progenity®

Preparent® Global Panel

See next section for Global+ Panel

Disorder	Gene	Variants	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
17-alpha-hydroxylase deficiency	*CYP17A1*	1118A>T, 160_162delTTC	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
17-beta-hydroxysteroid dehydrogenase deficiency, type III	*HSD17B3*	239G>A	Other or Mixed Ethnicity	1 in 192	10%	1 in 213
3-beta-hydroxysteroid dehydrogenase deficiency, type II	*HSD3B2*	29C>A, 512G>A, 776C>T	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
3-hydroxy-3-methylglutaryl CoA lyase deficiency	*HMGCL*	109G>T, 122G>A, 561+1G>A	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
3-methylglutaconic aciduria, type III	*OPA3*	143-1G>C	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
3-phosphoglycerate dehydrogenase deficiency	*PHGDH*	1468G>A	Other or Mixed Ethnicity	1 in 500	83%	1 in 2936
Abetalipoproteinemia	*MTTP*	2593G>T	Ashkenazi Jewish Other or Mixed Ethnicity	1 in 131 1 in 500	75% 10%	1 in 521 1 in 555
Achalasia-addisonianism-alacrima syndrome	*AAAS*	1331+1G>A, 787T>C	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Achromatopsia, *CNGA3*-related	*CNGA3*	1585G>A	Other or Mixed Ethnicity	1 in 173	10%	1 in 192
Achromatopsia, *CNGB3*-related	*CNGB3*	1148delC, 1304C>T	Other or Mixed Ethnicity	1 in 123	78%	1 in 556
Acrodermatitis enteropathica	*SLC39A4*	599C>T, 970_973delTCAG	Other or Mixed Ethnicity	1 in 354	10%	1 in 393
Adenosine deaminase deficiency	*ADA*	467G>A, 986C>T	Other or Mixed Ethnicity	1 in 71	53%	1 in 150
Adrenoleukodystrophy (X-linked)*	*ABCD1*	686T>C	Other or Mixed Ethnicity	1 in 10,000	10%	1 in 11111
Aicardi-Goutieres syndrome, *RNASEH2C*-related	*RNASEH2C*	205C>T	Other or Mixed Ethnicity	1 in 1444	10%	1 in 1604
Aicardi-Goutieres syndrome, *SAMHD1*-related	*SAMHD1*	1411-2A>G	Other or Mixed Ethnicity	1 in 1388	10%	1 in 1542
Aicardi-Goutieres syndrome, *TREX1*-related	*TREX1*	341G>A, 490C>T	Other or Mixed Ethnicity	1 in 1037	50%	1 in 2073
Alkaptonuria	*HGD*	1102A>G, 1111dupC, 360T>G, 457dupG, 481G>A, 808G>A	Caucasian/White Other or Mixed Ethnicity	1 in 250 1 in 250	20% 10%	1 in 312 1 in 278
Alpha-mannosidosis	*MAN2B1*	1830+1G>C, 2248C>T, 2426T>C	Other or Mixed Ethnicity	1 in 354	41%	1 in 599
Alpha-thalassemia	*HBA1* *HBA2*	-alpha3.7, -alpha4.2, -alpha(20.5), --SEA, --MED, --FIL, --THAI	African American/Black Asian Other or Mixed Ethnicity	1 in 30 1 in 20 1 in 56	90% 90% 90%	1 in 291 1 in 191 1 in 551

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Alport syndrome, *COL4A3*-related	*COL4A3*	2T>C, 4001G>A, 40del24	Other or Mixed Ethnicity	1 in 408	10%	1 in 453
Alport syndrome, *COL4A5*-related (X-linked)*	*COL4A5*	4946T>G, 5030G>A	Other or Mixed Ethnicity	1 in 31,250	12%	1 in 35,511
Amish infantile epilepsy syndrome	*ST3GAL5*	862C>T	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Andermann syndrome	*SLC12A6*	2436+1delG, 3031C>T	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Argininosuccinic aciduria	*ASL*	1153C>T, 346C>T, c. 1060C>T	Other or Mixed Ethnicity	1 in 132	13%	1 in 152
Aromatase deficiency	*CYP19A1*	1094G>A, 1123C>T, 1224delC, 1303C>T, 1310G>A, 469delC, 628G>A, 629-3C>A, c.451+1G>A, c.858+2T>C	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Arthrogryposis, mental retardation and seizures	*SLC35A3*	514C>T	Ashkenazi Jewish Other or Mixed Ethnicity	1 in 373 1 in 500	10% 10%	1 in 414 1 in 555
Arts syndrome (X-linked)*	*PRPS1*	398A>C, 455T>C	Other or Mixed Ethnicity	1 in 500000	10%	1 in 555555
Aspartylglycosaminuria	*AGA*	200_201delAG, 214T>C, 488G>C	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Ataxia-telangiectasia	*ATM*	103C>T, 1120C>T, 1564_1565delGA, 2251-10T>G, 2806_2809dupCTAG, 3576G>A, 3894dupT, 4507C>T, 4852C>T, 5908C>T, 5932G>T, 6095G>A, 7010_7011delGT, 7327C>T, 7449G>A, 7517_7520delGAGA, 7638_7646delTAGAATTTC, 7926A>C, 8201_8211delTGTGTAATACAinsGACCTG, 8264_8268delATAAG, 8494C>T	Other or Mixed Ethnicity	1 in 100	10%	1 in 111
Ataxia-telangiectasia-like disorder	*MRE11A*	630G>C	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Ataxia with vitamin E deficiency	*TTPA*	303T>G, 400C>T, 513_514insTT, 744delA	Caucasian/White Other or Mixed Ethnicity	1 in 500 1 in 500	45% 10%	1 in 907 1 in 555
Autoimmune polyglandular syndrome, type I	*AIRE*	254A>G, 415C>T, 769C>T, 967_979delCTGTCCCCTCCGC,	Other or Mixed Ethnicity	1 in 167	72%	1 in 594
Autosomal recessive polycystic kidney disease	*PKHD1*	10412T>G, 107C>T, 1486C>T	Other or Mixed Ethnicity	1 in 70	19%	1 in 86
Bardet-Biedl syndrome, *BBS1*-related	*BBS1*	1169T>G	Other or Mixed Ethnicity	1 in 328	46%	1 in 607
Bardet-Biedl syndrome, *BBS10*-related	*BBS10*	271dupT	Other or Mixed Ethnicity	1 in 354	46%	1 in 655
Bardet-Biedl syndrome, *BBS12*-related	*BBS12*	1063C>T, 1115_1116delTT	Other or Mixed Ethnicity	1 in 708	10%	1 in 787
Bartter syndrome, type IV	*BSND*	1A>T, 28G>A	Other or Mixed Ethnicity	1 in 500	25%	1 in 666
Beta-ketothiolase deficiency	*ACAT1*	814C>T	Other or Mixed Ethnicity	1 in 500	10%	1 in 555

