

Residual risks by ethnicity

This table displays residual risks after a negative result for each of the genes and corresponding disorders. If a patient is reported to be a carrier of a disease, their residual risk is 1 and this table does not apply for that disease.

Disease (Inheritance)	Gene	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk	Analytical Detection Rate
Abetalipoproteinemia (AR) NM_000253.3	<i>MTTP</i>	Caucasian	< 1 in 500	81%	1 in 2627	>95%
		Ashkenazi Jewish	1 in 186	>95%	1 in 3701	>95%
		Worldwide	< 1 in 500	70%	1 in 1664	95%
Achromatopsia (AR) NM_019098.4	<i>CNGB3</i>	Caucasian	1 in 91	85%	1 in 601	>95%
		Worldwide	1 in 98	>95%	1 in 1941	>95%
Acrodermatitis Enteropathica (AR) NM_130849.3	<i>SLC39A4</i>	Worldwide	1 in 354	75%	1 in 1413	>95%
Acute Infantile Liver Failure (AR) NM_018006.4	<i>TRMU</i>	Worldwide	< 1 in 500	87%	1 in 3839	>95%
		Sephardic Jewish (Yemenite)	1 in 34	81%	1 in 175	94%
Acyl-CoA Oxidase I Deficiency (AR) NM_004035.6	<i>ACOX1</i>	Worldwide	< 1 in 500	80%	1 in 2496	91%
Adenosine Deaminase Deficiency (AR) NM_000022.2	<i>ADA</i>	Worldwide	1 in 337	73%	1 in 1245	>95%
Adrenoleukodystrophy, X-Linked (XL) NM_000033.3 <i>Exception: Exons 8 and 9</i>	<i>ABCD1</i>	Worldwide	< 1 in 500	47%	1 in 943	85%
		Sephardic Jewish	< 1 in 500	73%	1 in 1849	>95%
Aicardi-Goutières Syndrome (SAMHD1-Related) (AR) NM_015474.3	<i>SAMHD1</i>	Worldwide	< 1 in 500	84%	1 in 3120	93%
Alpha-Mannosidosis (AR) NM_000528.3	<i>MAN2B1</i>	Caucasian	1 in 485	85%	1 in 3228	94%
		Worldwide	< 1 in 500	81%	1 in 2627	>95%
Alpha-Thalassemia (AR) NM_000558.4 / NM_000517.4 <i>MLPA only (Copy-number analysis)</i>	<i>HBA1 / HBA2</i>	Caucasian	1 in 500	90%	1 in 4991	90%
		African American	1 in 30	90%	1 in 291	90%
		Asian	1 in 20	90%	1 in 191	90%
		Worldwide	1 in 25	90%	1 in 241	90%
Alpha-Thalassemia Mental Retardation Syndrome (XL) NM_000489.4	<i>ATRX</i>	Worldwide	< 1 in 500	56%	1 in 1135	95%
Alport Syndrome (COL4A3-Related) (AR) NM_000091.4	<i>COL4A3</i>	Caucasian	1 in 284	86%	1 in 2022	95%
		Ashkenazi Jewish	1 in 192	>95%	1 in 3821	>95%
		Worldwide	1 in 354	87%	1 in 2716	>95%
Alport Syndrome (COL4A4-Related) (AR) NM_000092.4	<i>COL4A4</i>	Worldwide	1 in 353	88%	1 in 2934	>95%
Alport Syndrome (COL4A5-Related) (XL) NM_000495.3	<i>COL4A5</i>	Worldwide	< 1 in 500	83%	1 in 2936	88%

Alstrom Syndrome (AR) NM_015120.4	<i>ALMS1</i>	Worldwide	1 in 500	79%	1 in 2377	>95%
Andermann Syndrome (AR) NM_133647.1	<i>SLC12A6</i>	Worldwide French Canadian (Saguenay Lac-St. Jean)	< 1 in 500 1 in 23	>95% >95%	1 in 9981 1 in 441	>95% >95%
Argininosuccinic Aciduria (AR) NM_000048.3	<i>ASL</i>	Worldwide	in 274	62%	1 in 719	>95%
Aromatase Deficiency (AR) NM_031226.2	<i>CYP19A1</i>	Worldwide	< 1 in 500	72%	1 in 1783	81%
Arthrogryposis, Mental Retardation, and Seizures (AR) NM_012243.2	<i>SLC35A3</i>	Ashkenazi Jewish Worldwide	1 in 453 < 1 in 500	>95% >95%	1 in 9041 1 in 9981	>95% >95%
Asparagine Synthetase Deficiency (AR) NM_001673.4	<i>ASNS</i>	Worldwide Sephardic Jewish (Iranian)	< 1 in 500 1 in 80	>95% >95%	1 in 9981 1 in 1581	>95% >95%
Aspartylglycosaminuria (AR) NM_000027.3	<i>AGA</i>	Caucasian Worldwide Finnish	< 1 in 500 < 1 in 500 1 in 63	>95% >95% >95%	1 in 9981 1 in 9981 1 in 1241	>95% >95% >95%
Ataxia With Isolated Vitamin E Deficiency (AR) NM_000370.3	<i>TTPA</i>	Caucasian Worldwide	< 1 in 500 < 1 in 500	88% 93%	1 in 3565 1 in 7130	90% >95%
Ataxia Telangiectasia (AR) NM_000051.3	<i>ATM</i>	Ashkenazi Jewish Worldwide Sephardic Jewish (Moroccan)	< 1 in 500 1 in 100 1 in 69	>95% 75% >95%	1 in 9981 1 in 397 1 in 1361	>95% 91% >95%
Autosomal Recessive Spastic Ataxia Of Charlevoix-Saguenay (AR) NM_014363.5	<i>SACS</i>	Caucasian Worldwide French Canadian (Charlevoix-Saguenay)	1 in 450 < 1 in 500 1 in 21	70% 86% >95%	1 in 1498 1 in 3565 1 in 401	>95% >95% >95%
Bardet-Biedl Syndrome (BBS1-Related) (AR) NM_024649.4	<i>BBS1</i>	Worldwide Faroese	1 in 392 1 in 30	93% >95%	1 in 5587 1 in 581	>95% >95%
Bardet-Biedl Syndrome (BBS2-Related) (AR) NM_031885.3	<i>BBS2</i>	Ashkenazi Jewish Worldwide Hutterite	1 in 140 < 1 in 500 1 in 22	>95% 69% >95%	1 in 2781 1 in 1611 1 in 421	>95% >95% >95%
Bardet-Biedl Syndrome (BBS10-Related) (AR) NM_024685.3	<i>BBS10</i>	Worldwide	1 in 423	78%	1 in 1919	>95%
Bardet-Biedl Syndrome (BBS12-Related) (AR) NM_152618.2	<i>BBS12</i>	Worldwide	< 1 in 500	77%	1 in 2171	>95%
Bare Lymphocyte Syndrome, Type II (AR) NM_000246.3	<i>CIITA</i>	Worldwide	< 1 in 500	95%	1 in 9981	>95%
Bartter Syndrome, Type 4A (AR) NM_057176.2	<i>BSND</i>	Worldwide	< 1 in 500	94%	1 in 8318	>95%
Bernard-Soulier Syndrome, Type A1 (AR) NM_000173.5	<i>GP1BA</i>	Worldwide	< 1 in 500	84%	1 in 3120	>95%

Bernard-Soulier Syndrome, Type C (AR) NM_000174.4	<i>GP9</i>	Worldwide	< 1 in 500	83%	1 in 2936	>95%
Beta-Globin Related Hemoglobinopathies:	<i>HBB</i>	Caucasian	1 in 373	>95%	1 in 7441	>95%
Beta- Thalassemia (AR) NM_000518.4		African American	1 in 100	>95%	1 in 1981	>95%
		Asian	1 in 54	>95%	1 in 1061	>95%
		Worldwide	1 in 158	>95%	1 in 3141	>95%
		Mediterranean	1 in 28	>95%	1 in 541	>95%
Beta-Globin Related Hemoglobinopathies:	<i>HBB</i>	Caucasian	< 1 in 500	>99%	1 in 49901	
HbC Variant (AR) NM_000518.4		African American	1 in 35	>99%	1 in 3401	
		Hispanic	< 1 in 500	>99%	1 in 49901	>99%
		Asian	< 1 in 500	>99%	1 in 49901	
<i>Variant Tested: c.19G>A, p.E7K</i>		Worldwide	< 1 in 500	>99%	1 in 49901	
Beta-Globin Related Hemoglobinopathies:	<i>HBB</i>	Caucasian	< 1 in 500	>99%	1 in 49901	
HbS Variant (Sickle Cell Disease) (AR) NM_000518.4		African American	1 in 12	>99%	1 in 1101	
		Hispanic	1 in 17	>99%	1 in 1601	>99%
		Asian	< 1 in 500	>99%	1 in 49901	
<i>Variant Tested: c.20A>T, p.E7V</i>		Worldwide	1 in 71	>99%	1 in 7001	
3-Beta-Hydroxysteroid Deficiency (AR) NM_000198.3	<i>HSD3B2</i>	Worldwide	< 1 in 500	58%	1 in 1189	>95%
Beta-Ketothiolase Deficiency (AR) NM_000019.3	<i>ACAT1</i>	Caucasian	1 in 354	46%	1 in 655	73%
		Asian	1 in 289	67%	1 in 874	>95%
		Worldwide	1 in 347	60%	1 in 866	>95%
Bilateral Frontoparietal Polymicrogyria (AR) NM_005682.6	<i>GPR56</i> (<i>ADGRG1</i>)	Worldwide	< 1 in 500	80%	1 in 2496	>95%
Biotinidase Deficiency (AR) NM_000060.3	<i>BTBD</i>	Caucasian	1 in 12	80%	1 in 56	86%
		Hispanic	1 in 30	72%	1 in 105	>95%
		Worldwide	1 in 25	74%	1 in 93	>95%
Bloom Syndrome (AR) NM_000057.2	<i>BLM</i>	Ashkenazi Jewish	1 in 134	>95%	1 in 2661	>95%
		Worldwide	< 1 in 500	88%	1 in 4159	>95%
Canavan Disease (AR) NM_000049.2	<i>ASPA</i>	Ashkenazi Jewish	1 in 55	>95%	1 in 1081	>95%
		Worldwide	1 in 158	82%	1 in 873	95%
Carbamoylphosphate Synthetase I Deficiency (AR) NM_001875.4	<i>CPS1</i>	Caucasian	1 in 284	25%	1 in 378	86%
		Asian	1 in 447	46%	1 in 827	84%
		Worldwide	< 1 in 500	54%	1 in 1086	>95%
Carnitine Palmitoyltransferase IA Deficiency (AR) NM_001876.3	<i>CPT1A</i>	Worldwide	< 1 in 500	56%	1 in 1135	>95%
		Hutterite	1 in 16	>95%	1 in 301	>95%
Carnitine Palmitoyltransferase II Deficiency (AR) NM_000098.2	<i>CPT2</i>	Caucasian	1 in 200	91%	1 in 2212	>95%
		African	1 in 308	75%	1 in 1229	>95%
		Asian	< 1 in 500	76%	1 in 2080	>95%
		Ashkenazi Jewish	1 in 45	>95%	1 in 881	>95%
		Worldwide	1 in 182	90%	1 in 1811	>95%

