Myriad Foresight® Residual Risk Table

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Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
3-methylcrotonyl-CoA Carboxylase Deficiency, MCC2-related (*MCCC2*) NM_022132:1-17 Inheritance: Autosomal Recessive	Worldwide: 1 in 190	Worldwide: 99%	Worldwide: < 1 in 19,000
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
* **ARX-related Disorders (*ARX*)** NM_139058:1,3-5 Inheritance: X-linked Recessive	Worldwide: 1 in 8,700	Worldwide: 31%	Worldwide: 1 in 12,000
* **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
Abetalipoproteinemia (*MTTP*) NM_000253:2-19 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 40,000
Achromatopsia, CNGB3-related (*CNGB3*) NM_019098:1-18 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 190 Middle East: 1 in 56 Northwestern Europe: 1 in 92 South Asia: 1 in 24 Other Populations: 1 in 98	Eastern Asia: 99% Middle East: 99% Northwestern Europe: 99% South Asia: 99% Other Populations: 99%	Eastern Asia: < 1 in 19,000 Middle East: < 1 in 5,500 Northwestern Europe: < 1 in 9,100 South Asia: < 1 in 2,300 Other Populations: < 1 in 9,700
Acute Liver Failure, TRMU-related (*TRMU*) NM_018006:1-11 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
Adenosine Deaminase Deficiency (*ADA*) NM_000022:1-12 Inheritance: Autosomal Recessive	Worldwide: 1 in 390	Worldwide: 98%	Worldwide: < 1 in 22,000
Aicardi-Goutières Syndrome (*RNASEH2B*) NM_024570:1-11 Inheritance: Autosomal Recessive	Worldwide: 1 in 220	Worldwide: 99%	Worldwide: < 1 in 22,000

* For X-linked diseases, female carrier frequencies are presented.
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Aldosterone Synthase Deficiency (*CYP11B2*) R181W, V386A, V35Afs*3 Inheritance: Autosomal Recessive	Middle East: < 1 in 500 Other Populations: < 1 in 500	Middle East: 99% Other Populations: 33%	Middle East: < 1 in 50,000 Other Populations: < 1 in 740
Alkaptonuria (*HGD*) NM_000187:1-14 Inheritance: Autosomal Recessive	Worldwide: 1 in 400	Worldwide: 99%	Worldwide: < 1 in 39,000
Alpha-1 Antitrypsin Deficiency (*SERPINA1*) NM_000295:2-5 Inheritance: Autosomal Recessive	African American: 1 in 190 Ashkenazi Jewish: 1 in 55 Eastern Asia: < 1 in 500 Finland: 1 in 28 French Canadian/Cajun: 1 in 28 Hispanic: 1 in 130 Middle East: 1 in 130 Native American: 1 in 80 Northwestern Europe: 1 in 28 South Asia: < 1 in 500 Southeast Asia: 1 in 250 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 18,000 Ashkenazi Jewish: < 1 in 5,400 Eastern Asia: < 1 in 50,000 Finland: < 1 in 2,700 French Canadian/Cajun: < 1 in 2,700 Hispanic: < 1 in 12,000 Middle East: < 1 in 13,000 Native American: < 1 in 7,900 Northwestern Europe: < 1 in 2,700 South Asia: < 1 in 50,000 Southeast Asia: < 1 in 25,000 Southern Europe: < 1 in 2,700
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
Alport Syndrome, COL4A3-related (*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
Alport Syndrome, COL4A4-related (*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
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

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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 Northwestern Europe: 1 in 160 Other Populations: 1 in 130	Eastern Asia: 99% Finland: 99% Hispanic: 99% Northwestern Europe: 99% Other Populations: 99%	Eastern Asia: < 1 in 45,000 Finland: < 1 in 19,000 Hispanic: < 1 in 29,000 Northwestern Europe: < 1 in 15,000 Other Populations: < 1 in 13,000
Asparagine Synthetase Deficiency (*ASNS*) NM_001673:3-13 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
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
Atransferrinemia (*TF*) NM_001063:1-17 Inheritance: Autosomal Recessive	Worldwide: 1 in 120	Worldwide: 99%	Worldwide: < 1 in 12,000
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