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Beta-thalassemia	*HBB*	-138C>T, -78A>G/-28A>G, -79A>G/-29A>G, 118C>T, 126_129delCTTT, 135delC, 25_26delAA, 27dupG, 315+1G>A, 316-2A>C, 316-2A>G, 47G>A, 51delC, 52A>T, 59A>G, 75T>A, 92+1G>A, 92+5G>C, 92+6T>C, 93-1G>A, 93-21G>A	African American/Black Asian Hispanic Other or Mixed Ethnicity	1 in 11 1 in 25 1 in 30 1 in 160	91% 70% 80% 69%	1 in 106 1 in 82 1 in 146 1 in 514
Bilateral frontoparietal polymicrogyria	*GPR56*	1036T>A	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Biotinidase deficiency	*BTD*	1368A>C, 1595C>T, 1612C>T, 755A>G, 98_104delGCGGCTGinsTCC	Other or Mixed Ethnicity	1 in 120	56%	1 in 273
Bloom syndrome	*BLM*	2281Del6/Ins7, 2407dupT	Ashkenazi Jewish Other or Mixed Ethnicity	1 in 100 1 in 500	99% 10%	1 in 9901 1 in 555
Canavan disease	*ASPA*	IVS2-2A>G, p.A305E, p.E285A, p.Y231X, p.Y288C	Ashkenazi Jewish Other or Mixed Ethnicity	1 in 41 1 in 158	98% 52%	1 in 2001 1 in 328
Carnitine palmitoyltransferase I deficiency	*CPT1A*	1079A>G, 1241C>T, 1361A>G, 1436C>T, 1493A>G, 2126G>A, 2129G>A	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Carnitine palmitoyltransferase II deficiency	*CPT2*	1145G>A, 1148T>A, 1239_1240delGA, 149C>A, 1507C>T, 1646G>A, 1883A>C, 1891C>T, 338C>T, 359A>G, 370C>T, 452G>A, 520G>A, 680C>T, 983A>G, p.Leu178_Ile186delinsPhe	Other or Mixed Ethnicity	1 in 500	62%	1 in 1314
Carpenter syndrome	*RAB23*	434T>A	Other or Mixed Ethnicity	1 in 500	72%	1 in 1783
Cartilage-hair hypoplasia	*RMRP*	c.262G>T, g.70A>G	Other or Mixed Ethnicity	1 in 500	48%	1 in 961
Charcot-Marie-Tooth disease, *GJB1*-related (X-linked)*	*GJB1*	164C>T, 187G>A, 223C>T, 225delG, 283G>A, 415G>A, 43C>T, 658C>T, c.536G>A, c.556G>A	Other or Mixed Ethnicity	1 in 20,625	10%	1 in 22917
Charcot-Marie-Tooth disease, *PRPS1*-related (X-linked)*	*PRPS1*	344T>C	Other or Mixed Ethnicity	1 in 500000	10%	1 in 555555
Chediak-Higashi syndrome	*LYST*	118dupG, 1467delG, 148C>T, 2623delT, 3085C>T, 9590delA	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Cholesteryl ester storage disease	*LIPA*	599T>C, p.G87V	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Choroideremia (X-linked)*	*CHM*	1609+2insT	Other or Mixed Ethnicity	1 in 29,000	10%	1 in 32222
Citrullinemia, type I	*ASS1*	1168G>A, 421-2A>G, 910C>T	Asian Other or Mixed Ethnicity	1 in 120 1 in 120	15% 40%	1 in 141 1 in 199
Congenital amegakaryocytic thrombocytopenia	*MPL*	IVS1+2T>A	Ashkenazi Jewish Other or Mixed Ethnicity	1 in 75 1 in 500	99% 10%	1 in 7,401 1 in 555
Congenital disorder of glycosylation, type IA	*PMM2*	338C>T, 357C>A, 422G>A, 691G>A	Caucasian/White Other or Mixed Ethnicity	1 in 71 1 in 71	75% 10%	1 in 281 1 in 79
Congenital disorder of glycosylation, type IB	*MPI*	884G>A	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Congenital lipoid adrenal hyperplasia	*STAR*	201_202delCT, 772C>T	Other or Mixed Ethnicity	1 in 500	10%	1 in 555

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Congenital neutropenia, *HAX1*-related	*HAX1*	568C>T	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Corneal dystrophy and perceptive deafness syndrome	*SLC4A11*	2528T>C	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Corticosterone methyloxidase deficiency	*CYP11B2*	541C>T	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Creatine transporter defect, *SLC6A8*-related* (X-linked)	*SLC6A8*	1006_1008delAAC, c.1222_1224delTTC	Other of Mixed Ethnicity	1 in 500000	10%	1 in 555555
Crigler-Najjar syndrome	*UGT1A1*	1070A>G	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Cystic fibrosis (600 mutations)	*CFTR*	1013delAA, 1058delC, 1078delT, 1112delT, 1119delA, 1138insG, 1150delA, 1154insTC, 1161delC, 1161insG, 1199delG, 1213delT, 1215delG, 1248+1G>A, 1259insA, 1283delA, 1289insTA, 1291delTT, 1309delG, 1323insA, 1341+1G>A, 1366delG, 1367del5, 1367delC, 1460delAT, 1461ins4, 1471delA, 1497delGG, 1504delG, 1525-1G>A, 1540del10, 1548delG, 1565delCA, 1571delG, 1576insT, 1601delTC, 1660delG, 1677delTA, 1717-1G>A, 1717-8G>A, 1742delAC, 1749insTA, 1774delCT, 1782delA, 1784delG, 1787delA, 1802delC, 1806delA, 1811+1.6kbA>G, 1811+1G>C, 1812-1G>A, 1813insC, 1824delA, 1833delT, 1845delAG/1846delGA, 1870delG, 1874insT, 1898+1G>A, 1898+1G>C, 1898+3A>G, 1898+5G>A, 1949del84, 2051delTT, 2113delA, 2116delCTAA, 211delG, 2141insA, 2143delT, 2176insC, 2183delAA>G, 2184delA, 2184insA, 2185insC, 2307insA, 2347delG, 237insA, 2406delCC, 2409delC, 241delAT, 2456delAC, 2512delG, 2522insC, 2556insAT, 2566insT, 2585delT, 2603delT, 2622+1G>A, 2634delT, 2634insT, 2640delT, 2694delT, 2711delT, 2721del11, 2723delTT, 2732insA, 2734G>AT, 2747delC, 2766del8, 2789+5G>A, 2790-1G>C, 284delA, 2896insAG, 2907delTT, 2909delT, 2942insT, 2948AT>C, 295ins8, 297-1G>A, 3007delG, 300delA, 3028delA, 3041delG, 3056delGA, 306delTAGA, 306insA, 3079delTT, 3100insA, 3120+1G>A, 3120G>A, 3121-1G>A, 3126del4, 3132delTG, 3171delC, 3171insC, 3173delAC, 3199del6, 3200_3204delTAGTG, 3238delA, 3272-26A>G, 3293delA, 3320ins5, 3359delCT, 3396delC, 3419delT, 3423delC, 3425delG, 3447delG, 347delC, 3532AC>GTA, 3577delT, 360_365insT, 360delT, 3617delGA, 3622insT, 3629delT, 3659delC, 3662delA, 3667ins4, 3670delA, 3724delG, 3732delA, 3737delA, 3750delAG, 3755delG, 3789insA, 3791delC, 379_381insT, 3821delT, 3849+10kbC>T, 3876delA, 3878delG, 3898insC, 3905insT, 3906insG, 3944delGT, 394delTT, 3960_3961delA, 4005+1G>A, 4006delA, 4010del4, 4015delA, 4016insT, 4022insT, 4040delA, 4048insCC, 405+1G>A, 406-1G>A, 4089ins4, 412del7- >TA, 4203TAG>AA, 4209TGTT>AA, 4218insT, 435insA, 4382delA, 441delA, 444delA, 451del8, 457TAT>G,	Ashkenazi Jewish Asian African American/Black Caucasian/White Hispanic Other or Mixed Ethnicity	1 in 24 1 in 94 1 in 61 1 in 25 1 in 58 1 in 30	99% 56% 82% 95% 84% N/A	1 in 2301 1 in 212 1 in 334 1 in 481 1 in 357 N/A

