencies are presented.

Detection rates are determined by using literature to estimate the fraction of disease alleles, weighted by frequency, that the methodology is unable to detect.

Other Populations denotes default ethnicity grouping based on identical risk and detection rates.

Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
	Northwestern Europe: 1 in 200 Southern Europe: 1 in 150 Other Populations: < 1 in 500	Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Northwestern Europe: < 1 in 14,000 Southern Europe: < 1 in 10,000 Other Populations: < 1 in 34,000
MKS1-related Disorders (*MKS1*) NM_017777:1-18 Inheritance: Autosomal Recessive	Finland: 1 in 48 Other Populations: < 1 in 500	Finland: 99% Other Populations: 99%	Finland: < 1 in 4,700 Other Populations: < 1 in 50,000
MYO7A-related Disorders (*MYO7A*) NM_000260:2-49 Inheritance: Autosomal Recessive	Finland: 1 in 210 Other Populations: 1 in 150	Finland: 99% Other Populations: 99%	Finland: < 1 in 21,000 Other Populations: < 1 in 15,000
Maple Syrup Urine Disease Type II (*DBT*) NM_001918:1-11 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 180 Eastern Asia: 1 in 470 Hispanic: 1 in 350 Middle East: 1 in 110 Northwestern Europe: 1 in 480 South Asia: 1 in 130 Southeast Asia: 1 in 180 Southern Europe: 1 in 220 Other Populations: 1 in 400	Ashkenazi Jewish: 97% Eastern Asia: 97% Hispanic: 97% Middle East: 97% Northwestern Europe: 97% South Asia: 97% Southeast Asia: 97% Southern Europe: 97% Other Populations: 97%	Ashkenazi Jewish: < 1 in 5,900 Eastern Asia: < 1 in 15,000 Hispanic: < 1 in 12,000 Middle East: < 1 in 3,500 Northwestern Europe: < 1 in 16,000 South Asia: < 1 in 4,100 Southeast Asia: < 1 in 5,700 Southern Europe: < 1 in 7,300 Other Populations: < 1 in 13,000
Maple Syrup Urine Disease Type Ia (*BCKDHA*) NM_000709:1-9 Inheritance: Autosomal Recessive	African American: 1 in 260 Ashkenazi Jewish: 1 in 320 Eastern Asia: 1 in 430 Finland: 1 in 320 French Canadian/Cajun: 1 in 290 Hispanic: 1 in 180 Middle East: 1 in 99 Native American: 1 in 320 Northwestern Europe: 1 in 390 South Asia: 1 in 95 Southeast Asia: 1 in 190 Southern Europe: 1 in 130	African American: 99% Ashkenazi Jewish: 99% Eastern Asia: 99% Finland: 99% French Canadian/Cajun: 99% Hispanic: 99% Middle East: 99% Native American: 99% Northwestern Europe: 99% South Asia: 99% Southeast Asia: 99% Southern Europe: 99%	African American: < 1 in 26,000 Ashkenazi Jewish: < 1 in 32,000 Eastern Asia: < 1 in 43,000 Finland: < 1 in 32,000 French Canadian/Cajun: < 1 in 29,000 Hispanic: < 1 in 18,000 Middle East: < 1 in 9,800 Native American: < 1 in 32,000 Northwestern Europe: < 1 in 39,000 South Asia: < 1 in 9,400 Southeast Asia: < 1 in 19,000 Southern Europe: < 1 in 13,000
Maple Syrup Urine Disease Type Ib (*BCKDHB*) NM_183050:1-10 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 98 Eastern Asia: 1 in 240 Middle East: 1 in 44 Northwestern Europe: 1 in 390 Southern Europe: 1 in 210 Other Populations: 1 in 360	Ashkenazi Jewish: 99% Eastern Asia: 99% Middle East: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 9,700 Eastern Asia: < 1 in 23,000 Middle East: < 1 in 4,300 Northwestern Europe: < 1 in 39,000 Southern Europe: < 1 in 21,000 Other Populations: < 1 in 36,000
Medium Chain Acyl-CoA Dehydrogenase Deficiency (*ACADM*) NM_000016:1-12 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 110 Middle East: 1 in 68 Northwestern Europe: 1 in 45 Southern Europe: 1 in 62 Other Populations: 1 in 61	Eastern Asia: 99% Middle East: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Eastern Asia: < 1 in 11,000 Middle East: < 1 in 6,700 Northwestern Europe: < 1 in 4,400 Southern Europe: < 1 in 6,100 Other Populations: < 1 in 6,000

* For X-linked diseases, female carrier frequencies are presented.