Carpenter Syndrome (AR) NM_001278667.1	<i>RAB23</i>	Caucasian	< 1 in 500	95%	1 in 9981	>95%
		Worldwide	< 1 in 500	84%	1 in 3120	>95%
Cartilage-Hair Hypoplasia (AR) NR_003051.3	<i>RMRP</i>	Worldwide	< 1 in 500	50%	1 in 999	>95%
		Amish	1 in 19	>95%	1 in 361	>95%
		Finnish	1 in 76	>95%	1 in 1501	>95%
Cerebral Creatine Deficiency Syndrome 1 (XL) NM_005629.3 <i>Exception: Exon 3</i>	<i>SLC6A8</i>	Worldwide	< 1 in 500	76%	1 in 2080	94%
Cerebral Creatine Deficiency Syndrome 2 (AR) NM_000156.5	<i>GAMT</i>	Worldwide	< 1 in 500	71%	1 in 1722	>95%
		Portuguese	1 in 125	>95%	1 in 2481	>95%
Cerebrotendinous Xanthomatosis (AR) NM_000784.3	<i>CYP27A1</i>	Worldwide	1 in 112	71%	1 in 384	>95%
		Sephardic Jewish (Moroccan)	1 in 76	>95%	1 in 1501	>95%
Charcot-Marie-Tooth Disease, Type 4D (AR) NM_001135242.1	<i>NDRG1</i>	Roma	1 in 22	>95%	1 in 421	>95%
Charcot-Marie-Tooth Disease, Type 5 / Arts Syndrome (XL) NM_002764.3	<i>PRPS1</i>	Worldwide	< 1 in 500	>95%	1 in 9981	>95%
Charcot-Marie-Tooth Disease, X-Linked (XL) NM_000166.5	<i>GJB1</i>	Worldwide	< 1 in 500	53%	1 in 1063	>95%
Choreoacanthocytosis (AR) NM_033305.2	<i>VPS13A</i>	Worldwide	< 1 in 500	88%	1 in 4159	95%
		Ashkenazi Jewish	N/A	>95%	N/A	>95%
Choroideremia (XL) NM_000390.2	<i>CHM</i>	Worldwide	< 1 in 500	92%	1 in 6239	94%
Chronic Granulomatous Disease (CYBA-Related) (AR) NM_000101.2	<i>CYBA</i>	Worldwide	< 1 in 500	57%	1 in 1161	85%
		Sephardic Jewish (Moroccan)	1 in 13	83%	1 in 72	>95%
Chronic Granulomatous Disease (CYBB-Related) (XL) NM_000397.3	<i>CYBB</i>	Worldwide	< 1 in 500	83%	1 in 2936	94%
Citrin Deficiency (AR) NM_014251.2	<i>SLC25A13</i>	Caucasian	< 1 in 500	92%	1 in 6239	>95%
		Asian	1 in 123	95%	1 in 2441	>95%
		Worldwide	< 1 in 500	89%	1 in 4537	>95%
Citrullinemia, Type I (AR) NM_000050.4	<i>ASS1</i>	Caucasian	1 in 195	53%	1 in 414	>95%
		Asian	1 in 123	78%	1 in 556	91%
		Worldwide	1 in 119	84%	1 in 739	>95%
Cohen Syndrome (AR) NM_017890.4	<i>VPS13B</i>	Worldwide	1 in 500	85%	1 in 3328	90%
Combined Malonic and Methylmalonic Aciduria (AR) NM_001127214.3	<i>ACSF3</i>	Worldwide	1 in 86	>95%	1 in 1701	>95%

Combined Oxidative Phosphorylation Deficiency 1 (AR) NM_024996.5	<i>GFM1</i>	Worldwide	< 1 in 500	>95%	1 in 9981	>95%
Combined Oxidative Phosphorylation Deficiency 3 (AR) NM_001172696.1	<i>TSMF</i>	Finnish Worldwide	1 in 80 < 1 in 500	83% >95%	1 in 466 1 in 9981	>95% >95%
Combined Pituitary Hormone Deficiency 2 (AR) NM_006261.4	<i>PROP1</i>	Worldwide	1 in 141	>95%	1 in 2801	>95%
Combined Pituitary Hormone Deficiency 3 (AR) NM_014564.3	<i>LHX3</i>	Worldwide	< 1 in 500	88%	1 in 4159	92%
Combined SAP Deficiency (AR) NM_002778.2	<i>PSAP</i>	Worldwide	< 1 in 500	89%	1 in 4537	>95%
Congenital Adrenal Hyperplasia due to 17-Alpha-Hydroxylase Deficiency (AR) NM_000102.3	<i>CYP17A1</i>	Worldwide	< 1 in 500	57%	1 in 1161	>95%
Congenital Amegakaryocytic Thrombocytopenia (AR) NM_005373.2	<i>MPL</i>	Caucasian Ashkenazi Jewish Worldwide	1 in 266 1 in 57 1 in 415	69% >95% 79%	1 in 856 1 in 1121 1 in 1972	>95% >95% >95%
Congenital Disorder of Glycosylation, Type Ia (AR) NM_000303.2	<i>PMM2</i>	Caucasian Asian Ashkenazi Jewish Worldwide	1 in 42 1 in 449 1 in 61 1 in 124	87% 45% >95% 84%	1 in 316 1 in 816 1 in 1201 1 in 770	>95% >95% >95% >95%
Congenital Disorder of Glycosylation, Type Ib (AR) NM_002435.2	<i>MPI</i>	Worldwide	< 1 in 500	74%	1 in 1920	>95%
Congenital Disorder of Glycosylation, Type Ic (AR) NM_013339.3	<i>ALG6</i>	Worldwide	< 1 in 500	87%	1 in 3839	>95%
Congenital Insensitivity to Pain with Anhidrosis (AR) NM_001012331.1	<i>NTRK1</i>	Asian Worldwide Sephardic Jewish (Moroccan)	1 in 387 < 1 in 500 N/A	91% 91% >95%	1 in 4290 1 in 5545 N/A	>95% >95% >95%
Congenital Myasthenic Syndrome (CHRNA-Related) (AR) NM_000080.3	<i>CHRNE</i>	Caucasian Worldwide Southeastern European Roma	1 in 383 1 in 408 1 in 25	65% 85% >95%	1 in 1092 1 in 2714 1 in 481	>95% >95% >95%
Congenital Myasthenic Syndrome (RAPSN-Related) (AR) NM_005055.4	<i>RAPSN</i>	Caucasian Worldwide Sephardic Jewish (Iraqi and Iranian)	1 in 176 1 in 252 N/A	90% 86% >95%	1 in 1751 1 in 1794 N/A	>95% >95% >95%
Congenital Neutropenia (HAX1-Related) (AR) NM_006118.3	<i>HAX1</i>	Worldwide	< 1 in 500	90%	1 in 4991	95%