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		458delAT, 460delG, 489delC, 519delT, 525delT, 541del4, 541delC, 546insCTA, 547insGA, 547insTA, 556delA, 557delT, 565delC, 574delA, 5T, 605insT, 621+1G>T, 657delA, 663delT, 675del4, 681delC, 710_711+5del7, 711+1G>T, 711+3A>G, 711+5G>A, 712-1G>T, 733delG, 852del22, 874insTACA, 905delG, 935delA, 989_992insA, 991del5, 9T, [delta]F311, [delta]F508, [delta]I507, A455E, A46D, A559T, A561E, c.1086T>A, c.1086T>G, c.1111dupA, c.1114C>T, c.1227delT, c.1353_1354insT, c.1469delT, c.1645_1648delAGTG, c.1660_1661insA, c.198_202delTCCTA, c.2044delA, c.234delC, c.2554dupT, c.2566_2567insT, c.264_268delATATT, c.2686_2687insT, c.2705delG, c.2789delG, c.2803_2813delCTACCACTGGT, c.2876delC, c.3180delA, c.3263dupA, c.3708delT, c.3835_3836delTT, c.3855delC, c.3908dupA, c.405_406dupAC, c.650_659delAGTTGTTACA, c.708delT, c.714delT, c.717delG, c.825C>G, C128X, C225X, C276X, C343X, C491X, C76X, C831X, CFTRdele2.3, CFTRdele22.23, D110H, D1152H, E1104X, E115X, E116X, E1308X, E1371X, E1418X, E193X, E217X, E257X, E264X, E267X, E286X, E292X, E33X, E379X, E384X, E391X, E395X, E402X, E403X, E407X, E410X, E449X, E474X, E476X, E479X, E504X, E514X, E51X, E535X, E543X, E54X, E56X, E585X, E588X, E60X, E656X, E664X, E7X, E822X, E823X, E827X, E831X, E838X, E883X, E92K, E92X, F1074L, F508C, G1003X, G103X, G1061R, G1244E, G1349D, G149X, G178R, G178X, G194X, G226X, G27X, G330X, G366X, G404X, G451X, G458X, G461X, G473X, G486X, G542X, G545X, G550X, G551D, G551S, G576X, G673X, G85E, G85X, G885X, G970R, H1054D, H199Y, I336K, K114X, K1177X, K14X, K162X, K163X, K174X, K190X, K246X, K254X, K273X, K294X, K298X, K329X, K370X, K377X, K381X, K411X, K413X, K442X, K447X, K481X, K483X, K503X, K52X, K536X, K564X, K64X, K65X, K688X, K68X, K710X, K830X, K857X, K946X, K978X, L101X (TAA), L101X (TGA), L1059X, L1065P, L1077P, L1254X, L127X (TAA), L127X (TGA), L159X, L197X, L206W, L206X, L218X, L219X (T>A), L219X (T>G), L288X (TAA), L288X (TGA), L320X (TAA), L320X (TGA), L34X, L375X, L383X, L387X (TAA), L387X (TGA), L408X (TAA), L408X (TGA), L453X, L454X, L467P, L558X (TAA), L558X (TGA), L568X, L570X (TAA), L570X (TGA), L571X (TAA), L571X (TGA), L581X (TAA), L581X (TGA), L732X, L859X (TAA), L859X (TGA), L867X, L88X (T>A), L88X (T>G), L90X (TAA), L90X (TGA), L927P, L94X, M1101K, M1K, M1L, M1T, M1V, N1303K, P205S, P67L, Q1071X, Q1186X, Q1281X, Q1291X, Q1313X, Q179K, Q179X, Q207X, Q220X, Q237X, Q270X, Q290X, Q2X, Q30X, Q353X, Q359X, Q376X, Q378X, Q39X, Q414X, Q452X, Q493X, Q552X, Q634X, Q637X, Q685X, Q781X, Q814X, Q890X, Q98X, R104X, R1066C, R1066H, R1102X, R1128X, R1158X, R1162X,				