Detection rates are determined by using literature to estimate the fraction of disease alleles, weighted by frequency, that the methodology is unable to detect.

Other Populations denotes default ethnicity grouping based on identical risk and detection rates.

Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
Megalencephalic Leukoencephalopathy with Subcortical Cysts (*MLC1*) NM_015166:2-12 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
Metachromatic Leukodystrophy (*ARSA*) NM_000487:1-8 Inheritance: Autosomal Recessive	Native American: 1 in 41 Other Populations: 1 in 160	Native American: 99% Other Populations: 99%	Native American: < 1 in 4,000 Other Populations: < 1 in 16,000
Methylmalonic Acidemia, MMUT-related (*MMUT*) NM_000255:2-13 Inheritance: Autosomal Recessive	African American: 1 in 180 Ashkenazi Jewish: 1 in 110 Eastern Asia: 1 in 120 Finland: 1 in 110 French Canadian/Cajun: 1 in 380 Hispanic: 1 in 120 Middle East: 1 in 76 Native American: 1 in 110 Northwestern Europe: 1 in 270 South Asia: 1 in 100 Southeast Asia: 1 in 64 Southern Europe: 1 in 97	African American: 99% Ashkenazi Jewish: 99% Eastern Asia: 99% Finland: 99% French Canadian/Cajun: 99% Hispanic: 99% Middle East: 99% Native American: 99% Northwestern Europe: 99% South Asia: 99% Southeast Asia: 99% Southern Europe: 99%	African American: < 1 in 18,000 Ashkenazi Jewish: < 1 in 11,000 Eastern Asia: < 1 in 12,000 Finland: < 1 in 11,000 French Canadian/Cajun: < 1 in 38,000 Hispanic: < 1 in 12,000 Middle East: < 1 in 7,500 Native American: < 1 in 11,000 Northwestern Europe: < 1 in 26,000 South Asia: < 1 in 10,000 Southeast Asia: < 1 in 6,300 Southern Europe: < 1 in 9,600
Methylmalonic Acidemia, cblA Type (*MMAA*) NM_172250:2-7 Inheritance: Autosomal Recessive	French Canadian/Cajun: 1 in 470 Other Populations: < 1 in 500	French Canadian/Cajun: 99% Other Populations: 99%	French Canadian/Cajun: < 1 in 47,000 Other Populations: < 1 in 50,000
Methylmalonic Acidemia, cblB Type (*MMAB*) NM_052845:1-9 Inheritance: Autosomal Recessive	French Canadian/Cajun: < 1 in 660 Northwestern Europe: 1 in 480 Other Populations: < 1 in 500	French Canadian/Cajun: 99% Northwestern Europe: 99% Other Populations: 99%	French Canadian/Cajun: < 1 in 66,000 Northwestern Europe: < 1 in 48,000 Other Populations: < 1 in 50,000
Methylmalonic Aciduria and Homocystinuria, cblC Type (*MMACHC*) NM_015506:1-4 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 330 French Canadian/Cajun: 1 in 230 Other Populations: 1 in 160	Eastern Asia: 99% French Canadian/Cajun: 99% Other Populations: 99%	Eastern Asia: < 1 in 33,000 French Canadian/Cajun: < 1 in 23,000 Other Populations: < 1 in 16,000
Mucolipidosis III Gamma (*GNPTG*) NM_032520:1-11 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 98%	Worldwide: < 1 in 20,000
Mucolipidosis IV (*MCOLN1*) NM_020533:1-14 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 90 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 8,900 Other Populations: < 1 in 50,000
Mucopolysaccharidosis Type I (*IDUA*) NM_000203:1-14 Inheritance: Autosomal Recessive	Middle East: 1 in 80 Other Populations: 1 in 160	Middle East: 99% Other Populations: 99%	Middle East: < 1 in 7,900 Other Populations: < 1 in 16,000
* **Mucopolysaccharidosis Type II (*IDS*)** NM_000202:1-9 Inheritance: X-linked Recessive	Ashkenazi Jewish: 1 in 54,000 Eastern Asia: 1 in 29,000 Other Populations: 1 in 75,000	Ashkenazi Jewish: 89% Eastern Asia: 89% Other Populations: 89%	Ashkenazi Jewish: 1 in 480,000 Eastern Asia: 1 in 260,000 Other Populations: 1 in 670,000