Congenital Neutropenia (VPS45-Related) (AR) NM_001279354.1	<i>VPS45</i>	Worldwide	< 1 in 500	>95%	1 in 9981	>95%
Corneal Dystrophy and Perceptive Deafness (AR) NM_032034.3	<i>SLC4A11</i>	Worldwide	< 1 in 500	60%	1 in 1249	>95%
Corticosterone Methyloxidase Deficiency (AR) NM_000498.3 <i>Exception: Exons 3 - 7</i>	<i>CYP11B2</i>	Worldwide Sephardic Jewish (Iranian)	< 1 in 500 1 in 30	68% >95%	1 in 1560 1 in 581	72% >95%
Cystic Fibrosis (AR) NM_000492.3 <i>Exception: Exon 10</i>	<i>CFTR</i>	Caucasian African American Hispanic Asian Ashkenazi Jewish Worldwide	1 in 25 1 in 61 1 in 58 1 in 94 1 in 24 1 in 45	94% 87% 87% 65% >95% 88%	1 in 401 1 in 463 1 in 439 1 in 267 1 in 461 1 in 368	>95% >95% >95% >95% >95% >95%
Cystinosis (AR) NM_004937.2	<i>CTNS</i>	Caucasian African Hispanic Asian Worldwide French Canadian (Saguenay-Lac St. Jean) Sephardic Jewish (Moroccan)	1 in 220 < 1 in 500 < 1 in 500 < 1 in 500 1 in 224 1 in 39 1 in 100	81% 83% 64% 90% 86% 90% 92%	1 in 1154 1 in 2936 1 in 1387 1 in 4991 1 in 1594 1 in 381 1 in 1239	90% >95% 75% >95% >95% 90% >95%
D-Bifunctional Protein Deficiency (AR) NM_000414.3	<i>HSD17B4</i>	Worldwide	< 1 in 500	64%	1 in 1387	87%
Deafness, Autosomal Recessive 77 (AR) NM_144612.6	<i>LOXHD1</i>	Ashkenazi Jewish Worldwide	1 in 180 < 1 in 500	>95% >95%	1 in 3581 1 in 9981	>95% >95%
Duchenne Muscular Dystrophy/ Becker Muscular Dystrophy (XL) NM_004006.2	<i>DMD</i>	Worldwide	< 1 in 500	90%	1 in 4991	>95%
Dyskeratosis Congenita (RTEL1-Related) (AR) NM_001283009.1	<i>RTEL1</i>	Ashkenazi Jewish Worldwide	1 in 165 < 1 in 500	>95% 78%	1 in 3281 1 in 2269	>95% >95%
Dystrophic Epidermolysis Bullosa (AR) NM_000094.3	<i>COL7A1</i>	Worldwide	1 in 370	85%	1 in 2461	>95%
Ehlers-Danlos Syndrome, Type VIIC (AR) NM_014244.4	<i>ADAMTS2</i>	Ashkenazi Jewish Worldwide	1 in 187 < 1 in 500	>95% 91%	1 in 3721 1 in 5545	>95% 93%
Ellis-van Creveld Syndrome (EVC-Related) (AR) NM_153717.2 <i>Exception: Exon 1</i>	<i>EVC</i>	Worldwide Lancaster County Amish	1 in 345 1 in 12	72% >95%	1 in 1230 1 in 221	90% >95%
Emery-Dreifuss Myopathy 1 (XL) NM_000117.2	<i>EMD</i>	Worldwide	< 1 in 500	94%	1 in 8318	>95%

Enhanced S-Cone Syndrome (AR) NM_014249.3	<i>NR2E3</i>	Ashkenazi Jewish Worldwide	N/A 1 in 204	>95% 82%	N/A 1 in 1129	>95% >95%
Ethylmalonic Encephalopathy (AR) NM_014297.3	<i>ETHE1</i>	Worldwide	< 1 in 500	72%	1 in 1783	94%
Fabry Disease (XL) NM_000169.2	<i>GLA</i>	Worldwide	< 1 in 500	74%	1 in 1920	>95%
Factor IX Deficiency (XL) NM_000133.3	<i>F9</i>	Worldwide	< 1 in 500	59%	1 in 1218	95%
Factor XI Deficiency (AR) NM_000128.3	<i>F11</i>	Caucasian Asian Ashkenazi Jewish Worldwide	1 in 101 1 in 163 1 in 11 1 in 92	36% 65% >95% 51%	1 in 157 1 in 464 1 in 201 1 in 187	>95% >95% >95% >95%
Familial Dysautonomia (AR) NM_003640.3	<i>IKBKAP</i>	Ashkenazi Jewish Worldwide	1 in 31 < 1 in 500	>95% >95%	1 in 601 1 in 9981	>95% >95%
Familial Hypercholesterolemia (AR) NM_000527.4	<i>LDLR</i>	Caucasian Ashkenazi Jewish Worldwide French Canadian Finnish South African Afrikaner	1 in 200 1 in 69 < 1 in 500 1 in 267 1 in 143 1 in 70	15% 35% 41% 17% 93% 94%	1 in 235 1 in 106 1 in 847 1 in 321 1 in 2030 1 in 1151	85% 85% 86% 26% >95% >95%
Familial Hypercholesterolemia, Autosomal Recessive (AR) NM_015627.2	<i>LDLRAP1</i>	Worldwide Sardinian	< 1 in 500 1 in 143	88% >95%	1 in 4159 1 in 2841	95% >95%
Familial Hyperinsulinism (ABCC8-Related) (AR) NM_000352.4	<i>ABCC8</i>	Ashkenazi Jewish Worldwide Finnish	1 in 52 1 in 167 1 in 100	>95% 56% 50%	1 in 1021 1 in 378 1 in 199	>95% >95% >95%
Familial Hyperinsulinism (KCNJ11-Related) (AR) NM_000525.3	<i>KCNJ11</i>	Worldwide	1 in 500	26%	1 in 675	>95%
Familial Mediterranean Fever (AR) NM_000243.2	<i>MEFV</i>	Ashkenazi Jewish Worldwide Sepharic Jewish Armenian Turkish	1 in 13 1 in 115 1 in 14 1 in 5 1 in 5	>95% 87% >95% >95% 75%	1 in 241 1 in 878 1 in 261 1 in 81 1 in 17	>95% >95% >95% >95% >95%
Fanconi Anemia, Group A (AR) NM_000135.2	<i>FANCA</i>	Worldwide Spanish Roma Sephardic Jewish (Moroccan and Tunisian)	1 in 345 1 in 64 1 in 133	60% >95% 86%	1 in 861 1 in 1261 1 in 944	76% >95% >95%
Fanconi Anemia, Group C (AR) NM_000136.2	<i>FANCC</i>	Ashkenazi Jewish Worldwide	1 in 89 1 in 417	>95% >95%	1 in 1761 1 in 8321	>95% >95%
Fanconi Anemia, Group G (AR) NM_004629.1	<i>FANCG</i>	African American Worldwide	1 in 100 < 1 in 500	80% 92%	1 in 496 1 in 6239	>95% >95%

Fumarase Deficiency (AR) NM_000143.3	<i>FH</i>	Worldwide	< 1 in 500	49%	1 in 979	95%
Galactokinase Deficiency (AR) NM_000154.1	<i>GALK1</i>	Asian	1 in 500	60%	1 in 1249	90%
		Worldwide	1 in 122	75%	1 in 485	>95%
		Roma	1 in 47	>95%	1 in 921	>95%
Galactosemia (AR) NM_000155.3	<i>GALT</i>	Caucasian	1 in 152	91%	1 in 1679	>95%
		African	1 in 87	>95%	1 in 1721	>95%
		Hispanic	1 in 305	>95%	1 in 6081	>95%
		Ashkenazi Jewish	1 in 156	>95%	1 in 3101	>95%
		Worldwide	1 in 112	88%	1 in 926	>95%
		Irish Travellers	1 in 11	>95%	1 in 201	>95%
Gaucher Disease (AR) NM_000157.3	<i>GBA</i>	Caucasian	1 in 164	67%	1 in 495	67%
		Ashkenazi Jewish	1 in 15	>95%	1 in 281	>95%
		Worldwide	1 in 158	56%	1 in 358	56%
<i>Variants tested: p.L29fs, c.115+1G>A, p.N409S, p.L422fs, p.V433L, p.D448H, p.D448V, p.L483P, p.R502C, p.R502H, p.R535C (Genotyping only)</i>						
Gitelman Syndrome (AR) NM_000339.2	<i>SLC12A3</i>	Worldwide	1 in 100	51%	1 in 203	>95%
Glutaric Acidemia, Type I (AR) NM_000159.3	<i>GCDH</i>	Caucasian	1 in 172	77%	1 in 744	>95%
		African	1 in 36	>95%	1 in 701	>95%
		Worldwide	1 in 158	70%	1 in 524	>95%
		Oji-Cree First Nations (N. Manitoba)	1 in 8	>95%	1 in 141	>95%
		Old Order Amish of Pennsylvania	1 in 11	>95%	1 in 201	>95%
		Lumbee Native American	1 in 16	>95%	1 in 301	>95%
Glutaric Acidemia, Type IIa (AR) NM_000126.3	<i>ETFA</i>	Worldwide	< 1 in 500	63%	1 in 1350	95%
Glutaric Acidemia, Type IIc (AR) NM_004453.3	<i>ETFDH</i>	Asian	1 in 87	64%	1 in 240	94%
		Worldwide	1 in 250	55%	1 in 554	>95%
Glycine Encephalopathy (AMT-Related) (AR) NM_000481.3	<i>AMT</i>	Caucasian	1 in 271	67%	1 in 819	94%
		Worldwide	1 in 319	74%	1 in 1224	>95%
Glycine Encephalopathy (GLDC-Related) (AR) NM_000170.2	<i>GLDC</i>	Caucasian	1 in 140	48%	1 in 268	66%
		Worldwide	1 in 165	41%	1 in 279	83%
<i>Exception: Exon 1</i>						
Glycogen Storage Disease, Type Ia (AR) NM_000151.3	<i>G6PC</i>	Caucasian	1 in 177	88%	1 in 1468	91%
		Asian	1 in 192	>95%	1 in 3821	>95%
		Ashkenazi Jewish	1 in 71	>95%	1 in 1401	>95%
		Worldwide	1 in 261	95%	1 in 5201	>95%
Glycogen Storage Disease, Type Ib (AR) NM_001164277.1	<i>SLC37A4</i>	Caucasian	< 1 in 500	94%	1 in 8318	>95%
		Worldwide	1 in 354	78%	1 in 1606	>95%
Glycogen Storage Disease, Type II (AR) NM_000152.3	<i>GAA</i>	Caucasian	1 in 100	68%	1 in 310	89%
		African	1 in 70	83%	1 in 407	>95%
		Asian	1 in 112	64%	1 in 309	>95%
		Ashkenazi Jewish	1 in 58	>95%	1 in 1141	>95%
		Worldwide	1 in 132	73%	1 in 486	>95%