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		R117C, R117H, R242X, R258X, R289X, R29X, R303X, R334W, R347H, R347P, R352Q, R450X, R487X, R516X, R532X, R553X, R555X, R560K, R560T, R59X, R709X, R75X, R764X, R785X, R792X, R80X, R851X, S1196X, S1206X (TAA), S1206X (TGA), S1251N, S1255P, S1255X (TAA), S1255X (TGA), S263X (TAA), S263X (TGA), S308X, S313X, S341P, S341X, S35X (TAA), S35X (TGA), S434X, S466X (TAA), S466X (TGA), S478X (TAA), S478X (TGA), S489X, S492F, S4X, S549N, S549R (AGG), S549R (CGT), S63X, S776X, S912X, S945L, T338I, T388X, V520F, W1063X, W1089X, W1098X (TGA), W1098X(TAG), W1204X (TAG), W1204X (TGA), W1274X, W1282X, W1310X, W1316X, W216X, W277X, W356X (TAG), W356X (TGA), W361X, W401X (TAG), W401X (TGA), W57X (TGA), W57X(TAG), W679X, W79X (TAG), W79X (TGA), W846X, W846X (2670TGG>TGA), W865X, W882X, Y1092X (TAA), Y1092X (TAG), Y109X, Y1182X, Y122X, Y1307X, Y161X (TAA), Y161X (TAG), Y247X (TAA), Y247X (TAG), Y28X, Y301X, Y304X, Y325X, Y380X, Y385X (TAA), Y385X (TAG), Y38X, Y512X, Y563X, Y569D, Y569X, Y577X (TAA), Y577X (TAG), Y849X, Y84X, Y852X, Y89X, Y913X				
Cystinosis	*CTNS*	deletion 27 bp nt. 898-IVS8+24	Other or Mixed Ethnicity	1 in 194	10%	1 in 215
D-bifunctional protein deficiency	*HSD17B4*	1369A>T, 46G>A	Other or Mixed Ethnicity	1 in 158	35%	1 in 243
Desbuquois dysplasia, type I	*CANT1*	676G>A, 899G>A	Other or Mixed Ethnicity	1 in 500	22%	1 in 641
Dihydrolipoamide dehydrogenase deficiency	*DLD*	p.G229C, p.Y35X	Ashkenazi Jewish Other or Mixed Ethnicity	1 in 107 1 in 500	95% 10%	1 in 2,121 1 in 555
Dihydropyrimidine dehydrogenase deficiency	*DPYD*	1905+1G>A	Other or Mixed Ethnicity	1 in 50	52%	1 in 103
Du Pan syndrome	*GDF5*	1133G>A, 1322T>C	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Dyskeratosis congenita, *RTEL1*-related	*RTEL1*	R1264H	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Dyskeratosis congenita, *DKC1*-related (X-linked)*	*DKC1*	1058C>T, 109_111delCTT, 1205G>A, 146C>T, 472C>T, 91C>A	Other or Mixed Ethnicity	1 in 2500000	40%	1 in 4166666
Dystrophic epidermolysis bullosa, *COL7A1*-related	*COL7A1*	2305_2314delGTGAGGACTGinsTT, 6527dupC, c.4899+1G>A, p.Arg2063Trp	Other or Mixed Ethnicity	1 in 345	10%	1 in 383
Early onset myopathy with fatal cardiomyopathy	*TTN*	106668delA, 97824_97831delAGTGACCA	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Ehlers Danlos syndrome, type VIIC	*ADAMTS2*	673C>T	Other or Mixed Ethnicity	1 in 500	83%	1 in 2936
Emery-Dreifuss muscular dystrophy (X-linked)*	*EMD*	c.130C>T	Other or Mixed Ethnicity	1 in 82000	10%	1 in 91111
Enhanced S-cone syndrome	*NR2E3*	932G>A	Other or Mixed Ethnicity	1 in 500	44%	1 in 892
Ethylmalonic encephalopathy	*ETHE1*	221dupA, 487C>T, 505+1G>T	Other or Mixed Ethnicity	1 in 500	38%	1 in 806

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Fabry disease (X-linked)*	*GLA*	101A>G, 1025G>A, 272T>C, 335G>A, 337T>C, 427G>C, 644A>G, 679C>T, 680G>A, 888G>A, 902G>A, 982G>A	Other or Mixed Ethnicity	1 in 25000	10%	1 in 27778
Familial dysautonomia	*IKBKAP*	IVS20+6T>C, p.R696P	Ashkenazi Jewish	1 in 31	99%	1 in 3,001
			Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Familial hyperinsulinism, *ABCC8*-related	*ABCC8*	c.3992-9G>A, delF1388	Ashkenazi Jewish	1 in 52	97%	1 in 1,701
			Other or Mixed Ethnicity	1 in 166	10%	1 in 184
Familial hyperinsulinism, *KCNJ11*-related	*KCNJ11*	158G>A, 175G>A, 176T>G, 499A>C, 601C>T, 602G>A	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Familial Mediterranean fever	*MEFV*	1958G>A, 2040G>A, 2076_2078delAAT, 2080A>G, 2082G>A, 2084A>G, 2177T>C, 2230G>T, 2282G>A	Ashkenazi Jewish	1 in 81	64%	1 in 223
			Other or Mixed Ethnicity	1 in 50	10%	1 in 55
Fanconi anemia, type A	*FANCA*	1115_1118delTTGG, 295C>T, 3398delA	Other or Mixed Ethnicity	1 in 248	10%	1 in 275
Fanconi anemia, type C	*FANCC*	c.322delG, IVS4+4A>T, p.R548X	Ashkenazi Jewish	1 in 89	99%	1 in 8801
			Other or Mixed Ethnicity	1 in 535	10%	1 in 594
Fetal akinesia deformation sequence, *DOK7*-related	*DOK7*	331+1G>T	Other or Mixed Ethnicity	1 in 1119	10%	1 in 1243
Fragile X syndrome (X-linked)*	*FMR1*	CGG repeat analysis	Other or Mixed Ethnicity	1 in 259	99%	1 in 25801
Fumarase deficiency	*FH*	1431_1433dupAAA	Other or Mixed Ethnicity	1 in 500	21%	1 in 633
Galactokinase deficiency	*GALK1*	1144C>T, 238G>T, 82C>A, 94G>A	Other or Mixed Ethnicity	1 in 158	10%	1 in 175
Galactosemia	*GALT*	253-2A>G, 404C>T, 512T>C, 563A>G, 584T>C, 626A>G, 855G>T	Ashkenazi Jewish	1 in 110	10%	1 in 122
			Other or Mixed Ethnicity	1 in 110	90%	1 in 1091
Gaucher disease	*GBA*	IVS2+1G>A, p.D409H, p.D409V, p.L444P, p.L47X, p.N370S, p.R496H, p.V394L	Ashkenazi Jewish	1 in 18	95%	1 in 341
			Other or Mixed Ethnicity	1 in 112	60%	1 in 279
Geroderma osteodysplastica	*SCYL1BP1*	Glu123Ter	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Gitelman syndrome	*SLC12A3*	1180+1G>T	Other or Mixed Ethnicity	1 in 100	10%	1 in 111
Glucose-6-phosphate dehydrogenase deficiency (X-linked)*	*G6PD*	1003G>A, 131C>G, 1376G>C, 1376G>T, 202G>A, 376A>G, 487G>A, 563C>T, 871G>A	African American/Black	1 in 5	95%	1 in 81
			Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Glutaric acidemia, type I	*GCDH*	1204C>T, 1262C>T, 680G>C	Caucasian/White	1 in 94	36%	1 in 146
			Other or Mixed Ethnicity	1 in 94	10%	1 in 104
Glycine encephalopathy, *AMT*-related	*AMT*	125A>G, 139G>A, 878-1G>A, 959G>A	Other or Mixed Ethnicity	1 in 316	50%	1 in 631
Glycine encephalopathy, *GLDC*-related	*GLDC*	1545G>C, 1691G>T, 2281G>A, 2267_2269delTCT	Other or Mixed Ethnicity	1 in 125	10%	1 in 139
Glycogen storage disease, type lA	*G6PC*	p.Q347X, p.R83C	Ashkenazi Jewish	1 in 71	99%	1 in 7,001
			Caucasian/White	1 in 177	53%	1 in 375
			Other or Mixed Ethnicity	1 in 177	10%	1 in 197

