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Preparent® Standard Panel

Disorder	Gene	Variants	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
Alpha-thalassemia	*HBA1* *HBA2*	-alpha3.7, -alpha4.2, -alpha20.5, --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
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
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
Congenital amegakaryocytic thrombocytopenia	*MPL*	IVS1+2T>A	Ashkenazi Jewish Other or Mixed Ethnicity	1 in 75 1 in 500	99% 10%	1 in 7401 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,	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

Disorder	Gene	Variants	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
		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, 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,				

Disorder	Gene	Variants	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
		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, 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				

Disorder	Gene	Variants	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
		(TAG), Y849X, Y84X, Y852X, Y89X, Y913X				
Dihydrolipoamide dehydrogenase deficiency	*DLD*	p.G229C, p.Y35X	Ashkenazi Jewish Other or Mixed Ethnicity	1 in 107 1 in 500	95% 10%	1 in 2121 1 in 555
Familial dysautonomia	*IKBKAP*	IVS20+6T>C, p.R696P	Ashkenazi Jewish Other or Mixed Ethnicity	1 in 31 1 in 500	99% 10%	1 in 3001 1 in 555
Familial hyperinsulinism, *ABCC8*-related	*ABCC8*	c.3992-9G>A, delF1388	Ashkenazi Jewish Other or Mixed Ethnicity	1 in 52 1 in 166	97% 10%	1 in 1701 1 in 184
Fanconi anemia, type C	*FANCC*	c.322delG, IVS4+4A>T, p.R548X	Ashkenazi Jewish Other or Mixed Ethnicity	1 in 89 1 in 535	99% 10%	1 in 8801 1 in 594
Fragile X syndrome (X-linked)*	*FMR1*	CGG repeat analysis	Other or Mixed Ethnicity	1 in 259	99%	1 in 25801
Gaucher disease	*GBA*	IVS2+1G>A, p.D409H, p.D409V, p.L444P, p.L47X, p.N370S, p.R496H, p.V394L	Ashkenazi Jewish Other or Mixed Ethnicity	1 in 18 1 in 112	95% 60%	1 in 341 1 in 279
Glycogen storage disease, type IA	*G6PC*	p.Q347X, p.R83C	Ashkenazi Jewish Caucasian/White Other or Mixed Ethnicity	1 in 71 1 in 177 1 in 177	99% 53% 10%	1 in 7,001 1 in 375 1 in 197
Hemoglobinopathy, Hb C	*HBB*	19G>A	African American/Black Asian Caucasian/White Hispanic Other or Mixed Ethnicity	1 in 48 1 in 20000 1 in 2521 1 in 1417 1 in 500	99% 99% 99% 99% 99%	1 in 4701 1 in 1999901 1 in 252001 1 in 141601 1 in 49901
Hemoglobinopathy, Hb D	*HBB*	364G>C	African American/Black Asian Caucasian/White Hispanic Other or Mixed Ethnicity	1 in 2129 1 in 3614 1 in 2391 1 in 1635 1 in 500	99% 99% 99% 99% 99%	1 in 212801 1 in 361301 1 in 239001 1 in 163401 1 in 49901
Hemoglobinopathy, Hb E	*HBB*	79G>A	African American/Black Asian Caucasian/White Hispanic Other or Mixed Ethnicity	1 in 1671 1 in 124 1 in 2227 1 in 5154 1 in 500	99% 99% 99% 99% 99%	1 in 167001 1 in 12301 1 in 222601 1 in 515301 1 in 49901
Hemoglobinopathy, Hb O	*HBB*	364G>A	Other or Mixed Ethnicity	1 in 500	99%	1 in 49901
Joubert syndrome 2	*TMEM216*	p.R12L	Ashkenazi Jewish Other or Mixed Ethnicity	1 in 92 1 in 500	95% 10%	1 in 1821 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

Disorder	Gene	Variants	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk	
Mucolipidosis type IV	*MCOLN1*	del6.4kb, IVS3-2A>G	Ashkenazi Jewish	1 in 127	95%	1 in 2521	
			Other or Mixed Ethnicity	1 in 500	10%	1 in 555	
Nemaline myopathy 2	*NEB*	p.R2478_D2512del	Ashkenazi Jewish	1 in 108	99%	1 in 10701	
			Other or Mixed Ethnicity	1 in 500	10%	1 in 555	
Niemann-Pick disease, type A and B	*SMPD1*	delR608, fs330, p.L302P, p.R496L	Ashkenazi Jewish	1 in 90	97%	1 in 2968	
			Caucasian/White	1 in 250	20%	1 in 312	
			Other or Mixed Ethnicity	1 in 500	20%	1 in 625	
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	
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	N/A	N/A	N/A
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	
Usher syndrome, type IF	*PCDH15*	p.R245X	Ashkenazi Jewish	1 in 147	75%	1 in 585	
			Other or Mixed Ethnicity	1 in 428	10%	1 in 475	
Usher syndrome, type III	*CLRN1*	528T>G, p.N48K	Ashkenazi Jewish	1 in 120	98%	1 in 5951	
			Other or Mixed Ethnicity	1 in 449	10%	1 in 499	
Walker-Warburg syndrome	*FKTN*	p.F390X	Ashkenazi Jewish	1 in 150	95%	1 in 2981	
			Other or Mixed Ethnicity	1 in 500	10%	1 in 555	

*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.

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