Glycogen Storage Disease, Type III (AR) NM_000028.2	AGL	Worldwide	1 in 158	95%	1 in 3141	>95%
		Sephardic Jewish (Moroccan)	1 in 34	>95%	1 in 661	>95%
		Faroese	1 in 28	>95%	1 in 541	>95%
Glycogen Storage Disease, Type IV / Adult Polyglucosan Body Disease (AR) NM_000158.3	GBE1	Caucasian	1 in 144	70%	1 in 478	70%
		Ashkenazi Jewish	1 in 68	>95%	1 in 1341	>95%
		Worldwide	1 in 387	73%	1 in 1431	95%
Glycogen Storage Disease, Type V (AR) NM_005609.2	PYGM	Caucasian	1 in 191	76%	1 in 793	94%
		Sephardic Jewish (Kurdish)	1 in 84	>95%	1 in 1661	>95%
Glycogen Storage Disease, Type VII (AR) NM_000289.5	PFKM	Ashkenazi Jewish	1 in 250	>95%	1 in 4981	>95%
		Worldwide	< 1 in 500	74%	1 in 1920	>95%
GRACILE Syndrome and Other BCS1L-Related Disorders (AR) NM_001257342.1	BCS1L	Caucasian	1 in 407	64%	1 in 1129	>95%
		Worldwide	< 1 in 500	95%	1 in 9981	>95%
		Finnish	1 in 108	>95%	1 in 2141	>95%
Hemochromatosis, Type 2A (AR) NM_213653.3	HFE2	Caucasian	< 1 in 500	84%	1 in 3120	>95%
		Worldwide	< 1 in 500	79%	1 in 2377	>95%
Hemochromatosis, Type 3 (AR) NM_003227.3	TFR2	Worldwide	< 1 in 500	73%	1 in 1849	>95%
Hereditary Fructose Intolerance (AR) NM_000035.3	ALDOB	Caucasian	1 in 80	95%	1 in 1581	>95%
		African	1 in 406	>95%	1 in 8101	>95%
		Hispanic	1 in 275	94%	1 in 4568	>95%
		Worldwide	1 in 121	87%	1 in 924	>95%
Hereditary Spastic Paraparesis 49 (AR) NM_014844.4	TECP2	Sephardic Jewish (Bukharian)	1 in 27	>95%	1 in 521	>95%
Hermansky-Pudlak Syndrome, Type 1 (AR) NM_000195.4	HPS1	Worldwide	< 1 in 500	>95%	1 in 9981	>95%
		Puerto Rican	1 in 59	>95%	1 in 1161	>95%
Hermansky-Pudlak Syndrome, Type 3 (AR) NM_032383.4	HPS3	Ashkenazi Jewish	1 in 235	>95%	1 in 4681	>95%
		Worldwide	< 1 in 500	87%	1 in 3839	>95%
HMG-CoA Lyase Deficiency (AR) NM_000191.2	HMGCL	Worldwide	< 1 in 500	73%	1 in 1849	94%
Holocarboxylase Synthetase Deficiency (AR) NM_000411.6	HLCS	Caucasian	1 in 500	83%	1 in 2936	>95%
		Asian	1 in 158	83%	1 in 925	94%
		Worldwide	1 in 500	79%	1 in 2377	>95%
		Faroese	1 in 20	>95%	1 in 381	>95%
Homocystinuria (CBS-Related) (AR) NM_000071.2	CBS	Caucasian	1 in 52	74%	1 in 197	>95%
		Worldwide	1 in 293	78%	1 in 1328	>95%
		Qatari	1 in 21	86%	1 in 144	>95%
Homocystinuria due to MTHFR Deficiency (AR) NM_005957.4	MTHFR	Sephardic Jewish (Bukharian)	1 in 39	>95%	1 in 761	>95%

Variant tested: p.G158G (Genotyping only)

Homocystinuria, cBLE Type (AR) NM_002454.2	<i>MTRR</i>	Caucasian	< 1 in 500	93%	1 in 7130	>95%
		Worldwide	< 1 in 500	73%	1 in 1849	>95%
Hydrolethalus Syndrome (AR) NM_001134793.1	<i>HYLS1</i>	Worldwide	1 in 455	>95%	1 in 9081	>95%
		Finnish	1 in 50	>95%	1 in 981	>95%
Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome (AR) NM_014252.3	<i>SLC25A15</i>	Worldwide	< 1 in 500	83%	1 in 2936	>95%
		Metis (Saskatchewan)	1 in 19	>95%	1 in 361	>95%
Hypohidrotic Ectodermal Dysplasia 1 (XL) NM_001399.4	<i>EDA</i>	Worldwide	< 1 in 500	73%	1 in 1849	95%
Hypophosphatasia (AR) NM_000478.4	<i>ALPL</i>	Asian	1 in 192	72%	1 in 683	>95%
		Worldwide	1 in 345	64%	1 in 957	>95%
		Mennonite	1 in 25	>95%	1 in 481	>95%
Inclusion Body Myopathy 2 (AR) NM_005476.5	<i>GNE</i>	Caucasian	< 1 in 500	86%	1 in 3565	>95%
		Asian	1 in 58	83%	1 in 336	>95%
		Ashkenazi Jewish	< 1 in 500	>95%	1 in 9981	>95%
		Worldwide	1 in 179	82%	1 in 990	>95%
		Sephardic Jewish (Iranian and Syrian)	1 in 10	>95%	1 in 181	>95%
Infantile Cerebral and Cerebellar Atrophy (AR) NM_004268.4	<i>MED17</i>	Sephardic Jewish (Bukharian and Kurdish)	1 in 20	>95%	1 in 381	>95%
Isovaleric Acidemia (AR) NM_002225.3	<i>IVD</i>	Caucasian	1 in 144	69%	1 in 462	>95%
		Asian	1 in 75	55%	1 in 165	>95%
		Worldwide	1 in 158	68%	1 in 492	>95%
Joubert Syndrome 2 (AR) NM_001173990.2	<i>TMEM216</i>	Ashkenazi Jewish	1 in 110	>95%	1 in 2181	>95%
		Worldwide	< 1 in 500	>95%	1 in 9981	>95%
Joubert Syndrome 7 / Meckel Syndrome 5 / COACH Syndrome (AR) NM_015272.2	<i>RPGRIPL</i>	Worldwide	1 in 259	81%	1 in 1359	>95%
Junctional Epidermolysis Bullosa (LAMA3-Related) (AR) NM_000227.4	<i>LAMA3</i>	Worldwide	< 1 in 500	91%	1 in 5545	>95%
Junctional Epidermolysis Bullosa (LAMB3-Related) (AR) NM_000228.2	<i>LAMB3</i>	Worldwide	1 in 500	92%	1 in 6239	>95%
Junctional Epidermolysis Bullosa (LAMC2-Related) (AR) NM_018891.2	<i>LAMC2</i>	Worldwide	< 1 in 500	90%	1 in 4991	>95%
Krabbe Disease (AR) NM_000153.3	<i>GALC</i>	Worldwide	1 in 158	80%	1 in 786	>95%
		Druze Northern Israel	1 in 6	>95%	1 in 101	>95%
		Muslim Arab (Jerusalem)	1 in 6	>95%	1 in 101	>95%
Lamellar Ichthyosis, Type 1 (AR) NM_000359.2	<i>TGM1</i>	Caucasian	1 in 253	83%	1 in 1483	>95%
		Worldwide	1 in 301	81%	1 in 1580	>95%
		Norwegian	1 in 151	80%	1 in 751	>95%