Disorder	Gene	Variants	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
Glycogen storage disease, type IB	*SLC37A4*	1015G>T, 1211delCT, 352T>C	Caucasian/White	1 in 354	46%	1 in 655
			Other or Mixed Ethnicity	1 in 354	10%	1 in 393
Glycogen storage disease, type II	*GAA*	2560C>T, 2707_2709delAAG, -32-13T>G, c.1935C>A, 525delT	African American/Black	1 in 60	60%	1 in 149
			Other or Mixed Ethnicity	1 in 100	10%	1 in 111
Glycogen storage disease, type III	*AGL*	1222C>T, 16C>T, 18_19delGA, 2039G>A, 2590C>T, 3682C>T, 3965delT, 4260-12A>G, 4456delT	Other or Mixed Ethnicity	1 in 158	45%	1 in 286
Glycogen storage disease, type IV	*GBE1*	986A>C	Ashkenazi Jewish	1 in 388	94%	1 in 6451
			Other or Mixed Ethnicity	1 in 388	10%	1 in 431
Glycogen storage disease, type V	*PYGM*	148C>T, 1627A>T, 1628A>C, 1827G>A, 2128_2130delTTC, 2392T>C, 255C>A, 613G>A	Other or Mixed Ethnicity	1 in 158	80%	1 in 786
Glycogen storage disease, type VII	*PFKM*	2003delC, 237+1G>A	Ashkenazi Jewish	1 in 500	94%	1 in 8318
			Other or Mixed Ethnicity	1 in 500	10%	1 in 555
GM1-gangliosidosis	*GLB1*	1445G>A, 152T>C, 176G>A, 622C>T	Other or Mixed Ethnicity	1 in 224	10%	1 in 249
GRACILE syndrome	*BCS1L*	232A>G	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Guanidinoacetate methyltransferase deficiency	*GAMT*	59G>C, c.299_311dupGGGACTGGGCCCC, c.327G>A	Other or Mixed Ethnicity	1 in 500	45%	1 in 908
Hemoglobinopathy evaluation	*N/A*	N/A	Other or Mixed Ethnicity	--	99%	--
Hemoglobinopathy, Hb C	*HBB*	19G>A	African American/Black	1 in 48	99%	1 in 4701
			Asian	1 in 20000	99%	1 in 1999901
			Caucasian/White	1 in 2521	99%	1 in 252001
			Hispanic	1 in 1417	99%	1 in 141601
			Other or Mixed Ethnicity	1 in 500	99%	1 in 49901
Hemoglobinopathy, Hb D	*HBB*	364G>C	African American/Black	1 in 2129	99%	1 in 212801
			Asian	1 in 3614	99%	1 in 361301
			Caucasian/White	1 in 2391	99%	1 in 239001
			Hispanic	1 in 1635	99%	1 in 163401
			Other or Mixed Ethnicity	1 in 500	99%	1 in 49901
Hemoglobinopathy, Hb E	*HBB*	79G>A	African American/Black	1 in 1671	99%	1 in 167001
			Asian	1 in 124	99%	1 in 12301
			Caucasian/White	1 in 2227	99%	1 in 222601
			Hispanic	1 in 5154	99%	1 in 515301
			Other or Mixed Ethnicity	1 in 500	99%	1 in 49901
Hemoglobinopathy, Hb O	*HBB*	364G>A	Other or Mixed Ethnicity	1 in 500	99%	1 in 49901

Disorder	Gene	Variants	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
Hemophilia A (X-linked)*	*F8*	1033G>C, 1636C>T, 1648C>T, 1729T>C, 1750C>A, 1804C>T, 1834C>T, 2167G>A, 43C>T, 5122C>T, 5143C>T, 5305G>A, 5399G>A, 5422C>T, 5536A>T, 5822A>G, 5878C>T, 5953C>T, 6046C>T, 6278A>G, 6360T>G, 6413C>A, 6506G>A, 6532C>T, 6545G>A, 6670_6672delCCT, 6682C>T, 6683G>A, 6744G>T, 6967C>T, 902G>A	Other or Mixed Ethnicity	1 in 2000	10%	1 in 2,222
Hemophilia B (X-linked)*	*F9*	1025C>T, 1328T>C, 223C>T, 316G>A	Other or Mixed Ethnicity	1 in 10000	41%	1 in 16948
Hepatocerebral mitochondrial DNA depletion syndrome, *MPV17*-related	*MPV17*	148C>T, 149G>A, 271_273delTTG, 359G>A, 498C>A, 70G>T	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Hermansky-Pudlak syndrome, type III	*HPS3*	1163+1G>A, 1189C>T, 1691+2T>G	Ashkenazi Jewish Other or Mixed Ethnicity	1 in 235 1 in 982	80% 10%	1 in 1171 1 in 1091
Holocarboxylase synthetase deficiency	*HLCS*	1522C>T, 1648G>A	Other or Mixed Ethnicity	1 in 224	25%	1 in 298
Homocystinuria, *CBS*-related	*CBS*	1058C>T, 833T>C, 919G>A	Other or Mixed Ethnicity	1 in 224	33%	1 in 334
Hyperphosphatemic familial tumoral calcinosis	*GALNT3*	1524+1G>A, 516-2A>T	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Hypohidrotic ectodermal dysplasia	*EDAR*	1144G>A	Other or Mixed Ethnicity	1 in 189	10%	1 in 210
Hypophosphatasia	*ALPL*	1001G>A, 1133A>T, 1559delT, 571G>A, 979T>C	Caucasian/White Other or Mixed Ethnicity	1 in 158 1 in 158	31% 10%	1 in 229 1 in 175
Isovaleric acidemia	*IVD*	941C>T	Other or Mixed Ethnicity	1 in 250	47%	1 in 471
Joubert syndrome 2	*TMEM216*	p.R12L	Ashkenazi Jewish Other or Mixed Ethnicity	1 in 92 1 in 500	95% 10%	1 in 1,821 1 in 555
Junctional epidermolysis bullosa, *LAMB3*-related	*LAMB3*	124C>T, 1903C>T, 727C>T	Other or Mixed Ethnicity	1 in 895	48%	1 in 1720
Juvenile retinoschisis (X-linked)*	*RS1*	214G>A, 221G>T, 325G>C	Other or Mixed Ethnicity	1 in 7500	10%	1 in 8333
Krabbe disease	*GALC*	1472delA, 1586C>T, 1630G>A, 1657G>A, 1700A>C, 1796T>G, 857G>A, c.1161+6532_polyA+9kbdel, c.121G>A, c.683_694delATCTCTGGGAGTinsCTC, c.749T>C	Ashkenazi Jewish Caucasian/White Hispanic Other or Mixed Ethnicity	1 in 158 1 in 158 1 in 158 1 in 158	73% 73% 35% 73%	1 in 582 1 in 582 1 in 243 1 in 582
Leber congenital amaurosis, *LCA5*-related	*LCA5*	1151delC, 1476dupA, 835C>T	Other or Mixed Ethnicity	1 in 643	10%	1 in 714
Leber congenital amaurosis, *RDH12*-related	*RDH12*	806_810delCCCTG	Other or Mixed Ethnicity	1 in 455	28%	1 in 632
Leigh syndrome, French Canadian	*LRPPRC*	1061C>T	Other or Mixed Ethnicity	1 in 100	10%	1 in 111
Limb-girdle muscular dystrophy, type 2A	*CAPN3*	1469G>A, 2306G>A, 550delA	Caucasian/White Other or Mixed Ethnicity	1 in 160 1 in 160	66% 10%	1 in 469 1 in 178
Limb-girdle muscular dystrophy, type 2D	*SGCA*	229C>T	Other or Mixed Ethnicity	1 in 126	32%	1 in 185

