Cystic fibrosis targeted expanded panel (EXP) v5.0

Division of Genome Diagnostics

at BC Children's and BC Women's Hospitals

Minor update: three (3) variants changed status in CFTR2 25Sept2024, denoted by $

This expanded panel includes all detectable CF-causing or varying clinical consequence CFTR variants classified by CFTR2 (September 24, 2021 release; www.cftr2.org) or by the Division of Genome Diagnostics.

- Classification is as per CFTR2 unless otherwise denoted by GD.
- cDNA and Protein are described using HGVS nomenclature version 20.05.

- Change relative to EXPv4.0:
    - Eleven (19) additional variants, denoted by #

Legacy Name	cDNA	Predicted Protein	CFTR2 24Sept2021
124del23bp	c.-9_14del [1]	p.?	CF-causing
M1V	c.1A>G	p.? [2]	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
S13F	c.38C>T	p.Ser13Phe	CF-causing
L15P	c.44T>C	p.Leu15Pro	CF-causing
182delT	c.50delT	p.Phe17SerfsTer8	CF-causing
185+1G>T	c.53+1G>T	p.?	CF-causing
W19X	c.57G>A	p.Trp19Ter	CF-causing
G27R	c.79G>A	p.Gly27Arg	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	p.?	CF-causing
296+1G>T	c.164+1G>T	p.?	CF-causing
296+2T>C	c.164+2T>C	p.?	CF-causing
296+3insT	c.164+4dupT	p.?	CF-causing
297-1G>A	c.165-1G>A	p.?	CF-causing
E56K	c.166G>A	p.Glu56Lys	CF-causing
300delA	c.168delA #	p.Glu56AspfsTer35	CF-causing
W57G	c.169T>G	p.Trp57Gly	CF-causing
W57X	c.170G>A	p.Trp57Ter	CF-causing

[1] Full variant name c.-9_14delCGAGAGACCATGCAGAGGTCGCC

[2] Alternative historical name for this variant is p.Met1Val

Legacy Name	cDNA	Predicted Protein	CFTR2 24Sept2021
W57X	c.171G>A	p.Trp57Ter	CF-causing
306insA	c.175dupA	p.Arg59LysfsTer10	CF-causing
306delTAGA	c.174_177delTAGA	p.Asp58GlufsTer32	CF-causing
E60K	c.178G>A	p.Glu60Lys	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
G91R	c.271G>A	p.Gly91Arg	CF-causing
405+1G>A	c.273+1G>A	p.?	CF-causing
405+3A>C	c.273+3A>C	p.?	CF-causing
406-2A>G	c.274-2A>G	p.?	CF-causing
406-1G>A	c.274-1G>A	p.?	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
L102R	c.305T>G	p.Leu102Arg	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
E116K	c.346G>A #	p.Glu116Lys	CF-causing
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
R117P	c.350G>C	p.Arg117Pro	CF-causing
R117L	c.350G>T	p.Arg117Leu	Varying clinical consequence
A120T	c.358G>A	p.Ala120Thr	Varying clinical consequence
Y122X	c.366T>A	p.Tyr122Ter	CF-causing
G126D	c.377G>A	p.Gly126Asp	CF-causing
541delC	c.409delC	p.Leu137SerfsTer16	CF-causing
L138ins	c.413_415dupTAC	p.Leu138dup	CF-causing
H139R	c.416A>G	p.His139Arg	CF-causing
556delA	c.424delA #	p.Ile142PhefsTer11	CF-causing