* For X-linked diseases, female carrier frequencies are presented.

Detection rates are determined by using literature to estimate the fraction of disease alleles, weighted by frequency, that the methodology is unable to detect.

Other Populations denotes default ethnicity grouping based on identical risk and detection rates.

Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
Mucopolysaccharidosis Type IIIA (*SGSH*) NM_000199:1-8 Inheritance: Autosomal Recessive	Middle East: 1 in 150 Northwestern Europe: 1 in 190 Other Populations: 1 in 160	Middle East: 99% Northwestern Europe: 99% Other Populations: 99%	Middle East: < 1 in 14,000 Northwestern Europe: < 1 in 19,000 Other Populations: < 1 in 16,000
Mucopolysaccharidosis Type IIIB (*NAGLU*) NM_000263:1-6 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 420 Middle East: 1 in 110 Northwestern Europe: 1 in 270 Southern Europe: 1 in 210 Other Populations: 1 in 260	Eastern Asia: 99% Middle East: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Eastern Asia: < 1 in 42,000 Middle East: < 1 in 11,000 Northwestern Europe: < 1 in 27,000 Southern Europe: < 1 in 21,000 Other Populations: < 1 in 26,000
Mucopolysaccharidosis Type IIIC (*HGSNAT*) NM_152419:1-18 Inheritance: Autosomal Recessive	Middle East: 1 in 350 Southern Europe: 1 in 470 Other Populations: < 1 in 500	Middle East: 99% Southern Europe: 99% Other Populations: 99%	Middle East: < 1 in 35,000 Southern Europe: < 1 in 47,000 Other Populations: < 1 in 50,000
Muscular Dystrophy, LAMA2-related (*LAMA2*) NM_000426:1-65 Inheritance: Autosomal Recessive	Southern Europe: 1 in 96 Other Populations: 1 in 120	Southern Europe: 98% Other Populations: 98%	Southern Europe: < 1 in 4,500 Other Populations: < 1 in 5,700
NEB-related Nemaline Myopathy (*NEB*) NM_001271208:3-80,117-183 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 110 Other Populations: 1 in 87	Ashkenazi Jewish: 99% Other Populations: 93%	Ashkenazi Jewish: < 1 in 11,000 Other Populations: < 1 in 1,200
Nephrotic Syndrome, NPHS1-related (*NPHS1*) NM_004646:1-29 Inheritance: Autosomal Recessive	Finland: 1 in 46 Other Populations: < 1 in 500	Finland: 99% Other Populations: 99%	Finland: < 1 in 4,500 Other Populations: < 1 in 50,000
Nephrotic Syndrome, NPHS2-related (*NPHS2*) NM_014625:1-8 Inheritance: Autosomal Recessive	French Canadian/Cajun: 1 in 360 Middle East: 1 in 360 Native American: 1 in 360 Southeast Asia: 1 in 360 Southern Europe: 1 in 360 Other Populations: 1 in 360	French Canadian/Cajun: 99% Middle East: 99% Native American: 99% Southeast Asia: 99% Southern Europe: 99% Other Populations: 99%	French Canadian/Cajun: < 1 in 35,000 Middle East: < 1 in 35,000 Native American: < 1 in 35,000 Southeast Asia: < 1 in 35,000 Southern Europe: < 1 in 35,000 Other Populations: < 1 in 35,000
Neuronal Ceroid Lipofuscinosis, CLN6-related (*CLN6*) NM_017882:1-7 Inheritance: Autosomal Recessive	Northwestern Europe: 1 in 200 Southern Europe: 1 in 390 Other Populations: < 1 in 500	Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Northwestern Europe: < 1 in 20,000 Southern Europe: < 1 in 39,000 Other Populations: < 1 in 50,000
Niemann-Pick Disease Type C1 (*NPC1*) NM_000271:1-25 Inheritance: Autosomal Recessive	Northwestern Europe: 1 in 190 Southern Europe: 1 in 110 Other Populations: 1 in 170	Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Northwestern Europe: < 1 in 19,000 Southern Europe: < 1 in 11,000 Other Populations: < 1 in 17,000
Niemann-Pick Disease Type C2 (*NPC2*) NM_006432:1-5 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000