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

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Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
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	Middle East: 1 in 470 Northwestern Europe: 1 in 360 Southern Europe: 1 in 440 Other Populations: < 1 in 500	Middle East: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Middle East: < 1 in 47,000 Northwestern Europe: < 1 in 36,000 Southern Europe: < 1 in 43,000 Other Populations: < 1 in 50,000
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 Globin-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
Beta-ketothiolase Deficiency (*ACAT1*) NM_000019:1-12 Inheritance: Autosomal Recessive	Worldwide: 1 in 200	Worldwide: 98%	Worldwide: < 1 in 9,600
Beta-sarcoglycanopathy (*SGCB*) NM_000232:1-6 Inheritance: Autosomal Recessive	Worldwide: 1 in 400	Worldwide: 99%	Worldwide: < 1 in 39,000

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Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
Biotin-thiamine–responsive Basal Ganglia Disease (*SLC19A3*) NM_025243:2-6 Inheritance: Autosomal Recessive	Worldwide: 1 in 110	Worldwide: 98%	Worldwide: < 1 in 4,500
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
CC2D2A-related Disorders (*CC2D2A*) NM_001080522:3-38 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 200 Other Populations: 1 in 200	Eastern Asia: 60% Other Populations: 99%	Eastern Asia: < 1 in 500 Other Populations: < 1 in 20,000
CEP290-related Disorders (*CEP290*) NM_025114:2-54 Inheritance: Autosomal Recessive	Finland: 1 in 69 French Canadian/Cajun: 1 in 69 Hispanic: 1 in 69 Northwestern Europe: 1 in 69 Southern Europe: 1 in 69 Other Populations: 1 in 69	Finland: 60% French Canadian/Cajun: 60% Hispanic: 70% Northwestern Europe: 60% Southern Europe: 60% Other Populations: 99%	Finland: 1 in 170 French Canadian/Cajun: 1 in 170 Hispanic: 1 in 230 Northwestern Europe: 1 in 170 Southern Europe: 1 in 170 Other Populations: < 1 in 6,800
CLN3-related Disorders (*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
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

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Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
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
Chronic Granulomatous Disease, CYBA-related (*CYBA*) NM_000101:1-6 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
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
Classical-like Ehlers-Danlos Syndrome, TNXB-related (*TNXB*) NM_019105:2-20,23-31 Inheritance: Autosomal Recessive	Worldwide: 1 in 28	Worldwide: 36%	Worldwide: 1 in 44
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
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

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Congenital Adrenal Hyperplasia, CYP11A1-related (*CYP11A1*) NM_000781:1-9 Inheritance: Autosomal Recessive	Worldwide: 1 in 110	Worldwide: 99%	Worldwide: < 1 in 11,000
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 Amegakaryocytic Thrombocytopenia (*MPL*) NM_005373:1-12 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 76 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 7,500 Other Populations: < 1 in 50,000
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
Congenital Disorder of Glycosylation, PMM2-related (*PMM2*) NM_000303:1-8 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 62 Eastern Asia: 1 in 450 Finland: 1 in 71 French Canadian/Cajun: 1 in 71 Northwestern Europe: 1 in 71 South Asia: 1 in 450 Southeast Asia: 1 in 450 Southern Europe: 1 in 71 Other Populations: 1 in 140	Ashkenazi Jewish: 99% Eastern Asia: 99% Finland: 99% French Canadian/Cajun: 99% Northwestern Europe: 99% South Asia: 99% Southeast Asia: 99% Southern Europe: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 6,100 Eastern Asia: < 1 in 45,000 Finland: < 1 in 7,000 French Canadian/Cajun: < 1 in 7,000 Northwestern Europe: < 1 in 7,000 South Asia: < 1 in 45,000 Southeast Asia: < 1 in 45,000 Southern Europe: < 1 in 7,000 Other Populations: < 1 in 14,000
Congenital Hydrocephalus, CCDC88C-related (*CCDC88C*) NM_001080414:1-30 Inheritance: Autosomal Recessive	Worldwide: 1 in 140	Worldwide: 98%	Worldwide: < 1 in 6,700
Congenital Insensitivity to Pain with Anhidrosis, NTRK1-related (*NTRK1*) NM_002529:1-17 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 440 Other Populations: < 1 in 500	Eastern Asia: 99% Other Populations: 99%	Eastern Asia: < 1 in 44,000 Other Populations: < 1 in 50,000