Leber Congenital Amaurosis 10 and Other CEP290-Related Ciliopathies (AR) NM_025114.3	<i>CEP290</i>	Worldwide	1 in 185	93%	1 in 2630	>95%
Leber Congenital Amaurosis 13 (AR) NM_152443.2	<i>RDH12</i>	Worldwide	1 in 456	59%	1 in 1111	>95%
Leber Congenital Amaurosis 2 / Retinitis Pigmentosa 20 (AR) NM_000329.2	<i>RPE65</i>	Worldwide Sephardic Jewish (North African)	1 in 228 1 in 90	73% >95%	1 in 842 1 in 1781	>95% >95%
Leber Congenital Amaurosis 5 (AR) NM_181714.3	<i>LCA5</i>	Worldwide	< 1 in 500	88%	1 in 4159	>95%
Leber Congenital Amaurosis 8 / Retinitis Pigmentosa 12 (AR) NM_201253.2	<i>CRB1</i>	Worldwide	1 in 112	78%	1 in 506	>95%
Leigh Syndrome, French-Canadian Type (AR) NM_133259.3	<i>LRPPRC</i>	Worldwide French Canadian (Saguenay- Lac St. Jean)	< 1 in 500 1 in 23	>95% >95%	1 in 9981 1 in 441	>95% >95%
Lethal Congenital Contracture Syndrome 1 / Cell Lethal Arthrogryposis with Anterior Horn Disease (AR) NM_001003722.1	<i>GLE1</i>	Finnish	1 in 100	>95%	1 in 1981	>95%
Leukoencephalopathy with Vanishing White Matter (AR) NM_003907.2	<i>EIF2B5</i>	Worldwide	< 1 in 500	72%	1 in 1783	>95%
Limb-Girdle Muscular Dystrophy, Type 2A (AR) NM_000070.2	<i>CAPN3</i>	Caucasian Hispanic Asian Worldwide Amish	1 in 130 1 in 260 1 in 238 1 in 158 N/A	56% 68% 80% 73% >95%	1 in 294 1 in 810 1 in 1186 1 in 582 N/A	90% >95% >95% >95% >95%
Limb-Girdle Muscular Dystrophy, Type 2B (AR) NM_003494.3	<i>DYSF</i>	Worldwide Sephardic Jewish (Libyan, Kavkazi, and Yemenite)	1 in 311 1 in 14	>95% >95%	1 in 6201 1 in 261	>95% >95%
Limb-Girdle Muscular Dystrophy, Type 2C (AR) NM_000231.2	<i>SGCG</i>	Worldwide Moroccan Roma	1 in 354 1 in 250 1 in 96	80% 77% >95%	1 in 1766 1 in 1084 1 in 1901	87% >95% >95%
Limb-Girdle Muscular Dystrophy, Type 2D (AR) NM_000023.2	<i>SGCA</i>	Caucasian Worldwide Finnish	1 in 290 < 1 in 500 1 in 150	83% 64% >95%	1 in 1701 1 in 1387 1 in 2981	>95% >95% >95%
Limb-Girdle Muscular Dystrophy, Type 2E (AR) NM_000232.4	<i>SGCB</i>	Caucasian Worldwide	1 in 406 1 in 500	59% 77%	1 in 989 1 in 2171	64% 93%
Limb-Girdle Muscular Dystrophy, Type 2I (AR) NM_024301.4	<i>FKRP</i>	Worldwide Norwegian	1 in 158 1 in 116	87% >95%	1 in 1209 1 in 2301	>95% >95%

Lipoamide Dehydrogenase Deficiency (AR) NM_000108.4	<i>DLD</i>	Ashkenazi Jewish	1 in 107	>95%	1 in 2121	>95%
		Worldwide	< 1 in 500	74%	1 in 1920	>95%
Lipoid Adrenal Hyperplasia (AR) NM_000349.2	<i>STAR</i>	East Asian	1 in 177	95%	1 in 3521	>95%
		Worldwide	< 1 in 500	82%	1 in 2773	>95%
Lipoprotein Lipase Deficiency (AR) NM_000237.2	<i>LPL</i>	Caucasian	< 1 in 500	55%	1 in 1110	68%
		Asian	1 in 189	40%	1 in 314	67%
		Worldwide	1 in 500	86%	1 in 3565	>95%
		French Canadian (Saguenay - Lac St. Jean)	1 in 46	>95%	1 in 901	>95%
		French Canadian (Other)	1 in 139	>95%	1 in 2761	>95%
Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (AR) NM_000182.4	<i>HADHA</i>	Caucasian	1 in 254	93%	1 in 3615	>95%
		Worldwide	1 in 351	93%	1 in 5001	>95%
Lysinuric Protein Intolerance (AR) NM_001126106.2	<i>SLC7A7</i>	Worldwide	< 1 in 500	88%	1 in 4159	>95%
		Japanese	1 in 119	88%	1 in 984	>95%
		Finnish	1 in 122	>95%	1 in 2421	>95%
Maple Syrup Urine Disease, Type 1a (AR) NM_000709.3	<i>BCKDHA</i>	Caucasian	1 in 320	75%	1 in 1277	94%
		Worldwide	1 in 289	63%	1 in 779	>95%
		Mennonite	1 in 10	>95%	1 in 181	>95%
		Portuguese Roma	1 in 71	>95%	1 in 1401	>95%
Maple Syrup Urine Disease, Type 1b (AR) NM_000056.3	<i>BCKDHB</i>	Caucasian	1 in 433	56%	1 in 983	>95%
		Asian	1 in 163	57%	1 in 378	>95%
		Ashkenazi Jewish	1 in 97	>95%	1 in 1921	>95%
		Worldwide	1 in 327	72%	1 in 1165	>95%
Meckel Syndrome 1 / Bardet-Biedl Syndrome 13 (AR) NM_017777.3	<i>MKS1</i>	Caucasian	1 in 260	83%	1 in 1525	>95%
		Worldwide	1 in 260	91%	1 in 2879	>95%
		Finnish	1 in 47	>95%	1 in 921	>95%
Medium Chain Acyl-CoA Dehydrogenase Deficiency (AR) NM_000016.5	<i>ACADM</i>	Caucasian	1 in 55	88%	1 in 451	93%
		Asian	1 in 178	53%	1 in 378	83%
		Worldwide	1 in 69	81%	1 in 359	>95%
Megalencephalic Leukoencephalopathy With Subcortical Cysts (AR) NM_015166.3	<i>MLC1</i>	Worldwide	< 1 in 500	93%	1 in 7130	>95%
		Sephardic Jewish (Libyan)	1 in 40	>95%	1 in 781	>95%
Menkes Disease (XL) NM_000052.6	<i>ATP7A</i>	Worldwide	< 1 in 500	71%	1 in 1722	87%
Metachromatic Leukodystrophy (AR) NM_000487.5	<i>ARSA</i>	Ashkenazi Jewish	< 1 in 500	>95%	1 in 9981	>95%
		Worldwide	1 in 100	67%	1 in 301	>95%
		Sephardic Jewish (Yemenite)	1 in 46	>95%	1 in 901	>95%
		Navajo	1 in 25	>95%	1 in 481	>95%

3-Methylcrotonyl-CoA Carboxylase Deficiency (MCCC1-Related) (AR) NM_020166.4	<i>MCCC1</i>	Caucasian Worldwide	1 in 137 1 in 147	77% 75%	1 in 592 1 in 585	88% >95%
3-Methylcrotonyl-CoA Carboxylase Deficiency (MCCC2-Related) (AR) NM_022132.4	<i>MCCC2</i>	Caucasian Worldwide	1 in 112 1 in 120	59% 69%	1 in 272 1 in 385	91% >95%
3-Methylglutaconic Aciduria, Type III (AR) NM_025136.3	<i>OPA3</i>	Worldwide Sephardic Jewish (Iraqi)	< 1 in 500 1 in 13	95% >95%	1 in 9981 1 in 241	>95% >95%
Methylmalonic Acidemia (MMAA-Related) (AR) NM_172250.2	<i>MMAA</i>	Caucasian Worldwide	1 in 316 1 in 316	92% 88%	1 in 3939 1 in 2626	>95% >95%
Methylmalonic Acidemia (MMAB-Related) (AR) NM_052845.3	<i>MMAB</i>	Caucasian Worldwide	1 in 456 1 in 456	91% 91%	1 in 5057 1 in 5057	>95% >95%
Methylmalonic Acidemia (MUT-Related) (AR) NM_000255.3	<i>MUT</i>	Caucasian African Hispanic Asian Worldwide	1 in 224 1 in 177 1 in 383 1 in 53 1 in 383	22% 52% 65% 50% 70%	1 in 287 1 in 368 1 in 1092 1 in 105 1 in 1274	>95% >95% >95% >95% >95%
Methylmalonic Aciduria and Homocystinuria, Cobalamin C Type (AR) NM_015506.2	<i>MMACHC</i>	Caucasian Asian Worldwide	1 in 138 1 in 113 1 in 138	95% 90% 91%	1 in 2741 1 in 1121 1 in 1523	>95% >95% >95%
Methylmalonic Aciduria and Homocystinuria, Cobalamin D Type (AR) NM_015702.2	<i>MMADHC</i>	Caucasian	< 1 in 500	>95%	1 in 9981	>95%
Microphthalmia / Anophthalmia (AR) NM_182894.2	<i>VSX2</i>	Worldwide Sephardic Jewish (Iranian and Syrian)	< 1 in 500 1 in 145	>95% >95%	1 in 9981 1 in 2881	>95% >95%
Mitochondrial Complex I Deficiency (ACAD9-Related) (AR) NM_014049.4	<i>ACAD9</i>	Worldwide	< 1 in 500	94%	1 in 8318	>95%
Mitochondrial Complex I Deficiency (NDUFA5-Related) (AR) NM_024120.4	<i>NDUFA5</i>	Ashkenazi Jewish Worldwide	1 in 290 < 1 in 500	>95% >95%	1 in 5781 1 in 9981	>95% >95%
Mitochondrial Complex I Deficiency (NDUFS6-Related) (AR) NM_004553.4	<i>NDUFS6</i>	Worldwide Sephardic Jewish (Caucasus)	< 1 in 500 1 in 24	93% >95%	1 in 7130 1 in 461	93% >95%
Mitochondrial DNA Depletion Syndrome 6 / Navajo Neurohepatopathy (AR) NM_002437.4	<i>MPV17</i>	Worldwide Navajo	< 1 in 500 1 in 20	88% >95%	1 in 4159 1 in 381	>95% >95%
Mitochondrial Myopathy and Sideroblastic Anemia 1 (AR) NM_025215.5	<i>PUS1</i>	Worldwide Sephardic Jewish (Iranian)	< 1 in 500 N/A	>95% >95%	1 in 9981 N/A	>95% >95%