* For X-linked diseases, female carrier frequencies are presented.

Detection rates are determined by using literature to estimate the fraction of disease alleles, weighted by frequency, that the methodology is unable to detect.

Other Populations denotes default ethnicity grouping based on identical risk and detection rates.

Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
Niemann-Pick Disease, SMPD1-related (*SMPD1*) NM_000543:1-6 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 100 Other Populations: 1 in 250	Ashkenazi Jewish: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 10,000 Other Populations: < 1 in 25,000
Nijmegen Breakage Syndrome (*NBN*) NM_002485:1-16 Inheritance: Autosomal Recessive	Northwestern Europe: 1 in 160 Southern Europe: 1 in 160 Other Populations: < 1 in 500	Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Northwestern Europe: < 1 in 16,000 Southern Europe: < 1 in 16,000 Other Populations: < 1 in 50,000
* **Ornithine Transcarbamylase Deficiency (*OTC*)** NM_000531:1-10 Inheritance: X-linked Recessive	Finland: 1 in 31,000 Other Populations: 1 in 34,000	Finland: 97% Other Populations: 97%	Finland: < 1 in 1,000,000 Other Populations: < 1 in 1,000,000
PCCA-related Propionic Acidemia (*PCCA*) NM_000282:1-24 Inheritance: Autosomal Recessive	Middle East: 1 in 91 Other Populations: 1 in 220	Middle East: 95% Other Populations: 95%	Middle East: < 1 in 1,700 Other Populations: < 1 in 4,200
PCCB-related Propionic Acidemia (*PCCB*) NM_000532:1-15 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 66 Middle East: 1 in 100 Other Populations: 1 in 220	Eastern Asia: 99% Middle East: 99% Other Populations: 99%	Eastern Asia: < 1 in 6,500 Middle East: < 1 in 10,000 Other Populations: < 1 in 22,000
PCDH15-related Disorders (*PCDH15*) NM_033056:2-33 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 78 Other Populations: 1 in 220	Ashkenazi Jewish: 93% Other Populations: 93%	Ashkenazi Jewish: < 1 in 1,200 Other Populations: < 1 in 3,300
POMGNT-related Disorders (*POMGNT1*) NM_017739:2-22 Inheritance: Autosomal Recessive	Finland: 1 in 110 Other Populations: < 1 in 500	Finland: 98% Other Populations: 96%	Finland: < 1 in 5,600 Other Populations: < 1 in 12,000
PPT1-related Neuronal Ceroid Lipofuscinosis (*PPT1*) NM_000310:1-9 Inheritance: Autosomal Recessive	Finland: 1 in 71 Other Populations: 1 in 78	Finland: 99% Other Populations: 99%	Finland: < 1 in 7,000 Other Populations: < 1 in 7,700
Pendred Syndrome (*SLC26A4*) NM_000441:2-21 Inheritance: Autosomal Recessive	French Canadian/Cajun: 1 in 83 Northwestern Europe: 1 in 83 Other Populations: 1 in 65	French Canadian/Cajun: 99% Northwestern Europe: 99% Other Populations: 99%	French Canadian/Cajun: < 1 in 8,200 Northwestern Europe: < 1 in 8,200 Other Populations: < 1 in 6,400
Peroxisome Biogenesis Disorder Type 1 (*PEX1*) NM_000466:1-24 Inheritance: Autosomal Recessive	Worldwide: 1 in 160	Worldwide: 99%	Worldwide: < 1 in 16,000
Peroxisome Biogenesis Disorder Type 3 (*PEX12*) NM_000286:1-3 Inheritance: Autosomal Recessive	Eastern Asia: < 1 in 500 Other Populations: 1 in 440	Eastern Asia: 99% Other Populations: 99%	Eastern Asia: < 1 in 50,000 Other Populations: < 1 in 44,000
Peroxisome Biogenesis Disorder Type 4 (*PEX6*) NM_000287:1-17 Inheritance: Autosomal Recessive	French Canadian/Cajun: 1 in 56 Other Populations: 1 in 310	French Canadian/Cajun: 97% Other Populations: 97%	French Canadian/Cajun: < 1 in 1,600 Other Populations: < 1 in 9,300