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Congenital Myasthenic Syndrome, CHRNE-related (*CHRNE*) NM_000080:1-12 Inheritance: Autosomal Recessive	Worldwide: 1 in 74	Worldwide: 98%	Worldwide: < 1 in 3,000
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
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
DYNC2H1-related Disorders (*DYNC2H1*) NM_001377:2-89 Inheritance: Autosomal Recessive	Worldwide: 1 in 67	Worldwide: 99%	Worldwide: < 1 in 6,700
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
Dihydropyrimidine Dehydrogenase Deficiency (*DPYD*) NM_000110:1-23 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 98%	Worldwide: < 1 in 29,000
Distal Renal Tubular Acidosis with Deafness, ATP6V1B1-related (*ATP6V1B1*) NM_001692:1-14 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000

* For X-linked diseases, female carrier frequencies are presented.
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Other Populations denotes default ethnicity grouping based on identical risk and detection rates.

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Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
Donnai–Barrow Syndrome (*LRP2*) NM_004525:1-79 Inheritance: Autosomal Recessive	Worldwide: 1 in 210	Worldwide: 99%	Worldwide: < 1 in 21,000
Dysferlinopathy (*DYSF*) NM_003494:1-55 Inheritance: Autosomal Recessive	Worldwide: 1 in 190	Worldwide: 98%	Worldwide: < 1 in 11,000
ERCC2-related Disorders (*ERCC2*) NM_000400:1-23 Inheritance: Autosomal Recessive	Worldwide: 1 in 66	Worldwide: 98%	Worldwide: < 1 in 3,900
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
Ehlers-Danlos Syndrome, ADAMTS2-related (*ADAMTS2*) NM_014244:1-22 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 250 Other Populations: < 1 in 500	Ashkenazi Jewish: 92% Other Populations: 92%	Ashkenazi Jewish: < 1 in 2,900 Other Populations: < 1 in 5,900
Enhanced S-cone Syndrome (*NR2E3*) NM_014249:1-9 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
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.

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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
Factor XI Deficiency (*F11*) NM_000128:2-15 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 10 Middle East: 1 in 110 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Middle East: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 870 Middle East: < 1 in 11,000 Other Populations: < 1 in 50,000
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 Hemophagocytic Lymphohistiocytosis, PRF1-related (*PRF1*) NM_001083116:2-3 Inheritance: Autosomal Recessive	Worldwide: 1 in 150	Worldwide: 99%	Worldwide: < 1 in 15,000
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

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Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
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
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
* **Fragile XE Syndrome (*AFF2*)** NM_002025:1-21 Inheritance: X-linked Recessive	Worldwide: 1 in 25,000	Worldwide: 31%	Worldwide: 1 in 36,000
Fraser Syndrome, GRIP1-related (*GRIP1*) NM_021150:1-24 Inheritance: Autosomal Recessive	Worldwide: 1 in 83	Worldwide: 98%	Worldwide: < 1 in 4,000
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