Legacy Name	cDNA	Predicted Protein	CFTR2 24Sept2021
557delT	c.429delT	p.Phe143LeufsTer10	CF-causing
574delA	c.442delA	p.Ile148LeufsTer5	CF-causing
602del14	c.470_483del [3]	p.Phe157Ter	CF-causing
Y161D	c.481T>G	p.Tyr161Asp	CF-causing
621+1G>T	c.489+1G>T	p.?	CF-causing
621+3A>G	c.489+3A>G	p.?	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
F191V	c.571T>G	p.Phe191Val	CF-causing
D192G	c.575A>G	p.Asp192Gly	CF-causing
E193X	c.577G>T	p.Glu193Ter	CF-causing
E193K	c.577G>A	p.Glu193Lys	CF-causing
711+1G>T	c.579+1G>T	p.?	CF-causing
711+3A>G	c.579+3A>G	p.?	CF-causing
711+5G>A	c.579+5G>A	p.?	CF-causing
712-1G>T	c.580-1G>T	p.?	CF-causing
712-2A->G	c.580-2A>G #	p.?	CF-causing
G194R	c.580G>A	p.Gly194Arg	CF-causing
G194V	c.581G>T	p.Gly194Val	Varying clinical consequence
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
L218X	c.653T>A ^	p.Leu218Ter	CF-causing
Q220X	c.658C>T	p.Gln220Ter	CF-causing
L227R	c.680T>G	p.Leu227Arg	CF-causing
V232D	c.695T>A	p.Val232Asp	CF-causing
Q237E	c.709C>G	p.Gln237Glu	Varying clinical consequence
849delG	c.717delG	p.Leu240Ter	CF-causing
852del22	c.723_743+1del [4]	p.? [5]	CF-causing
876-2A->G	c.744-2A>G #	p.?	CF-causing
892delA	c.761delA #	p.Lys254ArgfsTer7	CF-causing
R258G	c.772A>G	p.Arg258Gly	Varying clinical consequence
M265R	c.794T>G	p.Met265Arg	Varying clinical consequence
935delA	c.803delA	p.Asn268IlefsTer17	CF-causing
Y275X	c.825C>G	p.Tyr275Ter	CF-causing

[3] Full variant name is c.470_483delTTAGTTTGATTTAT

[4] Full variant name is c.723_743+1delGAGAATGATGATGAAGTACAGG

[5] Alternative historical name for this variant is p.Gly241GlufsTer13

Legacy Name	cDNA	Predicted Protein	CFTR2 24Sept2021
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>A [#]	p.Phe311Leu	CF-causing
F311L	c.933C>G	p.Phe311Leu	CF-causing
F312del	c.935_937delTCT [#]	p.Phe312del	Varying clinical consequence
G314E	c.941G>A	p.Gly314Glu	Varying clinical consequence
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
L346P	c.1037T>C	p.Leu346Pro	CF-causing
R347H	c.1040G>A	p.Arg347His	CF-causing
R347P	c.1040G>C	p.Arg347Pro	CF-causing
R352W	c.1054C>T	p.Arg352Trp	Varying clinical consequence
R352Q	c.1055G>A	p.Arg352Gln	CF-causing
Q359K/T360K	c.[1075C>A;1079C>A] [6]	p.Gln359_Thr360delinsLysLys	CF-causing
Q359R	c.1076A>G	p.Gln359Arg	CF-causing [$]
1213delT	c.1081delT	p.Trp361GlyfsTer8	CF-causing
1248+1G>A	c.1116+1G>A	p.?	CF-causing
1249-1G>A	c.1117-1G>A	p.?	CF-causing
1259insA	c.1130dupA	p.Gln378AlafsTer4	CF-causing
E379X	c.1135G>T	p.Glu379Ter	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	p.?	CF-causing
1342-2A->C	c.1210-2A>C [#]	p.?	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

[6] Alternative historical names for this variant are c.[1075C>A;1079C>A] p.[Gln359Lys;Thr360Lys]. and c.1075_1079delinsAAAAA, p.Gln359_Thr360delinsLysLys