Mucopolipidosis II / IIIA (AR) NM_024312.4	<i>GNPTAB</i>	Caucasian	1 in 225	90%	1 in 2241	95%
		Asian	1 in 389	79%	1 in 1849	84%
		Worldwide	1 in 408	84%	1 in 2545	>95%
Mucopolipidosis III Gamma (AR) NM_032520.4	<i>GNPTG</i>	Caucasian	1 in 273	94%	1 in 4534	94%
		Worldwide	< 1 in 500	81%	1 in 2627	>95%
Mucopolipidosis IV (AR) NM_020533.2	<i>MCOLN1</i>	Ashkenazi Jewish	1 in 89	>95%	1 in 1761	>95%
		Worldwide	< 1 in 500	92%	1 in 6239	>95%
Mucopolysaccharidosis, Type I (AR) NM_000203.4	<i>IDUA</i>	Worldwide	1 in 144	72%	1 in 512	>95%
Mucopolysaccharidosis, Type II (XL) NM_000202.6 <i>Exception: Exon 1</i>	<i>IDS</i>	Worldwide	< 1 in 500	67%	1 in 1513	86%
Mucopolysaccharidosis, Type IIIA (AR) NM_000199.3	<i>SGSH</i>	Caucasian	1 in 253	68%	1 in 789	95%
		Worldwide	1 in 415	56%	1 in 942	>95%
Mucopolysaccharidosis, Type IIIB (AR) NM_000263.3	<i>NAGLU</i>	Caucasian	1 in 346	59%	1 in 842	80%
		Asian	1 in 298	70%	1 in 991	>95%
		Worldwide	< 1 in 500	64%	1 in 1387	>95%
Mucopolysaccharidosis, Type IIIC (AR) NM_152419.2	<i>HGSNAT</i>	Caucasian	1 in 259	81%	1 in 1359	93%
		Asian	< 1 in 500	>95%	1 in 9981	>95%
		Worldwide	1 in 482	77%	1 in 2092	>95%
Mucopolysaccharidosis, Type IIID (AR) NM_002076.3	<i>GNS</i>	Worldwide	< 1 in 500	90%	1 in 4991	90%
Mucopolysaccharidosis, Type IVb / GM1 Gangliosidosis (AR) NM_000404.2	<i>GLB1</i>	Caucasian	1 in 278	57%	1 in 645	>95%
		Worldwide	1 in 158	69%	1 in 507	>95%
		Roma	1 in 50	>95%	1 in 981	>95%
		South Brazilian	1 in 58	>95%	1 in 1141	>95%
Mucopolysaccharidosis, Type VI (AR) NM_000046.3	<i>ARSB</i>	Caucasian	1 in 273	67%	1 in 825	>95%
		Asian	1 in 423	53%	1 in 899	>95%
		Worldwide	1 in 291	54%	1 in 631	>95%
Mucopolysaccharidosis, Type IX (AR) NM_153281.1	<i>HYAL1</i>	Worldwide	< 1 in 500	>95%	1 in 9981	>95%
Multiple Sulfatase Deficiency (AR) NM_182760.3	<i>SUMF1</i>	Ashkenazi Jewish	1 in 279	>95%	1 in 5561	>95%
		Worldwide	< 1 in 500	65%	1 in 1427	>95%
Muscle-Eye-Brain Disease and Other POMGNT1-Related Congenital Muscular Dystrophy- Dystroglycanopathies (AR) NM_017739.3	<i>POMGNT1</i>	Worldwide	1 in 462	74%	1 in 1774	95%
		Finnish	1 in 111	>95%	1 in 2201	>95%
Myoneurogastrointestinal Encephalopathy (AR) NM_001113755.2	<i>TYMP</i>	Caucasian	1 in 500	69%	1 in 1611	>95%
		Worldwide	< 1 in 500	62%	1 in 1314	>95%
		Sephardic Jewish (Iranian)	1 in 158	>95%	1 in 3141	>95%

Myotubular Myopathy 1 (XL) NM_000252.2	<i>MTM1</i>	Worldwide	< 1 in 500	86%	1 in 3565	>95%
N-Acetylglutamate Synthase Deficiency (AR) NM_153006.2	<i>NAGS</i>	Worldwide	< 1 in 500	59%	1 in 1218	>95%
Nemaline Myopathy 2 (AR) NM_001271208.1		Ashkenazi Jewish	1 in 168	>95%	1 in 3341	>95%
		Worldwide	1 in 224	81%	1 in 1175	94%
		Finnish	1 in 112	75%	1 in 445	75%
<i>Exception: Exons 82 - 105</i>						
Nephrogenic Diabetes Insipidus, Type II (AR) NM_000486.5	<i>AQP2</i>	Worldwide	< 1 in 500	44%	1 in 892	>95%
Nephrotic Syndrome (NPHS1-Related) / Congenital Finnish Nephrosis (AR) NM_004646.3	<i>NPHS1</i>	Worldwide	1 in 325	80%	1 in 1621	>95%
		Finnish	1 in 45	>95%	1 in 881	>95%
		Groffdale Conference Mennonites	1 in 12	>95%	1 in 221	>95%
Nephrotic Syndrome (NPHS2-Related) / Steroid-Resistant Nephrotic Syndrome (AR) NM_014625.3	<i>NPHS2</i>	Worldwide	1 in 377	80%	1 in 1881	>95%
Neuronal Ceroid-Lipofuscinosis (CLN3-Related) (AR) NM_000086.2	<i>CLN3</i>	Caucasian	1 in 188	94%	1 in 3118	>95%
		Worldwide	1 in 233	94%	1 in 3868	>95%
Neuronal Ceroid-Lipofuscinosis (CLN5-Related) (AR) NM_006493.2	<i>CLN5</i>	Worldwide	< 1 in 500	91%	1 in 5545	>95%
		Finnish	1 in 100	>95%	1 in 1981	>95%
Neuronal Ceroid-Lipofuscinosis (CLN6-Related) (AR) NM_017882.2	<i>CLN6</i>	Worldwide	< 1 in 500	86%	1 in 3565	>95%
Neuronal Ceroid-Lipofuscinosis (CLN8-Related) (AR) NM_018941.3	<i>CLN8</i>	Worldwide	< 1 in 500	84%	1 in 3120	>95%
		Finnish	1 in 135	>95%	1 in 2681	>95%
Neuronal Ceroid-Lipofuscinosis (MFSD8-Related) (AR) NM_152778.2	<i>MFSD8</i>	Worldwide	< 1 in 500	82%	1 in 2773	>95%
Neuronal Ceroid-Lipofuscinosis (PPT1-Related) (AR) NM_000310.3	<i>PPT1</i>	Worldwide	1 in 368	77%	1 in 1597	>95%
		Finnish	1 in 70	94%	1 in 1151	>95%
Neuronal Ceroid-Lipofuscinosis (TPP1-Related) (AR) NM_000391.3	<i>TPP1</i>	Worldwide	1 in 314	87%	1 in 2409	>95%
		Newfoundland	1 in 59	>95%	1 in 1161	>95%
Niemann-Pick Disease, Type A/B (AR) NM_000543.4	<i>SMPD1</i>	Caucasian	1 in 244	37%	1 in 387	>95%
		Ashkenazi Jewish	1 in 115	>95%	1 in 2281	>95%
		Worldwide	1 in 196	85%	1 in 1301	>95%

