Cystic fibrosis targeted expanded panel

Division of Genome Diagnostics
at BC Children's and BC Women's Hospitals

CFTR expanded variant list: EXPv1.0

Legacy Name	cDNA	Protein	Variant Determination (CFTR2 Dec 2017)
124del23bp	c.-9_14del23	-	CF-causing
M1V	c.1A>G	p.Met1Val	CF-causing
Q2X	c.4C>T	p.Gln2Ter	CF-causing
S4X	c.11C>A	p.Ser4Ter	CF-causing
P5L	c.14C>T	p.Pro5Leu	Varying clinical consequence
182delT	c.50delT	p.Phe17SerfsTer8	CF-causing
185+1G>T	c.53+1G>T	-	CF-causing
W19X	c.57G>A	p.Trp19Ter	CF-causing
G27X	c.79G>T	p.Gly27Ter	CF-causing
Q30X	c.88C>T	p.Gln30Ter	CF-causing
Q39X	c.115C>T	p.Gln39Ter	CF-causing
A46D	c.137C>A	p.Ala46Asp	CF-causing
296+1G>A	c.164+1G>A	-	CF-causing
296+1G>T	c.164+1G>T	-	CF-causing
296+2T>C	c.164+2T>C	-	CF-causing
296+3insT	c.164+4dupT	-	CF-causing
297-3C>T	c.165-3C>T	-	CF-causing
297-1G>A	c.165-1G>A	-	CF-causing
E56K	c.166G>A	p.Glu56Lys	CF-causing
W57G	c.169T>G	p.Trp57Gly	CF-causing
W57X	c.170G>A	p.Trp57Ter	CF-causing
W57X	c.171G>A	p.Trp57Ter	CF-causing
306insA	c.175dupA	p.Arg59LysfsTer10	CF-causing
306delTAGA	c.174_177delTAGA	p.Asp58GlufsTer32	CF-causing
E60X	c.178G>T	p.Glu60Ter	CF-causing
P67L	c.200C>T	p.Pro67Leu	CF-causing
R74W	c.220C>T	p.Arg74Trp	Varying clinical consequence
R75X	c.223C>T	p.Arg75Ter	CF-causing
365-366insT	c.233dupT	p.Trp79LeufsTer32	CF-causing
G85E	c.254G>A	p.Gly85Glu	CF-causing
394delTT	c.262_263delTT	p.Leu88IlefsTer22	CF-causing
L88X	c.263T>A	p.Leu88Ter	CF-causing
L88X	c.263T>G	p.Leu88Ter	CF-causing
405+1G>A	c.273+1G>A	-	CF-causing
405+3A>C	c.273+3A>C	-	CF-causing

Legacy Name	cDNA	Protein	Variant Determination (CFTR2 Dec 2017)
406-2A>G	c.274-2A>G	-	CF-causing
406-1G>A	c.274-1G>A	-	CF-causing
E92K	c.274G>A	p.Glu92Lys	CF-causing
E92X	c.274G>T	p.Glu92Ter	CF-causing
Q98X	c.292C>T	p.Gln98Ter	CF-causing
Q98R	c.293A>G	p.Gln98Arg	CF-causing
P99L	c.296C>T	p.Pro99Leu	CF-causing
442delA	c.310delA	p.Arg104GlufsTer3	CF-causing
444delA	c.313delA	p.Ile105SerfsTer2	CF-causing
457TAT>G	c.325_327delinsG	p.Tyr109GlyfsTer4	CF-causing
D110H	c.328G>C	p.Asp110His	CF-causing
D110E	c.330C>A	p.Asp110Glu	Varying clinical consequence
R117C	c.349C>T	p.Arg117Cys	CF-causing
R117G	c.349C>G	p.Arg117Gly	Varying clinical consequence
R117H	c.350G>A	p.Arg117His	Varying clinical consequence
Y122X	c.366T>A	p.Tyr122Ter	CF-causing
541delC	c.409delC	p.Leu137SerfsTer16	CF-causing
574delA	c.442delA	p.Ile148LeufsTer5	CF-causing
602del14	c.470_483del14	p.Phe157Ter	CF-causing
621+1G>T	c.489+1G>T	-	CF-causing
621+3A>G	c.489+3A>G	-	Varying clinical consequence
L165S	c.494T>C	p.Leu165Ser	CF-causing
663delT	c.531delT	p.Ile177MetfsTer12	CF-causing
G178R	c.532G>A	p.Gly178Arg	CF-causing
675del4	c.543_546delTAGT	p.Leu183PhefsTer5	CF-causing
E193X	c.577G>T	p.Glu193Ter	CF-causing
711+1G>T	c.579+1G>T	-	CF-causing
711+3A>G	c.579+3A>G	-	CF-causing
711+5G>A	c.579+5G>A	-	CF-causing
712-1G>T	c.580-1G>T	-	CF-causing
H199Y	c.595C>T	p.His199Tyr	CF-causing
P205S	c.613C>T	p.Pro205Ser	CF-causing
L206W	c.617T>G	p.Leu206Trp	CF-causing
W216X	c.647G>A	p.Trp216Ter	CF-causing
Q220X	c.658C>T	p.Gln220Ter	CF-causing
L227R	c.680T>G	p.Leu227Arg	CF-causing
849delG	c.717delG	p.Leu240Ter	CF-causing
852del22	c.723_743+1del22	p.Gly241GlufsTer13	CF-causing
M265R	c.794T>G	p.Met265Arg	Varying clinical consequence
935delA	c.803delA	p.Asn268IlefsTer17	CF-causing
Y275X	c.825C>G	p.Tyr275Ter	CF-causing