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

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Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
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
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 (*GBA1*) 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
* **Glucose-6-phosphate Dehydrogenase Deficiency (*G6PD*)** V68M, S188F, R459P, R459L, A335T, G163S, V291M Inheritance: X-linked Recessive	African American: 1 in 7 Eastern Asia: 1 in 11 Hispanic: 1 in 15 Middle East: 1 in 9 Northwestern Europe: 1 in 14 South Asia: 1 in 11 Southeast Asia: 1 in 11 Southern Europe: 1 in 14 Other Populations: 1 in 11	African American: 90% Eastern Asia: 30% Hispanic: 90% Middle East: 75% Northwestern Europe: 50% South Asia: 50% Southeast Asia: 80% Southern Europe: 50% Other Populations: 50%	African American: 1 in 61 Eastern Asia: 1 in 15 Hispanic: 1 in 140 Middle East: 1 in 31 Northwestern Europe: 1 in 26 South Asia: 1 in 21 Southeast Asia: 1 in 51 Southern Europe: 1 in 26 Other Populations: 1 in 20
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

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Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
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
Glycogen Storage Disease, GBE1-related (*GBE1*) NM_000158:1-16 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 36 Other Populations: 1 in 420	Ashkenazi Jewish: 99% Other Populations: 97%	Ashkenazi Jewish: < 1 in 3,500 Other Populations: < 1 in 12,000
Glycogen Storage Disease, PFKM-related (*PFKM*) NM_001166686:2-25 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 250 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 25,000 Other Populations: < 1 in 50,000
Glycogen Storage Disease, PYGM-related (*PYGM*) NM_005609:1-20 Inheritance: Autosomal Recessive	Worldwide: 1 in 110	Worldwide: 99%	Worldwide: < 1 in 11,000
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
Hearing Loss and Deafness, LOXHD1-related (*LOXHD1*) NM_144612:1-40 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 180 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 18,000 Other Populations: < 1 in 50,000

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Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
* **Hemophilia B (*F9*)** NM_000133:1-8 Inheritance: X-linked Recessive	Finland: 1 in 13,000 Northwestern Europe: 1 in 13,000 Other Populations: 1 in 13,000	Finland: 96% Northwestern Europe: 96% Other Populations: 97%	Finland: 1 in 350,000 Northwestern Europe: 1 in 350,000 Other Populations: 1 in 490,000
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
Hermansky-Pudlak Syndrome, HPS1-related (*HPS1*) NM_000195:3-20 Inheritance: Autosomal Recessive	Worldwide: 1 in 59	Worldwide: 95%	Worldwide: < 1 in 1,100
Hermansky-Pudlak Syndrome, HPS3-related (*HPS3*) NM_032383:1-17 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 240 Hispanic: 1 in 86 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Hispanic: 94% Other Populations: 99%	Ashkenazi Jewish: < 1 in 23,000 Hispanic: < 1 in 1,400 Other Populations: < 1 in 50,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) (*HEXA*) NM_000520:1-14 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 26 French Canadian/Cajun: 1 in 62 Northwestern Europe: 1 in 230 Other Populations: 1 in 280	Ashkenazi Jewish: 99% French Canadian/Cajun: 99% Northwestern Europe: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 2,500 French Canadian/Cajun: < 1 in 6,100 Northwestern Europe: < 1 in 23,000 Other Populations: < 1 in 28,000
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
Homocystinuria, MTHFR-related (*MTHFR*) NM_005957:2-12 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
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
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

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Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
Inherited Retinal Dystrophy, RPE65-related (*RPE65*) NM_000329:1-14 Inheritance: Autosomal Recessive	Worldwide: 1 in 450	Worldwide: 99%	Worldwide: < 1 in 45,000
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
Joubert Syndrome, AHI1-related (*AHI1*) NM_001134831:4-29 Inheritance: Autosomal Recessive	Worldwide: 1 in 100	Worldwide: 98%	Worldwide: < 1 in 4,500
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
* **L1 Syndrome (*L1CAM*)** NM_001278116:2-29 Inheritance: X-linked Recessive	Worldwide: 1 in 15,000	Worldwide: 98%	Worldwide: 1 in 640,000
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

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Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
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 Northwestern Europe: 1 in 200 Southern Europe: 1 in 150 Other Populations: < 1 in 500	African American: 99% Ashkenazi Jewish: 99% French Canadian/Cajun: 99% Hispanic: 99% Middle East: 99% Native American: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 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 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