Disorder	Gene	Variants	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
Limb-girdle muscular dystrophy, type 2E	*SGCB*	341C>T, 377_384dupCAGTAGGA	Other or Mixed Ethnicity	1 in 126	25%	1 in 168
Limb-girdle muscular dystrophy, type 2I	*FKRP*	826C>A	Other or Mixed Ethnicity	1 in 246	35%	1 in 378
Lipoprotein lipase deficiency	*LPL*	644G>A	Other or Mixed Ethnicity	1 in 500	24%	1 in 658
Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency	*HADHA*	1528G>C	Other or Mixed Ethnicity	1 in 250	87%	1 in 1916
Luteinizing hormone resistance	*LHCGR*	1060G>A, 1660C>T, 1777G>C	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Maple syrup urine disease, type IA	*BCKDHA*	p.Y438N	Other or Mixed Ethnicity	1 in 321	10%	1 in 357
Maple syrup urine disease, type IB	*BCKDHB*	p.E372X, p.G278S, p.R183P	Ashkenazi Jewish Other or Mixed Ethnicity	1 in 97 1 in 364	99% 10%	1 in 9601 1 in 404
Medium-chain acyl-CoA dehydrogenase deficiency	*ACADM*	362C>T, 583G>A, 799G>A, 985A>G	Caucasian/White Other or Mixed Ethnicity	1 in 50 1 in 62	74% 10%	1 in 189 1 in 69
MEDNIK syndrome	*AP1S1*	183-2A>G	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Metachromatic leukodystrophy	*ARSA*	1136C>T, 1232C>T, 1283C>T, 1408_1418delGCAGCTGTGAC, 257G>A, 293C>T, 542T>G, 641C>T, 739G>A, 769G>C, c.465+1G>A, c.302G>A, p.T274M	Other or Mixed Ethnicity	1 in 100	47%	1 in 188
Methylmalonic aciduria, cblA type	*MMAA*	1076G>A, 161G>A, 266T>C, 283C>T, 358C>T, 397C>T, 503delC, 562G>C, 64C>T, 650T>A, 653G>A, 733+1G>A, 988C>T	Other or Mixed Ethnicity	1 in 224	19%	1 in 276
Methylmalonic aciduria, cblB type	*MMAB*	197-1G>T, 287T>C, 291-1G>A, 556C>T, 568C>T, 569G>A, 571C>T, 572G>A, 700C>T	Other or Mixed Ethnicity	1 in 323	66%	1 in 948
Methylmalonic aciduria, cblC type	*MMACHC*	271dupA, 331C>T, 394C>T	Other or Mixed Ethnicity	1 in 224	62%	1 in 588
Methylmalonic aciduria, *MUT*-related	*MUT*	1097A>G, 1105C>T, 1106G>A, 1280G>A, 1867G>A, 2054T>G, 2150G>T, 278G>A, 281G>T, 299A>G, 313T>C, 322C>T, 521T>C, 572C>A, 607G>A, 643G>A, 655A>T, 691T>A, 935G>T	African American/Black Asian Caucasian/White Hispanic Other or Mixed Ethnicity	1 in 145 1 in 145 1 in 145 1 in 145 1 in 145	55% 17% 23% 52% 10%	1 in 318 1 in 174 1 in 188 1 in 301 1 in 161
Mitochondrial complex IV deficiency	*PET100*	3G>C	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Mitochondrial myopathy and sideroblastic anemia	*PUS1*	430C>T	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Mucolipidosis, type II/III alpha/beta	*GNPTAB*	1120T>C, 3503_3504delTC, 3565C>T	Other or Mixed Ethnicity	1 in 158	25%	1 in 210
Mucolipidosis, type IV	*MCOLN1*	del6.4kb, IVS3-2A>G	Ashkenazi Jewish Other or Mixed Ethnicity	1 in 127 1 in 500	95% 10%	1 in 2521 1 in 555
Mucopolysaccharidosis, type I Hurler syndrome	*IDUA*	1598C>G, 192C>A, 979G>C, p.R89Q, p.Q70X, Trp402Ter	Caucasian/White Other of Mixed Ethnicity	1 in 158 1 in 158	83% 10%	1 in 925 1 in 175

Disorder	Gene	Variants	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
Mucopolysaccharidosis, type II Hunter syndrome (X-linked)*	*IDS*	1327C>T, 1402C>T, 1403G>A, 1403G>T, 998C>T	Other or Mixed Ethnicity	1 in 50000	14%	1 in 57836
Mucopolysaccharidosis, type IIIC	*HGSNAT*	1553C>T, 234+1G>A, 372-2A>G	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Mucopolysaccharidosis, type VI	*ARSB*	1143-1G>C, 1143-8T>G, 629A>G	Other or Mixed Ethnicity	1 in 250	14%	1 in 291
Mulibrey nanism	*TRIM37*	2212delG, 493-2A>G, 860G>A	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Multiple sulfatase deficiency	*SUMF1*	1033C>T, 463T>C, 519+5_519+8delGTAA, 788G>T, 836C>T	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Muscle-eye-brain disease	*POMGNT1*	1539+1G>A	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Myotubular myopathy, *MTM1*-related (X-linked)*	*MTM1*	109C>T, 1261-10A>G, 1261C>T, 1262G>A, 142G>T, 205C>T, 614C>T, 721C>T	Other or Mixed Ethnicity	1 in 25000	21%	1 in 31645
Nemaline myopathy 2	*NEB*	p.R2478_D2512del	Ashkenazi Jewish Other or Mixed Ethnicity	1 in 108 1 in 500	99% 10%	1 in 10,701 1 in 555
Nephrotic syndrome, type I	*NPHS1*	121_122delCT, 1481delC, 3325C>T	Other or Mixed Ethnicity	1 in 91	10%	1 in 101
Neuronal ceroid lipofuscinosis, *CLN5*-related	*CLN5*	1175_1176delAT	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Neuronal ceroid lipofuscinosis, *CLN6*-related	*CLN6*	214G>T, 461_463delTCA, c.316dupC	Other or Mixed Ethnicity	1 in 430	10%	1 in 478
Neuronal ceroid lipofuscinosis, *CLN8*-related	*CLN8*	70C>G, 789G>C	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Neuronal ceroid-lipofuscinosis, *MFSD8*-related	*MFSD8*	754+2T>A, 881C>A	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Neuronal ceroid lipofuscinosis, *PPT1*-related	*PPT1*	223A>C, 364A>T, 451C>T	Other or Mixed Ethnicity	1 in 500	63%	1 in 1350
Neuronal ceroid lipofuscinosis, *TPP1*-related	*TPP1*	509-1G>C, 622C>T, 851G>T	Other or Mixed Ethnicity	1 in 300	53%	1 in 637
Niemann-Pick disease, type A and B	*SMPD1*	delR608, fs330, p.L302P, p.R496L	Ashkenazi Jewish Caucasian/White Other or Mixed Ethnicity	1 in 90 1 in 250 1 in 500	97% 20% 20%	1 in 2,968 1 in 312 1 in 625
Niemann-Pick disease, type CI	*NPC1*	3182T>C	Other or Mixed Ethnicity	1 in 199	20%	1 in 249
Niemann-Pick disease, type CII	*NPC2*	133C>T, 141C>A, 190+5G>A, 199T>C, 295T>C, 352G>T, 58G>T	Other or Mixed Ethnicity	1 in 866	10%	1 in 962
Nijmegen breakage syndrome	*NBN*	657_661delACAAA	Other or Mixed Ethnicity	1 in 158	70%	1 in 524
Nonsyndromic hearing loss, *GJB2*-related	*GJB2*	167delT, 229T>C, 235delC, 269T>C, 35delG, 71G>A	Ashkenazi Jewish Caucasian/White Other or Mixed Ethnicity	1 in 20 1 in 33 1 in 42	84% 78% 10%	1 in 120 1 in 147 1 in 47
Omenn syndrome	*DCLRE1C*	597C>A	Other or Mixed Ethnicity	1 in 500	10%	1 in 555