Legacy Name	cDNA	Protein	Variant Determination (CFTR2 Dec 2017)
C276X	c.828C>A	p.Cys276Ter	CF-causing
977insA	c.850dupA	p.Met284AsnfsTer3	CF-causing
991del5	c.861_865delCTTAA	p.Asn287LysfsTer19	CF-causing
F311L	c.933C>G	p.Phe311Leu	CF-causing
1078delT	c.948delT	p.Phe316LeufsTer12	CF-causing
1119delA	c.987delA	p.Gly330GlufsTer39	CF-causing
G330X	c.988G>T	p.Gly330Ter	CF-causing
R334W	c.1000C>T	p.Arg334Trp	CF-causing
R334Q	c.1001G>A	p.Arg334Gln	Varying clinical consequence
R334L	c.1001G>T	p.Arg334Leu	CF-causing
1138insG	c.1006_1007insG	p.Ile336SerfsTer28	CF-causing
I336K	c.1007T>A	p.Ile336Lys	CF-causing
T338I	c.1013C>T	p.Thr338Ile	CF-causing
S341P	c.1021T>C	p.Ser341Pro	CF-causing
1154insTC	c.1021_1022dupTC	p.Phe342HisfsTer28	CF-causing
1161delC	c.1029delC	p.Cys343Ter	CF-causing
R347H	c.1040G>A	p.Arg347His	CF-causing
R347P	c.1040G>C	p.Arg347Pro	CF-causing
R352Q	c.1055G>A	p.Arg352Gln	CF-causing
1213delT	c.1081delT	p.Trp361GlyfsTer8	CF-causing
1248+1G>A	c.1116+1G>A	-	CF-causing
1249-1G>A	c.1117-1G>A	-	CF-causing
1259insA	c.1130dupA	p.Gln378AlafsTer4	CF-causing
1288insTA	c.1155_1156dupTA	p.Asn386IlefsTer3	CF-causing
W401X	c.1202G>A	p.Trp401Ter	CF-causing
W401X	c.1203G>A	p.Trp401Ter	CF-causing
1341+1G>A	c.1209+1G>A	-	CF-causing
1343delG	c.1211delG	p.Gly404AspfsTer38	CF-causing
Q414X	c.1240C>T	p.Gln414Ter	CF-causing
1429del7	c.1301_1307delCACTTCT	p.Ser434LeufsTer6	CF-causing
D443Y	c.1327G>T	p.Asp443Tyr	Varying clinical consequence
1461ins4	c.1327_1330dupGATA	p.Ile444ArgfsTer3	CF-causing
1471delA	c.1340delA	p.Lys447ArgfsTer2	CF-causing
A455E	c.1364C>A	p.Ala455Glu	CF-causing
1497delGG	c.1365_1366delGG	p.Val456CysfsTer25	CF-causing
V456A	c.1367T>C	p.Val456Ala	CF-causing
1504delG	c.1373delG	p.Gly458AspfsTer11	CF-causing
1525-1G>A	c.1393-1G>A	-	CF-causing
1525-2A>G	c.1393-2A>G	-	CF-causing
S466X	c.1397C>A	p.Ser466Ter	CF-causing
S466X	c.1397C>G	p.Ser466Ter	CF-causing

