progenity®

# Preparent® CF (149 and 600 mutations)

| Disorder | Gene | Variants | Ethnicity | Carrier Frequency | Detection Rate | Residual Risk |
|---|---|---|---|---|---|---|
| Cystic fibrosis (149 and 600 mutations*) | *CFTR* | See Variants Tested on page 2 | Ashkenazi Jewish | 1 in 24 | 99% | 1 in 2301 |
| | | | Asian | 1 in 94 | 56% | 1 in 212 |
| | | | African American/Black | 1 in 61 | 82% | 1 in 334 |
| | | | Caucasian/White | 1 in 25 | 95% | 1 in 481 |
| | | | Hispanic | 1 in 58 | 84% | 1 in 357 |
| | | | Other or Mixed Ethnicity | 1 in 30 | N/A | N/A |

*The 600-mutation assay includes rare but clinically relevant mutations. Detection rates and residual risk calculations for this assay are consistent with the assay comprised of 149 mutations.

# Preparent® CF (149 and 600 mutations)

## Variants Tested

**Cystic Fibrosis (149 mutations):** 1078delT, 1154insTC, 1213delT, 1248+1G>A, 1259insA, 1341+1G>A, 1461ins4, 1525-1G>A, 1548delG, 1677delTA, 1717-1G>A, 1717-8G>A, 1811+1.6kbA>G, 1812-1G>A, 1898+1G>A, 1898+1G>C, 1898+3A>G, 1898+5G>A, 1949del84, 2143delT, 2183delAA>G, 2184delA, 2184insA, 2307insA, 2347delG, 2585delT, 2622+1G>A, 2711delT, 2789+5G>A, 3007delG, 3120+1G>A, 3120G>A, 3121-1G>A, 3171delC, 3199del6, 3272-26A>G, 3659delC, 3791delC, 3821delT, 3849+10kbC>T, 3876delA, 3905insT, 394delTT, 4005+1G>A, 4016insT, 405+1G>A, 406-1G>A, 4209TGTT>AA, 4382delA, 444delA, 457TAT>G, 574delA, 5T, 621+1G>T, 663delT, 711+1G>T, 711+3A>G, 711+5G>A, 712-1G>T, 852del22, 935delA, 9T, [delta]F311, [delta]F508, [delta]I507, A455E, A559T, CFTRdele2.3, CFTRdele22.23, D110H, D1152H, E1104X, E585X, E60X, E822X, E831X, E92K, E92X, F508C, G1244E, G178R, G330X, G542X, G551D, G85E, G970R, H199Y, I336K, K710X, L1065P, L1077P, L206W, L467P, L732X, L927P, M1101K, M1V, N1303K, P205S, P67L, Q1313X, Q179K, Q220X, Q39X, Q493X, Q552X, Q890X, Q98X, R1066C, R1066H, R1158X, R1162X, R117C, R117H, R334W, R347H, R347P, R352Q, R553X, R560K, R560T, R709X, R75X, R764X, R851X, S1196X, S1251N, S1255X (TAA), S1255X (TGA), S341P, S466X (TAA), S466X (TGA), S489X, S492F, S549N, S549R (AGG), S549R (CGT), S945L, T338I, V520F, W1089X, W1204X (TAG), W1204X (TGA), W1282X, W401X (TAG), W401X (TGA), W846X, Y1092X (TAA), Y1092X (TAG), Y122X, Y569D

**Cystic Fibrosis (600 mutations):** 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, 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, 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

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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.
2. American College of Medical Genetics and Genomics (ACMG) Practice Guidelines. Carrier screening in individuals of Ashkenazi Jewish descent. Genet Med. 2008;10(1):54-6.
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.