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Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
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
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
Mevalonate Kinase Deficiency (*MVK*) NM_000431:2-11 Inheritance: Autosomal Recessive	Worldwide: 1 in 170	Worldwide: 99%	Worldwide: < 1 in 15,000
Microcephaly with Seizures and Brain Atrophy, MED17-related (*MED17*) L371P Inheritance: Autosomal Recessive	Eastern Asia: < 1 in 500 Other Populations: < 1 in 500	Eastern Asia: < 1 in 1,000,000 Other Populations: 89%	Eastern Asia: < 1 in 500 Other Populations: < 1 in 4,500

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Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
Microphthalmia, Anophthalmia, and Coloboma, VSX2-related (*VSX2*) NM_182894:1-5 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
Mitochondrial Complex I Deficiency, NDUFAF5-related (*NDUFAF5*) G250V Inheritance: Autosomal Recessive	Ashkenazi Jewish: < 1 in 500 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Other Populations: 29%	Ashkenazi Jewish: < 1 in 50,000 Other Populations: < 1 in 700
Mitochondrial Complex I Deficiency, NDUFS4-related (*NDUFS4*) NM_002495:1-5 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 97%	Worldwide: < 1 in 17,000
Mitochondrial Complex I Deficiency, NDUFS6-related (*NDUFS6*) NM_004553:1-4 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 94%	Worldwide: < 1 in 8,000
Mitochondrial Complex IV Deficiency, SCO2-related (*SCO2*) NM_005138:2 Inheritance: Autosomal Recessive	Worldwide: 1 in 150	Worldwide: 99%	Worldwide: < 1 in 15,000
Mitochondrial Neurogastrointestinal Encephalopathy Disease (*TYMP*) NM_001257989:2-10 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
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
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

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Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
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
Multiple Sulfatase Deficiency (*SUMF1*) NM_182760:1-9 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 300 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 30,000 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
Myopathy, Lactic Acidosis, and Sideroblastic Anemia 1 (*PUS1*) NM_025215:1-6 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
Myotonia Congenita (*CLCN1*) NM_000083:1-23 Inheritance: Autosomal Recessive	Middle East: 1 in 180 Other Populations: 1 in 180	Middle East: 94% Other Populations: 98%	Middle East: < 1 in 3,100 Other Populations: < 1 in 11,000
NAGA-related Disorders (*NAGA*) NM_000262:1-9 Inheritance: Autosomal Recessive	Worldwide: 1 in 94	Worldwide: 99%	Worldwide: < 1 in 9,300
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
Neuronal Ceroid Lipofuscinosis, PPT1-related (*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

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Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
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
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
Normophosphatemic Familial Tumoral Calcinosis (*SAMD9*) NM_017654:3 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
Oculocutaneous Albinism, OCA2-related (*OCA2*) NM_000275:2-24 Inheritance: Autosomal Recessive	African American: 1 in 76 Native American: 1 in 76 South Asia: 1 in 76 Other Populations: 1 in 76	African American: 83% Native American: 85% South Asia: 93% Other Populations: 96%	African American: 1 in 430 Native American: < 1 in 500 South Asia: < 1 in 1,000 Other Populations: < 1 in 1,700
Oculocutaneous Albinism, TYR-related (*TYR*) NM_000372:1-5 Inheritance: Autosomal Recessive	South Asia: 1 in 20 Southern Europe: 1 in 20 Other Populations: 1 in 20	South Asia: 93% Southern Europe: 98% Other Populations: 99%	South Asia: 1 in 270 Southern Europe: < 1 in 940 Other Populations: < 1 in 1,600
* **Opitz G/BBB Syndrome, MID1-related (*MID1*)** NM_000381:2-10 Inheritance: X-linked Recessive	Worldwide: 1 in 25,000	Worldwide: 87%	Worldwide: 1 in 190,000
Ornithine Aminotransferase Deficiency (*OAT*) NM_000274:2-10 Inheritance: Autosomal Recessive	Finland: 1 in 130 Other Populations: < 1 in 500	Finland: 99% Other Populations: 99%	Finland: < 1 in 13,000 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