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L467P	c.1400T>C	p.Leu467Pro	CF-causing
1548delG	c.1418delG	p.Gly473GlufsTer54	CF-causing
S489X	c.1466C>A	p.Ser489Ter	CF-causing
S492F	c.1475C>T	p.Ser492Phe	CF-causing
1609delCA	c.1477_1478delCA	p.Gln493ValfsTer10	CF-causing
Q493X	c.1477C>T	p.Gln493Ter	CF-causing
W496X	c.1487G>A	p.Trp496Ter	CF-causing
I506V	c.1516A>G	p.Ile506Val	Non CF-causing
I507del	c.1519_1521delATC	p.Ile507del	CF-causing
I507V	c.1519A>G	p.Ile507Val	Non CF-causing
F508del	c.1521_1523delCTT	p.Phe508del	CF-causing
F508C	c.1523T>G	p.Phe508Cys	Non CF-causing
D513G	c.1538A>G	p.Asp513Gly	CF-causing
1677delTA	c.1545_1546delTA	p.Tyr515Ter	CF-causing
V520F	c.1558G>T	p.Val520Phe	CF-causing
C524X	c.1572C>A	p.Cys524Ter	CF-causing
Q525X	c.1573C>T	p.Gln525Ter	CF-causing
1716+1G>A	c.1584+1G>A	-	CF-causing
1717-1G>A	c.1585-1G>A	-	CF-causing
1717-8G>A	c.1585-8G>A	-	CF-causing
G542X	c.1624G>T	p.Gly542Ter	CF-causing
S549R	c.1645A>C	p.Ser549Arg	CF-causing
S549N	c.1646G>A	p.Ser549Asn	CF-causing
S549R	c.1647T>G	p.Ser549Arg	CF-causing
G550X	c.1648G>T	p.Gly550Ter	CF-causing
1782delA	c.1650delA	p.Gly551ValfsTer8	CF-causing
G551S	c.1651G>A	p.Gly551Ser	CF-causing
G551D	c.1652G>A	p.Gly551Asp	CF-causing
Q552X	c.1654C>T	p.Gln552Ter	CF-causing
R553X	c.1657C>T	p.Arg553Ter	CF-causing
1802delC	c.1670delC	p.Ser557PhefsTer2	CF-causing
A559T	c.1675G>A	p.Ala559Thr	CF-causing
1811+1.6kbA>G	c.1680-886A>G	-	CF-causing
1811+1G>C	c.1679+1G>C	-	CF-causing
R560K	c.1679G>A	p.Arg560Lys	CF-causing
R560T	c.1679G>C	p.Arg560Thr	CF-causing
1811+1G>A	c.1679+1G>A	-	CF-causing
1811+1643G>T	c.1680-877G>T	-	CF-causing
1812-1G>A	c.1680-1G>A	-	CF-causing
R560S	c.1680A>C	p.Arg560Ser	CF-causing
A561E	c.1682C>A	p.Ala561Glu	CF-causing