Niemann-Pick Disease, Type C (NPC1-Related) (AR) NM_000271.4	<i>NPC1</i>	Caucasian	1 in 185	73%	1 in 682	>95%
		Asian	1 in 404	45%	1 in 734	85%
		Worldwide	1 in 282	62%	1 in 740	>95%
Niemann-Pick Disease, Type C (NPC2-Related) (AR) NM_006432.3	<i>NPC2</i>	Worldwide	< 1 in 500	83%	1 in 2936	>95%
Nijmegen Breakage Syndrome (AR) NM_002485.4	<i>NBN</i>	Caucasian	1 in 155	90%	1 in 1541	>95%
		Worldwide	< 1 in 500	>95%	1 in 9981	>95%
Non-Syndromic Hearing Loss (GJB2-Related) (AR) NM_004004.5	<i>GJB2</i>	Caucasian	1 in 42	88%	1 in 343	>95%
		Asian	1 in 50	83%	1 in 289	>95%
		Ashkenazi Jewish	1 in 21	>95%	1 in 401	>95%
		Worldwide	1 in 43	82%	1 in 234	>95%
Odonto-Onycho-Dermal Dysplasia / Schopf-Schulz-Passarge Syndrome (AR) NM_025216.2	<i>WNT10A</i>	Worldwide	1 in 305	67%	1 in 922	>95%
Omenn Syndrome (RAG2-Related) (AR) NM_000536.2	<i>RAG2</i>	Worldwide	< 1 in 500	51%	1 in 1019	>95%
		Sephardic Jewish (Iraqi)	N/A	88%	N/A	>95%
Omenn Syndrome / Severe Combined Immunodeficiency, Athabaskan-Type (AR) NM_001033855.1	<i>DCLRE1C</i>	Worldwide	< 1 in 500	54%	1 in 1086	81%
		Navajo and Apache Native American	1 in 48	>95%	1 in 941	>95%
Ornithine Aminotransferase Deficiency (AR) NM_000274.3	<i>OAT</i>	Worldwide	< 1 in 500	89%	1 in 4537	>95%
		Finnish	1 in 147	>95%	1 in 2921	>95%
		Sephardic Jewish (Iraqi and Syrian)	1 in 177	>95%	1 in 3521	>95%
Ornithine Transcarbamylase Deficiency (XL) NM_000531.5	<i>OTC</i>	Worldwide	< 1 in 500	70%	1 in 1664	95%
Osteopetrosis 1 (AR) NM_006019.2	<i>TCIRG1</i>	Ashkenazi Jewish	1 in 350	>95%	1 in 6981	>95%
		Worldwide	1 in 316	67%	1 in 956	95%
		Costa Rican	1 in 86	>95%	1 in 1701	>95%
		Chuvashiyian	1 in 60	>95%	1 in 1181	>95%
Pendred Syndrome (AR) NM_000441.1	<i>SLC26A4</i>	Caucasian	1 in 88	86%	1 in 622	93%
		African	1 in 76	>95%	1 in 1501	>95%
		Asian	1 in 74	83%	1 in 430	>95%
		Worldwide	1 in 80	81%	1 in 417	>95%
Phenylalanine Hydroxylase Deficiency (AR) NM_000277.1	<i>PAH</i>	Caucasian	1 in 50	94%	1 in 818	>95%
		African	1 in 143	87%	1 in 1093	>95%
		Asian	1 in 78	79%	1 in 368	>95%
		Ashkenazi Jewish	1 in 225	75%	1 in 897	93%
		Worldwide	1 in 65	77%	1 in 279	>95%
		Turkish	1 in 32	63%	1 in 85	>95%
		Irish	1 in 34	91%	1 in 368	92%
		Sicilian	1 in 26	48%	1 in 49	>95%
		Sephardic Jewish (Iranian, Bukharian, Kavkazi, Tunisian, and Moroccan)	1 in 18	88%	1 in 143	91%

3-Phosphoglycerate Dehydrogenase Deficiency (AR) NM_006623.3	<i>PHGDH</i>	Ashkenazi Jewish Worldwide	1 in 453 < 1 in 500	>95% >95%	1 in 9041 1 in 9981	>95% >95%
Polycystic Kidney Disease, Autosomal Recessive (AR) NM_138694.3	<i>PKHD1</i>	Caucasian Ashkenazi Jewish Worldwide South African Afrikaner	1 in 100 1 in 106 1 in 144 1 in 52	70% >95% 69% >95%	1 in 331 1 in 2101 1 in 462 1 in 1021	>95% >95% >95% >95%
Polyglandular Autoimmune Syndrome, Type 1 (AR) NM_000383.2	<i>AIRE</i>	Worldwide Finnish Sardinian Sephardic Jewish (Iranian)	1 in 354 1 in 79 1 in 60 1 in 27	93% >95% 95% >95%	1 in 5044 1 in 1561 1 in 1181 1 in 521	>95% >95% 95% >95%
Pontocerebellar Hypoplasia, Type 1A (AR) NM_003384.2	<i>VRK1</i>	Ashkenazi Jewish Worldwide	1 in 225 < 1 in 500	>95% >95%	1 in 4481 1 in 9981	>95% >95%
Pontocerebellar Hypoplasia, Type 6 (AR) NM_020320.3	<i>RARS2</i>	Worldwide Sephardic Jewish (Iraqi, Syrian, and Tunisian)	< 1 in 500 N/A	>95% >95%	1 in 9981 N/A	>95% >95%
Primary Carnitine Deficiency (AR) NM_003060.2	<i>SLC22A5</i>	Caucasian Asian Worldwide Faroese	1 in 110 1 in 100 1 in 200 1 in 20	94% 70% 68% >95%	1 in 1818 1 in 331 1 in 623 1 in 381	94% 81% >95% >95%
Primary Ciliary Dyskinesia (DNAH5-Related) (AR) NM_001369.2	<i>DNAH5</i>	Ashkenazi Jewish Worldwide	1 in 174 1 in 120	>95% 78%	1 in 3461 1 in 542	>95% >95%
Primary Ciliary Dyskinesia (DNAI1-Related) (AR) NM_012144.3	<i>DNAI1</i>	Ashkenazi Jewish Worldwide	1 in 352 1 in 182	>95% 92%	1 in 7021 1 in 2264	>95% >95%
Primary Ciliary Dyskinesia (DNAI2-Related) (AR) NM_023036.4	<i>DNAI2</i>	Ashkenazi Jewish Worldwide	1 in 200 1 in 500	>95% >95%	1 in 3981 1 in 9981	>95% >95%
Primary Hyperoxaluria, Type 1 (AR) NM_000030.2	<i>AGXT</i>	Worldwide	1 in 158	77%	1 in 684	>95%
Primary Hyperoxaluria, Type 2 (AR) NM_012203.1	<i>GRHPR</i>	Worldwide	< 1 in 500	92%	1 in 6239	>95%
Primary Hyperoxaluria, Type 3 (AR) NM_138413.3	<i>HOGA1</i>	Ashkenazi Jewish Worldwide	N/A 1 in 309	>95% 88%	N/A 1 in 2568	>95% >95%
Progressive Cerebello-Cerebral Atrophy (AR) NM_016955.3	<i>SEPSECS</i>	Worldwide Sephardic Jewish (Moroccan and Iraqi)	< 1 in 500 1 in 41	>95% >95%	1 in 9981 1 in 801	>95% >95%
Progressive Familial Intrahepatic Cholestasis, Type 2 (AR) NM_003742.2	<i>ABCB11</i>	Worldwide	1 in 158	61%	1 in 404	>95%

Propionic Acidemia (PCCA-Related) (AR) NM_000282.3	<i>PCCA</i>	Caucasian	1 in 380	54%	1 in 825	73%
		Asian	1 in 162	72%	1 in 576	90%
		Worldwide	1 in 224	52%	1 in 466	85%
Propionic Acidemia (PCCB-Related) (AR) NM_000532.4	<i>PCCB</i>	Caucasian	1 in 202	60%	1 in 504	88%
		Asian	1 in 145	83%	1 in 848	>95%
		Worldwide	1 in 224	70%	1 in 744	>95%
Pycnodysostosis (AR) NM_000396.3	<i>CTSK</i>	Worldwide	1 in 438	60%	1 in 1094	>95%
Pyruvate Dehydrogenase E1-Alpha Deficiency (XL) NM_000284.3	<i>PDHA1</i>	Worldwide	< 1 in 500	64%	1 in 1387	94%
Pyruvate Dehydrogenase E1-Beta Deficiency (AR) NM_000925.3	<i>PDHB</i>	Worldwide	< 1 in 500	74%	1 in 1920	>95%
6-Pyruvoyl-Tetrahydropterin Synthase Deficiency (AR) NM_000317.2	<i>PTS</i>	Asian	1 in 122	87%	1 in 932	>95%
		Worldwide	< 1 in 500	71%	1 in 1722	>95%
Renal Tubular Acidosis and Deafness (AR) NM_001692.3	<i>ATP6V1B1</i>	Worldwide	< 1 in 500	86%	1 in 3565	>95%
		Sephardic Jewish (Syrian)	1 in 140	>95%	1 in 2781	>95%
Retinitis Pigmentosa 25 (AR) NM_001142800.1	<i>EYS</i>	Caucasian	1 in 53	60%	1 in 131	72%
		Ashkenazi Jewish	< 1 in 500	>95%	1 in 9981	>95%
		Worldwide	1 in 129	63%	1 in 347	92%
		Sephardic Jewish (Moroccan)	1 in 42	22%	1 in 54	>95%
Retinitis Pigmentosa 26 (AR) NM_001030311.2	<i>CERKL</i>	Worldwide	1 in 137	>95%	1 in 2721	>95%
		Sephardic Jewish - Yemenite	1 in 24	>95%	1 in 461	>95%
Retinitis Pigmentosa 28 (AR) NM_032180.2	<i>FAM161A</i>	Ashkenazi Jewish	1 in 214	>95%	1 in 4261	>95%
		Worldwide	1 in 289	>95%	1 in 5761	>95%
		Sephardic Jewish (Libyan, Moroccan, Tunisian, and Bulgarian)	1 in 41	>95%	1 in 801	>95%
Retinitis Pigmentosa 59 (AR) NM_001243564.1	<i>DHDDS</i>	Ashkenazi Jewish	1 in 117	>95%	1 in 2321	>95%
Rhizomelic Chondrodysplasia Punctata, Type 1 (AR) NM_000288.3	<i>PEX7</i>	Caucasian	1 in 158	87%	1 in 1209	>95%
		Worldwide	< 1 in 500	87%	1 in 3839	>95%
Rhizomelic Chondrodysplasia Punctata, Type 3 (AR) NM_003659.3	<i>AGPS</i>	Worldwide	< 1 in 500	87%	1 in 3839	93%
Roberts Syndrome (AR) NM_001017420.2	<i>ESCO2</i>	Worldwide	< 1 in 500	>95%	1 in 9981	>95%
Salla Disease (AR) NM_012434.4	<i>SLC17A5</i>	Worldwide	< 1 in 500	94%	1 in 8318	>95%
		Finnish	1 in 100	>95%	1 in 1981	>95%
		Swedish	1 in 125	>95%	1 in 2481	>95%
		Canadian Inuit	1 in 129	>95%	1 in 2561	>95%