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Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
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
* **PLP1-related Disorders (*PLP1*)** NM_000533:1-7 Inheritance: X-linked Recessive	Worldwide: 1 in 100,000	Worldwide: 32%	Worldwide: 1 in 150,000
POLG-related Disorders (*POLG*) NM_002693:2-23 Inheritance: Autosomal Recessive	Worldwide: 1 in 190	Worldwide: 99%	Worldwide: < 1 in 19,000
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
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
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	African American: 99% Ashkenazi Jewish: 99% Eastern Asia: 99% Finland: 99% French Canadian/Cajun: 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

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Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
	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	Hispanic: 99% Middle East: 99% Native American: 99% Northwestern Europe: 99% South Asia: 99% Southeast Asia: 99% Southern Europe: 99%	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
Pontocerebellar Hypoplasia, RARS2-related (*RARS2*) Q12R, c.110+5A>G, M1? Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 16%	Worldwide: < 1 in 590
Pontocerebellar Hypoplasia, SEPSECS-related (*SEPSECS*) NM_016955:1-11 Inheritance: Autosomal Recessive	Finland: 1 in 270 Middle East: 1 in 44 Other Populations: < 1 in 500	Finland: 99% Middle East: 99% Other Populations: 99%	Finland: < 1 in 27,000 Middle East: < 1 in 4,300 Other Populations: < 1 in 50,000
Pontocerebellar Hypoplasia, VPS53-related (*VPS53*) NM_001128159:1-22 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
Pontocerebellar Hypoplasia, VRK1-related (*VRK1*) NM_003384:2-13 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
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 Ciliary Dyskinesia, DNAH5-related (*DNAH5*) NM_001369:1-79 Inheritance: Autosomal Recessive	Worldwide: 1 in 140	Worldwide: 99%	Worldwide: < 1 in 13,000
Primary Ciliary Dyskinesia, DNAI1-related (*DNAI1*) NM_012144:1-20 Inheritance: Autosomal Recessive	Worldwide: 1 in 230	Worldwide: 99%	Worldwide: < 1 in 23,000
Primary Ciliary Dyskinesia, DNAI2-related (*DNAI2*) NM_023036:2-13 Inheritance: Autosomal Recessive	Worldwide: 1 in 450	Worldwide: 99%	Worldwide: < 1 in 45,000

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Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
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 Southern Europe: 1 in 130 Other Populations: 1 in 200	African American: 99% Ashkenazi Jewish: 99% Finland: 99% French Canadian/Cajun: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 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 Southern Europe: < 1 in 13,000 Other Populations: < 1 in 20,000
Primary Microcephaly, MCPH1-related (*MCPH1*) NM_024596:1-14 Inheritance: Autosomal Recessive	Worldwide: 1 in 150	Worldwide: 88%	Worldwide: < 1 in 1,300
Primary Trimethylaminuria (*FMO3*) NM_001002294:2-9 Inheritance: Autosomal Recessive	Worldwide: 1 in 140	Worldwide: 99%	Worldwide: < 1 in 14,000
Pseudocholinesterase Deficiency (*BCHE*) NM_000055:2-4 Inheritance: Autosomal Recessive	Northwestern Europe: 1 in 26 Southern Europe: 1 in 35 Other Populations: 1 in 32	Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Northwestern Europe: < 1 in 2,500 Southern Europe: < 1 in 3,400 Other Populations: < 1 in 3,100
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
RAPSN-related Disorders (*RAPSN*) NM_005055:1-8 Inheritance: Autosomal Recessive	Northwestern Europe: 1 in 320 Other Populations: 1 in 480	Northwestern Europe: 98% Other Populations: 98%	Northwestern Europe: < 1 in 17,000 Other Populations: < 1 in 26,000
RTEL1-related Disorders (*RTEL1*) NM_001283009:2-34 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
Refsum Disease, PHYH-related (*PHYH*) NM_006214:1-9 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000