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Y563N	c.1687T>A	p.Tyr563Asn	CF-causing
Y563D	c.1687T>G	p.Tyr563Asp	CF-causing
1824delA	c.1692delA	p.Asp565MetfsTer7	CF-causing
1833delT	c.1703delT	p.Leu568CysfsTer4	CF-causing
Y569D	c.1705T>G	p.Tyr569Asp	CF-causing
P574H	c.1721C>A	p.Pro574His	CF-causing
F575Y	c.1724T>A	p.Phe575Tyr	Varying clinical consequence
D579G	c.1736A>G	p.Asp579Gly	Varying clinical consequence
E585X	c.1753G>T	p.Glu585Ter	CF-causing
E588V	c.1763A>T	p.Glu588Val	Varying clinical consequence
1898+1G>A	c.1766+1G>A	-	CF-causing
1898+1G>C	c.1766+1G>C	-	CF-causing
1898+1G>T	c.1766+1G>T	-	CF-causing
1898+3A>G	c.1766+3A>G	-	CF-causing
1898+5G>T	c.1766+5G>T	-	CF-causing
1924del7	c.1792_1798delAAAACTA	p.Lys598GlyfsTer11	CF-causing
H609R	c.1826A>G	p.His609Arg	CF-causing
D614G	c.1841A>G	p.Asp614Gly	Varying clinical consequence
G622D	c.1865G>A	p.Gly622Asp	Varying clinical consequence
2055del9>A	c.1923_1931delinsA	p.Ser641ArgfsTer5	CF-causing
2075delA	c.1943delA	p.Asp648ValfsTer15	CF-causing
2105-2117del13insAGAAA	c.1973_1985delinsAGAAA	p.Arg658LysfsTer4	CF-causing
2118del4	c.1986_1989delAACT	p.Thr663ArgfsTer8	CF-causing
2143delT	c.2012delT	p.Leu671Ter	CF-causing
G673X	c.2017G>T	p.Gly673Ter	CF-causing
2183AA>G	c.2051_2052delinsG	p.Lys684SerfsTer38	CF-causing
2184insA	c.2052dupA	p.Gln685ThrfsTer4	CF-causing
2184delA	c.2052delA	p.Lys684AsnfsTer38	CF-causing
2185insC	c.2053dupC	p.Gln685ProfsTer4	CF-causing
Q685X	c.2053C>T	p.Gln685Ter	CF-causing
R709X	c.2125C>T	p.Arg709Ter	CF-causing
K710X	c.2128A>T	p.Lys710Ter	CF-causing
Q715X	c.2143C>T	p.Gln715Ter	CF-causing
Q720X	c.2158C>T	p.Gln720Ter	CF-causing
2307insA	c.2175dupA	p.Glu726ArgfsTer4	CF-causing
L732X	c.2195T>G	p.Leu732Ter	CF-causing
2347delG	c.2215delG	p.Val739TyrfsTer16	CF-causing
2372del8	c.2241_2248delGATACTGC	p.Ile748SerfsTer28	CF-causing
P750L	c.2249C>T	p.Pro750Leu	Varying clinical consequence
R764X	c.2290C>T	p.Arg764Ter	CF-causing
R785X	c.2353C>T	p.Arg785Ter	CF-causing

Legacy Name	cDNA	Protein	Variant Determination (CFTR2 Dec 2017)
R792X	c.2374C>T	p.Arg792Ter	CF-causing
2556insAT	c.2423_2424dupAT	p.Ser809IlefsTer13	CF-causing
2585delT	c.2453delT	p.Leu818TrpfsTer3	CF-causing
2594delGT	c.2463_2464delTG	p.Ser821ArgfsTer4	CF-causing
E822X	c.2464G>T	p.Glu822Ter	CF-causing
2622+1G>A	c.2490+1G>A	-	CF-causing
E831X	c.2491G>T	p.Glu831Ter	CF-causing
W846X	c.2537G>A	p.Trp846Ter	CF-causing
W846X	c.2538G>A	p.Trp846Ter	CF-causing
Y849X	c.2547C>A	p.Tyr849Ter	CF-causing
R851X	c.2551C>T	p.Arg851Ter	CF-causing
2711delT	c.2583delT	p.Phe861LeufsTer3	CF-causing
2721del11	c.2589_2599delAATTTGGTGCT	p.Ile864SerfsTer28	CF-causing
2732insA	c.2601dupA	p.Val868SerfsTer28	CF-causing
2752-26A>G	c.2620-26A>G	-	Varying clinical consequence
W882X	c.2645G>A	p.Trp882Ter	CF-causing
2789+5G>A	c.2657+5G>A	-	CF-causing
2790-1G>C	c.2658-1G>C	-	CF-causing
Q890X	c.2668C>T	p.Gln890Ter	CF-causing
S912X	c.2735C>A	p.Ser912Ter	CF-causing
2869insG	c.2737_2738insG	p.Tyr913Ter	CF-causing
Y913X	c.2739T>A	p.Tyr913Ter	CF-causing
2896insAG	c.2763_2764dupAG	p.Val922GlufsTer2	CF-causing
L927P	c.2780T>C	p.Leu927Pro	CF-causing
2942insT	c.2810dupT	p.Val938GlyfsTer37	CF-causing
2957delT	c.2825delT	p.Ile942ThrfsTer26	CF-causing
S945L	c.2834C>T	p.Ser945Leu	CF-causing
2991del32	c.2859_2890del32	p.Leu953PhefsTer11	CF-causing
3007delG	c.2875delG	p.Ala959HisfsTer9	CF-causing
3028delA	c.2896delA	p.Thr966ArgfsTer2	CF-causing
L967S	c.2900T>C	p.Leu967Ser	Varying clinical consequence
G970R	c.2908G>C	p.Gly970Arg	CF-causing
S977F	c.2930C>T	p.Ser977Phe	Varying clinical consequence
D979V	c.2936A>T	p.Asp979Val	CF-causing
3120G>A	c.2988G>A	-	CF-causing
3120+1G>A	c.2988+1G>A	-	CF-causing
3121-1G>A	c.2989-1G>A	-	CF-causing
3121-2A>G	c.2989-2A>G	-	CF-causing
3132delTG	c.3002_3003delTG	p.Val1001AspfsTer45	CF-causing
3171delC	c.3039delC	p.Tyr1014ThrfsTer9	CF-causing
3171insC	c.3039dupC	p.Tyr1014LeufsTer33	CF-causing