Disorder	Gene	Variants	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
Ornithine transcarbamylase deficiency (X-linked)*	*OTC*	274C>T, 421C>T, 77G>A	Other or Mixed Ethnicity	1 in 7000	10%	1 in 7778
Ornithine translocase deficiency	*SLC25A15*	535C>T, 562_564delTTC, 95C>G	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Pendred syndrome	*SLC26A4*	1001+1G>A, 1151A>G, 1246A>C, 1540C>A, 2168A>G, 919-2A>G	Caucasian/White Other or Mixed Ethnicity	1 in 298 1 in 298	39% 10%	1 in 488 1 in 331
Phenylalanine hydroxylase deficiency	*PAH*	1066-11G>A, 1222C>T, 1223G>A, 1315+1G>A, 721C>T, 856G>A	Other or Mixed Ethnicity	1 in 50	15%	1 in 59
Pontocerebellar hypoplasia, type IA	*VRK1*	1072C>T	Ashkenazi Jewish Other or Mixed Ethnicity	1 in 225 1 in 500	90% 10%	1 in 2241 1 in 555
Primary congenital glaucoma	*CYP1B1*	1103G>A, 1120G>A, 1159G>A, 1202_1211dupATGCCACCAC, 1267A>T, 1405C>T, 171G>A, 174delA, 182G>A, 241T>A, 4339G 1 bp del	Other or Mixed Ethnicity	1 in 92	47%	1 in 173
Primary hyperoxaluria, type I	*AGXT*	121G>A, 122G>T, 245G>A, 33dupC, 454T>A, 466G>A, 508G>A, 697C>T, 731T>C,	Other or Mixed Ethnicity	1 in 289	59%	1 in 703
Primary hyperoxaluria, type II	*GRHPR*	103delG, 404+3_404+6delAAGT	Asian Caucasian/White Other or Mixed Ethnicity	1 in 381 1 in 381 1 in 381	16% 58% 37%	1 in 453 1 in 906 1 in 604
Progressive familial intrahepatic cholestasis, type II	*ABCB11*	1723C>T, 890A>G	Other or Mixed Ethnicity	1 in 114	10%	1 in 127
Prolidase deficiency	*PEPD*	605C>T, 826G>A	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Propionic acidemia, *PCCA*-related	*PCCA*	1685C>G, 412G>A, 491T>C, 862A>G, 937C>T	Other or Mixed Ethnicity	1 in 112	10%	1 in 124
Propionic acidemia, *PCCB*-related	*PCCB*	1218_1231delGGGCATCATCCGGCinsTAGAGCACAGGA, 1228C>T, 1283C>T, 1495C>T, 1534C>T, 1538_1540dupCCC, 1606A>G, 280G>T, 335G>A, 457G>C, 502G>A	Caucasian Other or Mixed Ethnicity	1 in 112 1 in 112	30% 10%	1 in 160 1 in 124
Pseudoxanthoma elasticum	*ABCC6*	3413G>A, 3421C>T, 4015C>T	Other or Mixed Ethnicity	1 in 80	34%	1 in 121
Pycnodysostosis	*CTSK*	926T>C	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Pyruvate dehydrogenase deficiency, *PDHB*-related	*PDHB*	301A>G	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Pyruvate dehydrogenase deficiency, *PDHA1*-related (X-linked)*	*PDHA1*	4-BP INS: 1251ACTA, 615C>A, 787C>G, 904C>T	Other or Mixed Ethnicity	1 in 625000	10%	1 in 694444
Retinitis pigmentosa 59	*DHDDS*	124A>G	Ashkenazi Jewish Other or Mixed Ethnicity	1 in 322 1 in 296	99% 10%	1 in 32101 1 in 329
Rhizomelic chondrodysplasia punctata, type I	*PEX7*	875T>A	Other or Mixed Ethnicity	1 in 150	51%	1 in 305
Salla disease	*SLC17A5*	115C>T	Other or Mixed Ethnicity	1 in 500	10%	1 in 555