* For X-linked diseases, female carrier frequencies are presented.

Detection rates are determined by using literature to estimate the fraction of disease alleles, weighted by frequency, that the methodology is unable to detect.

Other Populations denotes default ethnicity grouping based on identical risk and detection rates.

Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
Peroxisome Biogenesis Disorder Type 5 (*PEX2*) NM_000318:4 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 120 Other Populations: < 1 in 710	Ashkenazi Jewish: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 12,000 Other Populations: < 1 in 71,000
Peroxisome Biogenesis Disorder Type 6 (*PEX10*) NM_153818:1-6 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
Phenylalanine Hydroxylase Deficiency (*PAH*) NM_000277:1-13 Inheritance: Autosomal Recessive	African American: 1 in 110 Ashkenazi Jewish: 1 in 220 Eastern Asia: 1 in 78 Finland: 1 in 180 French Canadian/Cajun: 1 in 75 Hispanic: 1 in 72 Middle East: 1 in 37 Native American: 1 in 56 Northwestern Europe: 1 in 49 South Asia: 1 in 87 Southeast Asia: < 1 in 500 Southern Europe: 1 in 42	African American: 99% Ashkenazi Jewish: 99% Eastern Asia: 99% Finland: 99% French Canadian/Cajun: 99% Hispanic: 99% Middle East: 99% Native American: 99% Northwestern Europe: 99% South Asia: 99% Southeast Asia: 99% Southern Europe: 99%	African American: < 1 in 11,000 Ashkenazi Jewish: < 1 in 22,000 Eastern Asia: < 1 in 7,700 Finland: < 1 in 18,000 French Canadian/Cajun: < 1 in 7,400 Hispanic: < 1 in 7,100 Middle East: < 1 in 3,600 Native American: < 1 in 5,500 Northwestern Europe: < 1 in 4,800 South Asia: < 1 in 8,600 Southeast Asia: < 1 in 50,000 Southern Europe: < 1 in 4,100
Pompe Disease (*GAA*) NM_000152:2-20 Inheritance: Autosomal Recessive	African American: 1 in 60 Eastern Asia: 1 in 100 Hispanic: 1 in 100 Northwestern Europe: 1 in 100 Southern Europe: 1 in 380 Other Populations: 1 in 100	African American: 99% Eastern Asia: 99% Hispanic: 99% Northwestern Europe: 98% Southern Europe: 98% Other Populations: 99%	African American: < 1 in 5,900 Eastern Asia: < 1 in 10,000 Hispanic: < 1 in 6,500 Northwestern Europe: < 1 in 4,000 Southern Europe: < 1 in 15,000 Other Populations: < 1 in 10,000
Primary Carnitine Deficiency (*SLC22A5*) NM_003060:1-10 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 100 Northwestern Europe: 1 in 110 Other Populations: 1 in 160	Eastern Asia: 99% Northwestern Europe: 99% Other Populations: 99%	Eastern Asia: < 1 in 10,000 Northwestern Europe: < 1 in 11,000 Other Populations: < 1 in 16,000
Primary Hyperoxaluria Type 1 (*AGXT*) NM_000030:1-11 Inheritance: Autosomal Recessive	African American: 1 in 200 French Canadian/Cajun: 1 in 170 Northwestern Europe: 1 in 170 Southern Europe: 1 in 170 Other Populations: 1 in 140	African American: 99% French Canadian/Cajun: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	African American: < 1 in 20,000 French Canadian/Cajun: < 1 in 17,000 Northwestern Europe: < 1 in 17,000 Southern Europe: < 1 in 17,000 Other Populations: < 1 in 13,000
Primary Hyperoxaluria Type 2 (*GRHPR*) NM_012203:1-9 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
Primary Hyperoxaluria Type 3 (*HOGA1*) NM_138413:1-7 Inheritance: Autosomal Recessive	African American: < 1 in 500 Ashkenazi Jewish: 1 in 57 Finland: 1 in 130 French Canadian/Cajun: 1 in 130 Northwestern Europe: 1 in 130	African American: 99% Ashkenazi Jewish: 99% Finland: 99% French Canadian/Cajun: 99% Northwestern Europe: 99%	African American: < 1 in 50,000 Ashkenazi Jewish: < 1 in 5,600 Finland: < 1 in 13,000 French Canadian/Cajun: < 1 in 13,000 Northwestern Europe: < 1 in 13,000