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3199del6[1]	c.3067_3072delATAGTG	p.Ile1023_Val1024del	-
Y1032C	c.3095A>G	p.Tyr1032Cys	Varying clinical consequence
Q1042X	c.3124C>T	p.Gln1042Ter	CF-causing
3271delGG	c.3139_3139+1delGG	p.Gly1047GlnfsTer28	CF-causing
3272-26A>G	c.3140-26A>G	-	CF-causing
F1052V	c.3154T>G	p.Phe1052Val	Varying clinical consequence
T1053I	c.3158C>T	p.Thr1053Ile	Varying clinical consequence
H1054D	c.3160C>G	p.His1054Asp	CF-causing
G1061R	c.3181G>C	p.Gly1061Arg	CF-causing
L1065P	c.3194T>C	p.Leu1065Pro	CF-causing
R1066C	c.3196C>T	p.Arg1066Cys	CF-causing
R1066H	c.3197G>A	p.Arg1066His	CF-causing
G1069R	c.3205G>A	p.Gly1069Arg	Varying clinical consequence
R1070W	c.3208C>T	p.Arg1070Trp	Varying clinical consequence
R1070Q	c.3209G>A	p.Arg1070Gln	Varying clinical consequence
3349insT	c.3217dupT	p.Tyr1073LeufsTer3	CF-causing
F1074L	c.3222T>A	p.Phe1074Leu	Varying clinical consequence
L1077P	c.3230T>C	p.Leu1077Pro	CF-causing
W1089X	c.3266G>A	p.Trp1089Ter	CF-causing
Y1092X	c.3276C>A	p.Tyr1092Ter	CF-causing
Y1092X	c.3276C>G	p.Tyr1092Ter	CF-causing
W1098X	c.3293G>A	p.Trp1098Ter	CF-causing
W1098X	c.3294G>A	p.Trp1098Ter	CF-causing
F1099L	c.3297C>A	p.Phe1099Leu	Varying clinical consequence
M1101K	c.3302T>A	p.Met1101Lys	CF-causing
R1102X	c.3304A>T	p.Arg1102Ter	CF-causing
E1104X	c.3310G>T	p.Glu1104Ter	CF-causing
S1118F	c.3353C>T	p.Ser1118Phe	CF-causing
3500-2A>G	c.3368-2A>G	-	CF-causing
W1145X	c.3435G>A	p.Trp1145Ter	CF-causing
D1152H	c.3454G>C	p.Asp1152His	Varying clinical consequence
V1153E	c.3458T>A	p.Val1153Glu	Varying clinical consequence
3600G>A	c.3468G>A	-	CF-causing
3600+2insT	c.3468+2dupT	-	CF-causing
3600+5G>A	c.3468+5G>A	-	CF-causing
R1158X	c.3472C>T	p.Arg1158Ter	CF-causing
S1159F	c.3476C>T	p.Ser1159Phe	CF-causing
R1162X	c.3484C>T	p.Arg1162Ter	CF-causing

[1] This variant is not present in the CFTR2 Dec 2017 release. It is the presumed pathogenic variant in linkage with I148T.

