progenity®

Preparent® Trio Panel

Disorder	Gene	Variants	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
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, 458delAT, 460delG, 489delC, 519delT, 525delT, 541del4, 541delC, 546insCTA, 547insGA, 547insTA, 556delA, 557delT, 565delC, 574delA, 5T, 605insT, 621+1G>T, 657delA, 663delT, 675del4, 681delC,	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
		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, R117C, R117H,				

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		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					
Fragile X syndrome (X-linked)*	*FMR1*	CGG repeat analysis	Other or Mixed Ethnicity	1 in 259	99%	1 in 25801	
Spinal muscular atrophy	*SMN1* *SMN2*	Copy number analysis				Two Copies *SMN1*	Three 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

*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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Test References

1. American College of Obstetricians and Gynecologists (ACOG) Committee Opinion Number 442. Preconception and Prenatal Carrier Screening for Genetic Diseases in Individuals of Eastern European Jewish Descent. Obstet Gyn. 2009 Oct;114(4):950-3.
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3. American College of Obstetrics and Gynecology Committee Opinion Number 486. Update on Carrier Screening for Cystic Fibrosis. Obstet Gyn. 2011 Apr;117(4):1028-31.
4. Hendrickson BC, Donohoe C, Akmaev VR, et al. Differences in SMN1 allele frequencies among ethnic groups within North America. J Med Genet. 2009 Sept;46(9):641-4. [PMID 19625283].