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Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
Retinitis Pigmentosa, CERKL-related (*CERKL*) NM_001030311:1-14 Inheritance: Autosomal Recessive	Southern Europe: 1 in 220 Other Populations: 1 in 440	Southern Europe: 99% Other Populations: 99%	Southern Europe: < 1 in 22,000 Other Populations: < 1 in 44,000
Retinitis Pigmentosa, DHDDS-related (*DHDDS*) K42E Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 250 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 25,000 Other Populations: < 1 in 50,000
Retinitis Pigmentosa, EYS-related (*EYS*) NM_001142800:4-43 Inheritance: Autosomal Recessive	Ashkenazi Jewish: < 1 in 500 Middle East: 1 in 170 Northwestern Europe: 1 in 200 Southern Europe: 1 in 200 Other Populations: 1 in 200	Ashkenazi Jewish: 99% Middle East: 96% Northwestern Europe: 98% Southern Europe: 99% Other Populations: 96%	Ashkenazi Jewish: < 1 in 50,000 Middle East: < 1 in 4,500 Northwestern Europe: < 1 in 11,000 Southern Europe: < 1 in 20,000 Other Populations: < 1 in 5,100
Retinitis Pigmentosa, FAM161A-related (*FAM161A*) NM_001201543:1-7 Inheritance: Autosomal Recessive	Middle East: 1 in 130 Northwestern Europe: 1 in 300 Other Populations: 1 in 440	Middle East: 99% Northwestern Europe: 99% Other Populations: 99%	Middle East: < 1 in 13,000 Northwestern Europe: < 1 in 30,000 Other Populations: < 1 in 44,000
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
Serine Deficiency Disorder, PHGDH-related (*PHGDH*) NM_006623:1-12 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
Severe Combined Immunodeficiency, RAG2-related (*RAG2*) NM_001243785:3 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
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

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Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
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
Spastic Paraplegia, TECPR2-related (*TECPR2*) NM_014844:2-20 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 310 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 31,000 Other Populations: < 1 in 50,000
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
Spinocerebellar Ataxia, ANO10-related (*ANO10*) NM_018075:2-13 Inheritance: Autosomal Recessive	Worldwide: 1 in 94	Worldwide: 97%	Worldwide: < 1 in 2,800
Spondylothoracic Dysostosis (*MESP2*) NM_001039958:1-2 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
Surfactant Deficiency, ABCA3-related (*ABCA3*) NM_001089:4-33 Inheritance: Autosomal Recessive	Worldwide: 1 in 120	Worldwide: 99%	Worldwide: < 1 in 12,000
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

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Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
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
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
VPS13A Disease (*VPS13A*) NM_033305:1-72 Inheritance: Autosomal Recessive	Eastern Asia: < 1 in 500 Other Populations: < 1 in 500	Eastern Asia: 97% Other Populations: 99%	Eastern Asia: < 1 in 16,000 Other Populations: < 1 in 40,000
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
Vitamin D–dependent Rickets, CYP27B1-related (*CYP27B1*) NM_000785:1-9 Inheritance: Autosomal Recessive	Worldwide: 1 in 180	Worldwide: 99%	Worldwide: < 1 in 18,000
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

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Disease (*gene*)	Carrier Frequency	Detection Rate	Residual Carrier Risk
* **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 19,000 Northwestern Europe: 1 in 16,000 Southern Europe: 1 in 14,000 Other Populations: 1 in 8,400	Eastern Asia: 77% Northwestern Europe: 77% Southern Europe: 77% Other Populations: 77%	Eastern Asia: 1 in 81,000 Northwestern Europe: 1 in 71,000 Southern Europe: 1 in 60,000 Other Populations: 1 in 36,000
* **X-linked Choroideremia (*CHM*)** NM_000390:1-15 Inheritance: X-linked Recessive	Worldwide: 1 in 25,000	Worldwide: 97%	Worldwide: 1 in 780,000
* **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 Retinal Dystrophy, RPGR-related (*RPGR*)** NM_000328:1-19 Inheritance: X-linked Recessive	Worldwide: 1 in 17,000	Worldwide: 38%	Worldwide: 1 in 28,000
* **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

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