Sandhoff Disease (AR) NM_000521.3	<i>HEXB</i>	Caucasian	1 in 235	26%	1 in 317	>95%
		Worldwide	1 in 180	82%	1 in 995	>95%
		Northern Saskatchewan Metis	1 in 15	75%	1 in 57	>95%
		Argentinian Creole	1 in 26	>95%	1 in 501	>95%
Schimke Immunoosseous Dysplasia (AR) NM_014140.3	<i>SMARCA1</i>	Worldwide	< 1 in 500	81%	1 in 2627	>95%
Segawa Syndrome (AR) NM_000360.3	<i>TH</i>	Caucasian	1 in 224	74%	1 in 859	>95%
		Asian	1 in 416	78%	1 in 1887	>95%
		Worldwide	< 1 in 500	78%	1 in 2269	>95%
Sjogren-Larsson Syndrome (AR) NM_000382.2	<i>ALDH3A2</i>	Worldwide	< 1 in 500	87%	1 in 3839	>95%
		Swedish	1 in 205	>95%	1 in 4081	>95%
Smith-Lemli-Opitz Syndrome (AR) NM_001360.2	<i>DHCR7</i>	Caucasian	1 in 48	81%	1 in 248	>95%
		African	1 in 93	67%	1 in 280	>95%
		Asian	< 1 in 500	84%	1 in 3120	>95%
		Ashkenazi Jewish	1 in 41	>95%	1 in 801	>95%
		Worldwide	1 in 68	80%	1 in 336	>95%
Spondylothoracic Dysostosis (AR) NM_001039958.1	<i>MESP2</i>	Worldwide	1 in 224	>95%	1 in 4461	>95%
		Puerto Rican	1 in 55	>95%	1 in 1081	>95%
Steel Syndrome (AR) NM_032888.2 <i>Variant tested: p.G697R (Genotyping only)</i>	<i>COL27A1</i>	Puerto Rican	< 1 in 500	>95%	1 in 9981	>95%
Stuve-Wiedemann Syndrome (AR) NM_002310.5	<i>LIFR</i>	Worldwide	< 1 in 500	94%	1 in 8318	>95%
Sulfate Transporter-Related Osteochondrodysplasia (AR) NM_000112.3	<i>SLC26A2</i>	Worldwide	1 in 158	>95%	1 in 3141	>95%
		Finnish	1 in 50	>95%	1 in 981	>95%
Tay-Sachs Disease (AR) NM_000520.4	<i>HEXA</i>	Caucasian	1 in 182	91%*	1 in 2012	>95%
		African American	1 in 271	90%*	1 in 2701	>95%
		Asian	1 in 126	70%*	1 in 418	91%
		Ashkenazi Jewish	1 in 27	>95%*	1 in 521	>95%
		Worldwide	1 in 288	94%*	1 in 4784	>95%
		French Canadian (Gaspesie)	1 in 13	>95%*	1 in 241	>95%
		French Canadian (Other)	1 in 73	>95%*	1 in 1441	>95%
		Irish	1 in 41	90%*	1 in 401	>95%
		Sephardic Jewish (Moroccan and Iraqi)	1 in 125	>95%*	1 in 2481	>95%
Tyrosinemia, Type I (AR) NM_000137.2	<i>FAH</i>	Caucasian	1 in 333	89%	1 in 3019	>95%
		African	1 in 478	92%	1 in 5964	>95%
		Asian	< 1 in 500	70%	1 in 1664	>95%
		Ashkenazi Jewish	1 in 143	>95%	1 in 2841	>95%
		Worldwide	< 1 in 500	82%	1 in 2773	>95%
		French Canadian (Saguenay Lac-St. Jean)	1 in 25	>95%	1 in 481	>95%
		French Canadian (Other)	1 in 66	>95%	1 in 1301	>95%

Usher Syndrome, Type IB (AR) NM_000260.3	<i>MYO7A</i>	Caucasian	1 in 145	75%	1 in 577	93%
		African	< 1 in 500	13%	1 in 575	>95%
		Asian	1 in 62	85%	1 in 408	>95%
		Worldwide	1 in 206	73%	1 in 760	>95%
Usher Syndrome, Type IC (AR) NM_005709.3	<i>USH1C</i>	Worldwide	1 in 353	77%	1 in 1531	>95%
		French Canadian/Acadian	1 in 227	>95%	1 in 4521	>95%
Usher Syndrome, Type ID (AR) NM_022124.5	<i>CDH23</i>	Worldwide	1 in 306	66%	1 in 898	>95%
Usher Syndrome, Type IF (AR) NM_001142764.1	<i>PCDH15</i>	Ashkenazi Jewish	1 in 78	>95%	1 in 1541	>95%
		Worldwide	1 in 395	76%	1 in 1643	92%
Usher Syndrome, Type IIA (AR) NM_206933.2	<i>USH2A</i>	Caucasian	1 in 73	77%	1 in 314	88%
		Worldwide	1 in 126	69%	1 in 404	>95%
		Sephardic Jewish (Iraqi and Iranian)	1 in 36	71%	1 in 122	75%
Usher Syndrome, Type III (AR) NM_174878.2	<i>CLRN1</i>	Ashkenazi Jewish	1 in 120	>95%	1 in 2381	>95%
		Worldwide	1 in 500	88%	1 in 4159	>95%
		Finnish	1 in 70	>95%	1 in 1381	>95%
Very Long Chain Acyl-CoA Dehydrogenase Deficiency (AR) NM_000018.3	<i>ACADVL</i>	Caucasian	1 in 88	62%	1 in 230	>95%
		Asian	1 in 194	53%	1 in 412	93%
		Worldwide	1 in 146	59%	1 in 355	>95%
Walker-Warburg Syndrome and Other <i>FKTN</i>-Related Dystrophies (AR) NM_001079802.1	<i>FKTN</i>	Ashkenazi Jewish	1 in 80	>95%	1 in 1581	>95%
		Worldwide	< 1 in 500	14%	1 in 581	20%
		Japanese	1 in 188	4%	1 in 196	5%
Wilson Disease (AR) NM_000053.3	<i>ATP7B</i>	Caucasian	1 in 90	64%	1 in 248	>95%
		Asian	1 in 50	63%	1 in 133	>95%
		Ashkenazi Jewish	1 in 67	>95%	1 in 1321	>95%
		Worldwide	1 in 90	79%	1 in 425	>95%
		Canary Islands	1 in 25	88%	1 in 201	>95%
		Sardinian	1 in 42	>95%	1 in 821	>95%
Wolman Disease / Cholesteryl Ester Storage Disease (AR) NM_000235.3	<i>LIPA</i>	Caucasian	1 in 145	>95%	1 in 2881	>95%
		Ashkenazi Jewish	< 1 in 500	>95%	1 in 9981	>95%
		Worldwide	< 1 in 500	86%	1 in 3565	>95%
		Sephardic Jewish (Iranian)	1 in 26	>95%	1 in 501	>95%
X-Linked Juvenile Retinoschisis (XL) NM_000330.3	<i>RS1</i>	Worldwide	< 1 in 500	74%	1 in 1920	>95%
X-Linked Severe Combined Immunodeficiency (XL) NM_000206.2	<i>IL2RG</i>	Worldwide	< 1 in 500	89%	1 in 4537	>95%

Zellweger Syndrome Spectrum (PEX1-Related) (AR)	<i>PEX1</i>	Caucasian	1 in 147	89%	1 in 1328	>95%
NM_000466.2		Worldwide	< 1 in 500	80%	1 in 2496	>95%
Zellweger Syndrome Spectrum (PEX2-Related) (AR)	<i>PEX2</i>	Caucasian	< 1 in 500	80%	1 in 2496	>95%
NM_000318.2		Ashkenazi Jewish	1 in 227	>95%	1 in 4521	>95%
		Worldwide	< 1 in 500	84%	1 in 3120	>95%
Zellweger Syndrome Spectrum (PEX6-Related) (AR)	<i>PEX6</i>	Worldwide	1 in 280	69%	1 in 901	>95%
NM_000287.3		French Canadian	1 in 55	>95%	1 in 1081	>95%
		Sephardic Jewish (Yemenite)	1 in 18	>95%	1 in 341	>95%
Zellweger Syndrome Spectrum (PEX10-Related) (AR)	<i>PEX10</i>	Asian	< 1 in 500	>95%	1 in 9981	>95%
NM_153818.1		Worldwide	< 1 in 500	89%	1 in 4537	>95%

*Carrier detection by HEXA enzyme analysis has a detection rate of approximately 98%.

AR=Autosomal recessive; N/A=Not available; XL=X-linked