Legacy Name	cDNA	Protein	Variant Determination (CFTR2 Dec 2017)
3659delC	c.3528delC	p.Lys1177SerfsTer15	CF-causing
3667ins4	c.3532_3535dupTCAA	p.Thr1179IlefsTer17	CF-causing
S1196X	c.3587C>G	p.Ser1196Ter	CF-causing
3737delA	c.3605delA	p.Asp1202AlafsTer9	CF-causing
W1204X	c.3611G>A	p.Trp1204Ter	CF-causing
W1204X	c.3612G>A	p.Trp1204Ter	CF-causing
3791delC	c.3659delC	p.Thr1220LysfsTer8	CF-causing
3821delT	c.3691delT	p.Ser1231ProfsTer4	CF-causing
I1234V	c.3700A>G	p.Ile1234Val	CF-causing
3849G>A	c.3717G>A	-	CF-causing
3849+4A>G	c.3717+4A>G	-	CF-causing
3849+5G>A	c.3717+5G>A	-	CF-causing
3849+40A>G	c.3717+40A>G	-	CF-causing
3849+10kbC>T	c.3718-2477C>T	-	CF-causing
3850-1G>A	c.3718-1G>A	-	CF-causing
3850-3T>G	c.3718-3T>G	-	CF-causing
G1244E	c.3731G>A	p.Gly1244Glu	CF-causing
T1246I	c.3737C>T	p.Thr1246Ile	Varying clinical consequence
3876delA	c.3744delA	p.Lys1250ArgfsTer9	CF-causing
3878delG	c.3747delG	p.Lys1250ArgfsTer9	CF-causing
S1251N	c.3752G>A	p.Ser1251Asn	CF-causing
L1254X	c.3761T>G	p.Leu1254Ter	CF-causing
S1255P	c.3763T>C	p.Ser1255Pro	CF-causing
S1255X	c.3764C>A	p.Ser1255Ter	CF-causing
3905insT	c.3773dupT	p.Leu1258PhefsTer7	CF-causing
D1270N	c.3808G>A	p.Asp1270Asn	Varying clinical consequence
W1282X	c.3846G>A	p.Trp1282Ter	CF-causing
R1283M	c.3848G>T	p.Arg1283Met	CF-causing
Q1291R	c.3872A>G	p.Gln1291Arg	Varying clinical consequence
4005+1G>A	c.3873+1G>A	-	CF-causing
4005+2T>C	c.3873+2T>C	-	CF-causing
4010del4	c.3883_3886delATTT	p.Ile1295PhefsTer32	CF-causing
4015delA	c.3883delA	p.Ile1295PhefsTer33	CF-causing
4016insT (4021dupT)[2]	c.3889dupT	p.Ser1297PhefsTer5	CF-causing
4022insT	c.3891dupT	p.Gly1298TrpfsTer4	CF-causing
4040delA	c.3908delA	p.Asn1303ThrfsTer25	CF-causing
N1303K	c.3909C>G	p.Asn1303Lys	CF-causing

[2] 4021dupT was incorrectly identified in CFTR2 as c.3899dupT, correspondence with CFTR2 has confirmed the cDNA name for this variant is c.3889dupT and it is therefore identical to 4016insT.

Legacy Name	cDNA	Protein	Variant Determination (CFTR2 Dec 2017)
Q1313X	c.3937C>T	p.Gln1313Ter	CF-causing
Q1330X	c.3988C>T	p.Gln1330Ter	CF-causing
L1335P	c.4004T>C	p.Leu1335Pro	CF-causing
4168delCTAAGCC	c.4036_4042delCTAAGCC	p.Leu1346MetfsTer6	CF-causing
G1349D	c.4046G>A	p.Gly1349Asp	CF-causing
4209TGTT>AA	c.4077_4080delinsAA	p.Val1360Thrfs*3	CF-causing
4218insT	c.4086dupT	p.Lys1363Ter	CF-causing
E1371X	c.4111G>T	p.Glu1371Ter	CF-causing
4259del5	c.4127_4131delTGGAT	p.Leu1376SerfsTer8	CF-causing
Q1382X	c.4144C>T	p.Gln1382Ter	CF-causing
4279insA	c.4147dupA	p.Ile1383AsnfsTer3	CF-causing
4326delTC	c.4197_4198delCT	p.Cys1400Ter	CF-causing
Q1411X	c.4231C>T	p.Gln1411Ter	CF-causing
Q1412X	c.4234C>T	p.Gln1412Ter	CF-causing
4374+1G>T	c.4242+1G>T	-	CF-causing
4374+1G>A	c.4242+1G>A	-	CF-causing
4382delA	c.4251delA	p.Glu1418ArgfsTer14	CF-causing
4428insGA	c.4300_4301dupAG	p.Ser1435GlyfsTer14	CF-causing

Reference sequence: NM_000492.3; HGVS nomenclature version 19.01

Assay QC metrics:

Analytical sensitivity for single nucleotide variants (95% CI): 100% (93.2-100%)

Analytical sensitivity for insertions/deletions (95% CI): 100% (95.3-100%)

Analytical specificity (95% CI): 100% (99.9-100%)

Minimum read depth: >100