Variant List – iPLEX® Pro CFTR Panel

Variant Name	Nucleotide Change	Amino Acid Change	dbSNP RS#
CFTRdele2,3	c.54- 5940_273+10250del21kb	p.Ser18ArgfsX16	–
E60X	c.178G>T	p.Glu60Ter	rs77284892
R75X	c.223C>T	p.Arg75Ter	rs121908749
G85E	c.254G>A	p.Gly85Glu	rs75961395
394delTT	c.262_263delTT	p.Leu88Ilefs	rs121908769
406-1G->A	c.274-1G>A	NA	rs121908792
R117C	c.349C>A	p.Arg117Cys	rs77834169
R117G	c.349C>G	p.Arg117Gly	rs77834169
R117H	c.350G>A	p.Arg117His	rs78655421
Y122X	c.366T>A	p.Tyr122Ter	rs79660178
621+1G>T	c.489+1G>T	NA	rs78756941
G178R	c.532G>A	p.Gly178Arg	rs80282562
711+1G->T	c.579+1G>T	NA	rs77188391
L206W	c.617T>G	p.Leu206Trp	rs121908752
935delA	c.803delA	p.Asn268Ilefs	rs121908772
1078delT	c.948delT	p.Phe316Leufs	rs121908744
G330X	c.988G>T	p.Gly330Ter	rs79031340
R334W	c.1000C>T	p.Arg334Trp	rs121909011
R347H	c.1040G>A	p.Arg347His	rs77932196
R347P	c.1040G>T	p.Arg347Pro	rs77932196
T5/T7/T9	c.1210-7T[5][7][9]	NA	rs1805177
A455E	c.1364C>A	p.Ala455Glu	rs74551128
A455V	c.1364C>T	p.Ala455Val	rs74551128
Q493X	c.1477C>T	p.Gln493Ter	rs77101217
I506V	c.1516A>G	p.Ile506Val	rs1800091
I507del	c.1519_1521delATC	p.Ile507del	rs121908745
I507V	c.1519A>G	p.Ile507Val	rs1801178
F508del	c.1521_1523delCTT	p.Phe508del	rs113993960
F508C	c.1523T>G	p.Phe508Cys	rs74571530
1677delTA	c.1545_1546delTA	p.Tyr515Terfs	rs121908776
V520F	c.1558G>T	p.Val520Phe	rs77646904
1717-1G>A	c.1585-1G>A	NA	rs76713772
G542X	c.1624G>T	p.Gly542Ter	rs113993959
S549R_1645A->C	c.1645A>C	p.Ser549Arg	rs121908757
S549N	c.1646G>A	p.Ser549Asn	rs121908755
S549R_1647T->G	c.1647T>G	p.Ser549Arg	rs121909005
G551S	c.1651G>A	p.Gly551Ser	rs121909013
G551D	c.1652G>A	p.Gly551Asp	rs75527207
R553X	c.1657C>T	p.Arg553Ter	rs74597325

Society-recommended variants

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Variant List – iPLEX® Pro CFTR Panel (PAGE 2 OF 2)

Variant Name	Nucleotide Change	Amino Acid Change	dbSNP RS#
A559T	c.1675G>A	p.Ala559Thr	rs75549581
R560K	c.1679G>A	p.Arg560Lys	rs80055610
R560T	c.1679G>C	p.Arg560Thr	rs80055610
1898+1G>A	c.1766+1G>A	NA	rs121908748
1898+5G->T	c.1766+5G>T	NA	rs121908796
2055del9->A	c.1923_1931delCTCAAAACTinsA	p.Ser641Argfs	rs121908779
2143delT	c.2012delT	p.Leu671Terfs	rs121908812
2183AA->G	c.2051_2052delAAinsG	p.Lys684Serfs	rs121908799
2184delA	c.2052delA	p.Lys684Asnfs	rs121908746
K710X	c.2128A>T	p.Lys710Ter	rs75115087
2307insA	c.2175dupA	p.Glu726Argfs	rs121908787
2789+5G>A	c.2657+5G>A	NA	rs80224560
Q890X	c.2668C>T	p.Gln890Ter	rs79633941
3120+1G>A	c.2988+1G>A	NA	rs75096551
3199del6	c.3067_3072delATAGTG	p.Ile1023_Val1024del	rs121908767
R1066C	c.3196C>T	p.Arg1066Cys	rs78194216
W1089X	c.3266G>A	p.Trp1089Ter	rs78802634
Y1092X	c.3276C>G	p.Tyr1092Ter	rs121908761
M1101K	c.3302T>A	p.Met1101Lys	rs36210737
D1152H	c.3454G>C	p.Asp1152His	rs75541969
R1158X	c.3472C>T	p.Arg1158Ter	rs79850223
R1162X	c.3484C>T	p.Arg1162Ter	rs74767530
R1162Q	c.3485G>A	p.Arg1162Gln	rs1800120
R1162L	c.3485G>T	p.Arg1162Leu	rs1800120
3659delC	c.3528delC	p.Lys1177Serfs	rs121908747
S1196X	c.3587C>G	p.Ser1196Ter	rs121908763
3791delC	c.3659delC	p.Thr1220Lysfs	rs121908811
3849+10kbC>T	c.3718-2477C>T	NA	rs75039782
G1244E	c.3731G>A	p.Gly1244Glu	rs267606723
3876delA	c.3744delA	p.Lys1250Argfs	rs121908784
S1251N	c.3752G>A	p.Ser1251Asn	rs74503330
S1255L	c.3764C>T	p.Ser1255Leu	rs76649725
S1255P	c.3763T>C	p.Ser1255Pro	rs121909041
S1255X	c.3764C>A	p.Ser1255Ter	rs76649725
3905insT	c.3773dupT	p.Leu1258Phefs	rs121908789
W1282X	c.3846G>A	p.Trp1282Ter	rs77010898
N1303K	c.3909C>G	p.Asn1303Lys	rs80034486
G1349D	c.4046G>A	p.Gly1349Asp	rs193922525

Society-recommended variants

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