* For X-linked diseases, female carrier frequencies are presented.

Detection rates are determined by using literature to estimate the fraction of disease alleles, weighted by frequency, that the methodology is unable to detect.

Other Populations denotes default ethnicity grouping based on identical risk and detection rates.

Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
	Southern Europe: 1 in 130 Other Populations: 1 in 200	Southern Europe: 99% Other Populations: 99%	Southern Europe: < 1 in 13,000 Other Populations: < 1 in 20,000
Pycnodysostosis (*CTSK*) NM_000396:2-8 Inheritance: Autosomal Recessive	Worldwide: 1 in 430	Worldwide: 99%	Worldwide: < 1 in 43,000
Pyruvate Carboxylase Deficiency (*PC*) NM_000920:3-22 Inheritance: Autosomal Recessive	Worldwide: 1 in 250	Worldwide: 99%	Worldwide: < 1 in 25,000
RTEL1-related Disorders (*RTEL1*) NM_032957:2-35 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 100 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 10,000 Other Populations: < 1 in 50,000
Rhizomelic Chondrodysplasia Punctata Type 1 (*PEX7*) NM_000288:1-10 Inheritance: Autosomal Recessive	Worldwide: 1 in 160	Worldwide: 99%	Worldwide: < 1 in 16,000
SLC26A2-related Disorders (*SLC26A2*) NM_000112:2-3 Inheritance: Autosomal Recessive	Finland: 1 in 87 Hispanic: 1 in 140 Other Populations: 1 in 160	Finland: 99% Hispanic: 99% Other Populations: 99%	Finland: < 1 in 8,600 Hispanic: < 1 in 14,000 Other Populations: < 1 in 16,000
Sandhoff Disease (*HEXB*) NM_000521:1-14 Inheritance: Autosomal Recessive	Ashkenazi Jewish: < 1 in 500 French Canadian/Cajun: 1 in 120 Other Populations: 1 in 320	Ashkenazi Jewish: 98% French Canadian/Cajun: 98% Other Populations: 98%	Ashkenazi Jewish: < 1 in 29,000 French Canadian/Cajun: < 1 in 6,500 Other Populations: < 1 in 18,000
Short-chain Acyl-CoA Dehydrogenase Deficiency (*ACADS*) NM_000017:1-10 Inheritance: Autosomal Recessive	Northwestern Europe: 1 in 110 Other Populations: 1 in 98	Northwestern Europe: 99% Other Populations: 99%	Northwestern Europe: < 1 in 11,000 Other Populations: < 1 in 9,700
Sjogren-Larsson Syndrome (*ALDH3A2*) NM_000382:1-10 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 96%	Worldwide: < 1 in 12,000
Smith-Lemli-Opitz Syndrome (*DHCR7*) NM_001360:3-9 Inheritance: Autosomal Recessive	African American: 1 in 180 Eastern Asia: < 1 in 500 Hispanic: 1 in 170 Middle East: < 1 in 500 South Asia: < 1 in 500 Southeast Asia: < 1 in 500 Other Populations: 1 in 95	African American: 99% Eastern Asia: 99% Hispanic: 99% Middle East: 99% South Asia: 99% Southeast Asia: 99% Other Populations: 99%	African American: < 1 in 18,000 Eastern Asia: < 1 in 50,000 Hispanic: < 1 in 17,000 Middle East: < 1 in 50,000 South Asia: < 1 in 50,000 Southeast Asia: < 1 in 50,000 Other Populations: < 1 in 9,400
Spastic Paraplegia Type 15 (*ZFYVE26*) NM_015346:2-42 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000

* For X-linked diseases, female carrier frequencies are presented.