Disorder	Gene	Variants	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk	
Sandhoff disease	*HEXB*	1376A>C, 445+1G>A, 850C>T	Caucasian/White	1 in 180	29%	1 in 253	
			Other or Mixed Ethnicity	1 in 500	10%	1 in 555	
Severe combined immunodeficiency, *RAG1*-related	*RAG1*	1186C>T, 1566G>T, 1681C>T, 1682G>A, 256_257delAA, 631delT	Other or Mixed Ethnicity	1 in 430	11%	1 in 480	
Severe combined immunodeficiency, *IL2RG*-related (X-linked)*	*IL2RG*	854G>A, 865C>T	Other or Mixed Ethnicity	1 in 25,000	12%	1 in 28,409	
Shwachman-Diamond syndrome	*SBDS*	258+2T>C	Other or Mixed Ethnicity	1 in 140	46%	1 in 258	
Sickle cell anemia (Hb S)	*HBB*	20A>T	African American/Black	1 in 14	99%	1 in 1301	
			Asian	1 in 1336	99%	1 in 133501	
			Caucasian/White	1 in 625	99%	1 in 62401	
			Hispanic	1 in 183	99%	1 in 18201	
			Other or Mixed Ethnicity	1 in 25	99%	1 in 2401	
Sjögren-Larsson syndrome	*ALDH3A2*	1297_1298delGA, 943C>T	Caucasian/White	1 in 167	36%	1 in 260	
			Other or Mixed Ethnicity	1 in 167	10%	1 in 185	
Smith-Lemli-Opitz syndrome	*DHCR7*	IVS8-1G>C	Other or Mixed Ethnicity	1 in 87	28%	1 in 121	
Spastic ataxia of Charlevoix-Saguenay (ARSACS)	*SACS*	12160C>T, 7504C>T, 8844delT	Other or Mixed Ethnicity	1 in 500	10%	1 in 555	
Spinal muscular atrophy	*SMN1* *SMN2*	Copy number analysis				2 Copies *SMN1*	3 Copies *SMN1*
			African American/Black	1 in 66	71%	1 in 225	1 in 3000
			Ashkenazi Jewish	1 in 41	90%	1 in 401	1 in 4000
			Asian	1 in 53	93%	1 in 744	1 in 5000
			Caucasian/White	1 in 35	95%	1 in 681	1 in 3500
			Hispanic	1 in 117	91%	1 in 1290	1 in 11000
			Other or Mixed Ethnicity	1 in 50	--	--	--
Steroid-resistant nephrotic syndrome	*NPHS2*	412C>T, 413G>A	Other or Mixed Ethnicity	1 in 91	57%	1 in 210	
Stuve-Wiedemann syndrome	*LIFR*	653dupT	Other or Mixed Ethnicity	1 in 500	10%	1 in 555	
Sulfate transporter-related osteochondrodysplasia	*SLC26A2*	1957T>A, -26+2T>C, 532C>T, 835C>T, 1020_1022delTGT	Other or Mixed Ethnicity	1 in 100	70%	1 in 331	
Tay-Sachs disease (DNA)	*HEXA*	1278+TATC, IVS12+1G>C, IVS7+1G>A, IVS9+1G>A, p.G269S, p.R178H, p.R247W, p.R249W, del7.6kb	Ashkenazi Jewish	1 in 31	94%	1 in 501	
			Other or Mixed Ethnicity	1 in 300	59%	1 in 730	
Tay-Sachs disease (hexosaminidase A)	*N/A*	N/A	Ashkenazi Jewish	1 in 31	98%	1 in 1501	
			Other or Mixed Ethnicity	1 in 300	98%	1 in 14951	
Tumoral calcinosis, normophosphatemic	*SAMD9*	1030C>T, 4483A>G	Other or Mixed Ethnicity	1 in 500	10%	1 in 555	
Tyrosinemia, type I	*FAH*	1062+5G>A	Other or Mixed Ethnicity	1 in 100	28%	1 in 139	
Usher syndrome, type IB	*MYO7A*	1996C>T, 448C>T, 635G>A, 700C>T, 731G>C, 93C>A	Other or Mixed Ethnicity	1 in 169	10%	1 in 188	

Disorder	Gene	Variants	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
Usher syndrome, type IC	*USH1C*	216G>A, 238dupC	Other or Mixed Ethnicity	1 in 485	10%	1 in 539
Usher syndrome, type ID	*CDH23*	5237G>A, 6442G>A	Other or Mixed Ethnicity	1 in 277	10%	1 in 308
Usher syndrome, type IF	*PCDH15*	p.R245X	Ashkenazi Jewish Other or Mixed Ethnicity	1 in 147 1 in 428	75% 10%	1 in 585 1 in 475
Usher syndrome, type III	*CLRN1*	528T>G, p.N48K	Ashkenazi Jewish Other or Mixed Ethnicity	1 in 120 1 in 449	98% 10%	1 in 5951 1 in 499
Very long chain acyl-CoA dehydrogenase deficiency	*ACADVL*	1322G>A, 1405C>T, 779C>T	Other or Mixed Ethnicity	1 in 87	10%	1 in 97
Vitamin D-dependent rickets, type I	*CYP27B1*	262delG	Other or Mixed Ethnicity	1 in 278	10%	1 in 309
Walker-Warburg syndrome	*FKTN*	p.F390X	Ashkenazi Jewish Other or Mixed Ethnicity	1 in 150 1 in 500	95% 10%	1 in 2,981 1 in 555
Wilson disease	*ATP7B*	1934T>G, 2333G>T, 2906G>A, 3207C>A, 3443T>C, 3809A>G	Asian Other or Mixed Ethnicity	1 in 90 1 in 90	57% 40%	1 in 208 1 in 149
Zellweger syndrome spectrum, *PEX6*-related	*PEX6*	802_815delGACGGACTGGCGCT	Other or Mixed Ethnicity	1 in 300	10%	1 in 333

*X-linked disorder; carrier frequency and residual risk provided apply to females only. X-linked disorders are not tested in males.

Residual risk tables are calculated based on the variants tested and the currently available evidence. Residual risks may change given the dynamic nature of both the variants in this assay and ongoing literature updates.

Preparent® Global+ Panel

Includes all Global Panel disorders plus the following.

Disorder	Gene	Variants	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
Alpha-1 antitrypsin deficiency	*SERPINA1*	1096G>A, 227_229delTCT	Other or Mixed Ethnicity	1 in 36	95%	1 in 701
Autosomal recessive woolly hair/hypotrichosis	*LIPH*	736T>A	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Hereditary fructose intolerance	*ALDOB*	1005C>G, 360_363delCAAA, 448G>C, 524C>A	Other or Mixed Ethnicity	1 in 71	59%	1 in 172
Hereditary hemochromatosis, *HFE*-related	*HFE*	187C>G, 845G>A	Caucasian/White Hispanic Other or Mixed Ethnicity	1 in 3 1 in 5 1 in 10	87% 10% 10%	1 in 16 1 in 5 1 in 11
Hereditary hemochromatosis, *HFE2*-related	*HFE2*	959G>T	Other or Mixed Ethnicity	1 in 500	50%	1 in 999
Hereditary hemochromatosis, *TFR2*-related	*TFR2*	2069A>C, 515T>A, 750C>G	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Inclusion body myopathy, type II	*GNE*	1714G>C, 2135T>C, 38G>C	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Lamellar ichthyosis, type I	*TGM1*	424C>T, 425G>A, 428G>A, c.877-2A>G	Other or Mixed Ethnicity	1 in 224	34%	1 in 339
Limb-girdle muscular dystrophy, type 2B	*DYSF*	1566C>G, 2372C>G, 2875C>T, 2997G>T, 3137G>A, 3373delG, 4872_4876delGCCCGinsCCCC, 4872delG	Other or Mixed Ethnicity	1 in 270	10%	1 in 300
Pseudocholinesterase deficiency	*BCHE*	293A>G	Other or Mixed Ethnicity	1 in 28	72%	1 in 97
Spastic paraplegia, *ZFYVE26*-related	*ZFYVE26*	1477C>T, 4312C>T	Other or Mixed Ethnicity	1 in 500	10%	1 in 555
Xeroderma pigmentosum	*XPC*	1735C>T, 413-24A>G, 413-9T>A	Other or Mixed Ethnicity	1 in 895	10%	1 in 994

Residual risk tables are calculated based on the variants tested and the currently available evidence. Residual risks may change given the dynamic nature of both the variants in this assay and ongoing literature updates.

Preparent® Global/Global+ Panel

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