Detection rates are determined by using literature to estimate the fraction of disease alleles, weighted by frequency, that the methodology is unable to detect.

Other Populations denotes default ethnicity grouping based on identical risk and detection rates.

Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
Spinal Muscular Atrophy (*SMN1*) SMN1 copy number Inheritance: Autosomal Recessive	African American: 1 in 66 Ashkenazi Jewish: 1 in 41 Eastern Asia: 1 in 53 Finland: 1 in 35 French Canadian/Cajun: 1 in 35 Hispanic: 1 in 120 Middle East: 1 in 50 Native American: 1 in 50 Northwestern Europe: 1 in 35 South Asia: 1 in 50 Southeast Asia: 1 in 53 Southern Europe: 1 in 57	African American: 71% Ashkenazi Jewish: 94% Eastern Asia: 93% Finland: 94% French Canadian/Cajun: 95% Hispanic: 91% Middle East: 92% Native American: 93% Northwestern Europe: 95% South Asia: 93% Southeast Asia: 93% Southern Europe: 94%	African American: 1 in 120 Ashkenazi Jewish: 1 in 350 Eastern Asia: < 1 in 630 Finland: < 1 in 560 French Canadian/Cajun: < 1 in 570 Hispanic: < 1 in 1,100 Middle East: < 1 in 560 Native American: < 1 in 690 Northwestern Europe: < 1 in 630 South Asia: < 1 in 630 Southeast Asia: < 1 in 630 Southern Europe: < 1 in 890
Spondylothoracic Dysostosis (*MESP2*) NM_001039958:1-2 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
TGM1-related Autosomal Recessive Congenital Ichthyosis (*TGM1*) NM_000359:2-15 Inheritance: Autosomal Recessive	Worldwide: 1 in 220	Worldwide: 99%	Worldwide: < 1 in 22,000
TPP1-related Neuronal Ceroid Lipofuscinosis (*TPP1*) NM_000391:1-13 Inheritance: Autosomal Recessive	Worldwide: 1 in 300	Worldwide: 99%	Worldwide: < 1 in 30,000
Tyrosine Hydroxylase Deficiency (*TH*) NM_199292:1-14 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
Tyrosinemia Type I (*FAH*) NM_000137:1-14 Inheritance: Autosomal Recessive	Finland: 1 in 120 French Canadian/Cajun: 1 in 64 Other Populations: 1 in 160	Finland: 99% French Canadian/Cajun: 99% Other Populations: 99%	Finland: < 1 in 12,000 French Canadian/Cajun: < 1 in 6,300 Other Populations: < 1 in 16,000
Tyrosinemia Type II (*TAT*) NM_000353:2-12 Inheritance: Autosomal Recessive	Worldwide: 1 in 250	Worldwide: 99%	Worldwide: < 1 in 25,000
USH1C-related Disorders (*USH1C*) NM_005709:1-21 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 450 French Canadian/Cajun: 1 in 100 Southern Europe: < 1 in 500 Other Populations: 1 in 300	Eastern Asia: 99% French Canadian/Cajun: 99% Southern Europe: 99% Other Populations: 99%	Eastern Asia: < 1 in 44,000 French Canadian/Cajun: < 1 in 10,000 Southern Europe: < 1 in 50,000 Other Populations: < 1 in 30,000
USH2A-related Disorders (*USH2A*) NM_206933:2-72 Inheritance: Autosomal Recessive	Hispanic: 1 in 180 Middle East: 1 in 110 Northwestern Europe: 1 in 88 Southern Europe: 1 in 140 Other Populations: 1 in 150	Hispanic: 98% Middle East: 98% Northwestern Europe: 98% Southern Europe: 98% Other Populations: 98%	Hispanic: < 1 in 7,200 Middle East: < 1 in 4,400 Northwestern Europe: < 1 in 4,100 Southern Europe: < 1 in 6,500 Other Populations: < 1 in 5,900

* For X-linked diseases, female carrier frequencies are presented.

Detection rates are determined by using literature to estimate the fraction of disease alleles, weighted by frequency, that the methodology is unable to detect.

Other Populations denotes default ethnicity grouping based on identical risk and detection rates.

Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
Usher Syndrome Type 3 (*CLRN1*) NM_174878:1-3 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 130 Finland: 1 in 130 Other Populations: 1 in 410	Ashkenazi Jewish: 99% Finland: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 13,000 Finland: < 1 in 13,000 Other Populations: < 1 in 41,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency (*ACADVL*) NM_000018:1-20 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 130 Northwestern Europe: 1 in 180 Southern Europe: 1 in 200 Other Populations: 1 in 140	Eastern Asia: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Eastern Asia: < 1 in 12,000 Northwestern Europe: < 1 in 18,000 Southern Europe: < 1 in 20,000 Other Populations: < 1 in 14,000
Wilson Disease (*ATP7B*) NM_000053:1-21 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 66 Northwestern Europe: 1 in 66 Southeast Asia: 1 in 66 Other Populations: 1 in 91	Eastern Asia: 99% Northwestern Europe: 99% Southeast Asia: 99% Other Populations: 99%	Eastern Asia: < 1 in 6,500 Northwestern Europe: < 1 in 6,500 Southeast Asia: < 1 in 6,500 Other Populations: < 1 in 9,000
* **X-linked Adrenal Hypoplasia Congenita (*NR0B1*)** NM_000475:1-2 Inheritance: X-linked Recessive	Worldwide: 1 in 300,000	Worldwide: 97%	Worldwide: < 1 in 1,000,000
* **X-linked Adrenoleukodystrophy (*ABCD1*)** NM_000033:1-6 Inheritance: X-linked Recessive	Eastern Asia: 1 in 20,000 Southern Europe: 1 in 14,000 Other Populations: 1 in 11,000	Eastern Asia: 77% Southern Europe: 77% Other Populations: 77%	Eastern Asia: 1 in 86,000 Southern Europe: 1 in 60,000 Other Populations: 1 in 45,000
* **X-linked Juvenile Retinoschisis (*RS1*)** NM_000330:1-6 Inheritance: X-linked Recessive	Ashkenazi Jewish: 1 in 10,000 Finland: 1 in 8,500 French Canadian/Cajun: 1 in 10,000 Northwestern Europe: 1 in 10,000 Southern Europe: 1 in 10,000 Other Populations: 1 in 13,000	Ashkenazi Jewish: 99% Finland: 99% French Canadian/Cajun: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Ashkenazi Jewish: 1 in 670,000 Finland: 1 in 570,000 French Canadian/Cajun: 1 in 670,000 Northwestern Europe: 1 in 670,000 Southern Europe: 1 in 670,000 Other Populations: 1 in 840,000
* **X-linked Severe Combined Immunodeficiency (*IL2RG*)** NM_000206:1-8 Inheritance: X-linked Recessive	Worldwide: 1 in 50,000	Worldwide: 99%	Worldwide: < 1 in 1,000,000
Xeroderma Pigmentosum Group A (*XPA*) NM_000380:1-6 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 100 Middle East: 1 in 280 South Asia: 1 in 280 Other Populations: < 1 in 500	Eastern Asia: 99% Middle East: 99% South Asia: 99% Other Populations: 99%	Eastern Asia: < 1 in 10,000 Middle East: < 1 in 28,000 South Asia: < 1 in 28,000 Other Populations: < 1 in 50,000
Xeroderma Pigmentosum Group C (*XPC*) NM_004628:1-16 Inheritance: Autosomal Recessive	Worldwide: 1 in 240	Worldwide: 97%	Worldwide: < 1 in 7,300

* For X-linked diseases, female carrier frequencies are presented.
Detection rates are determined by using literature to estimate the fraction of disease alleles, weighted by frequency, that the methodology is unable to detect.
Other Populations denotes default ethnicity grouping based on identical risk and detection rates.