Legacy Name	cDNA	Predicted Protein	CFTR2 24Sept2021
S434X	c.1301C>A	p.Ser434Ter	CF-causing
S434X	c.1301C>G	p.Ser434Ter	CF-causing
D443Y	c.1327G>T	p.Asp443Tyr	Varying clinical consequence
1461ins4	c.1327_1330dupGATA	p.Ile444ArgfsTer3	CF-causing
1460delAT	c.1330_1331delAT #	p.Ile444Ter	CF-causing
1471delA	c.1340delA	p.Lys447ArgfsTer2	CF-causing
L453S	c.1358T>C	p.Leu453Ser	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	p.?	CF-causing
1525-2A>G	c.1393-2A>G	p.?	CF-causing
S466X	c.1397C>A	p.Ser466Ter	CF-causing
S466X	c.1397C>G	p.Ser466Ter	CF-causing
L467P	c.1400T>C	p.Leu467Pro	CF-causing
1548delG	c.1418delG	p.Gly473GlufsTer54	CF-causing
E474K	c.1420G>A	p.Glu474Lys	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
I502T	c.1505T>C	p.Ile502Thr	CF-causing
I507del	c.1519_1521delATC	p.Ile507del	CF-causing
F508del	c.1521_1523delCTT	p.Phe508del	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	p.?	CF-causing
1717-1G>A	c.1585-1G>A	p.?	CF-causing
1717-8G>A	c.1585-8G>A	p.?	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>A	p.Ser549Arg	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

Legacy Name	cDNA	Predicted Protein	CFTR2 24Sept2021
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
L558S	c.1673T>C	p.Leu558Ser	CF-causing
A559T	c.1675G>A	p.Ala559Thr	CF-causing
1811+1.6kbA>G	c.1680-886A>G	p.?	CF-causing
1811+1G>C	c.1679+1G>C	p.?	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	p.?	CF-causing
1811+1643G>T	c.1680-877G>T	p.?	CF-causing
1812-1G>A	c.1680-1G>A	p.?	CF-causing
R560S	c.1680A>C	p.Arg560Ser	CF-causing
A561E	c.1682C>A	p.Ala561Glu	CF-causing
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	p.?	CF-causing
1898+1G>C	c.1766+1G>C	p.?	CF-causing
1898+1G>T	c.1766+1G>T	p.?	CF-causing
1898+3A>G	c.1766+3A>G	p.?	CF-causing
1898+5G>T	c.1766+5G>T	p.?	CF-causing
1924del7	c.1792_1798delAAAACTA	p.Lys598GlyfsTer11	CF-causing
I601F	c.1801A>T	p.Ile601Phe	CF-causing
H609R	c.1826A>G	p.His609Arg	CF-causing
A613T	c.1837G>A	p.Ala613Thr	CF-causing
D614G	c.1841A>G	p.Asp614Gly	Varying clinical consequence
I618T	c.1853T>C	p.Ile618Thr	CF-causing [$]
G622D	c.1865G>A	p.Gly622Asp	Varying clinical consequence
G628R	c.1882G>A	p.Gly628Arg	CF-causing
G628R	c.1882G>C	p.Gly628Arg	CF-causing
2043delG	c.1911delG [#]	p.Gln637HisfsTer26	CF-causing
2055del9>A	c.1923_1931delinsA	p.Ser641ArgfsTer5	CF-causing
2075delA	c.1943delA	p.Asp648ValfsTer15	CF-causing

Legacy Name	cDNA	Predicted Protein	CFTR2 24Sept2021
E656X	c.1966G>T	p.Glu656Ter	CF-causing
2105-2117del13insAGAAA	c.1973_1985delinsAGAAA	p.Arg658LysfsTer4	CF-causing
2118del4	c.1986_1989delAACT	p.Thr663ArgfsTer8	CF-causing
E664X	c.1990G>T	p.Glu664Ter	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
G745X	c.2233G>T	p.Gly745Ter	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
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	p.?	CF-causing
E831X	c.2491G>T	p.Glu831Ter	CF-causing
2634insT	c.2502dupT [#]	p.Asp835Ter	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_2599del [7]	p.Ile864SerfsTer28	CF-causing
2732insA	c.2601dupA	p.Val868SerfsTer28	CF-causing
W882X	c.2645G>A	p.Trp882Ter	CF-causing
2789+5G>A	c.2657+5G>A	p.?	CF-causing

[7] Full variant name is c.2589_2599delAATTTGGTGCT

Legacy Name	cDNA	Predicted Protein	CFTR2 24Sept2021
2790-1G>C	c.2658-1G>C	p.?	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
R933G	c.2797A>G #	p.Arg933Gly	Varying clinical consequence
2942insT	c.2810dupT	p.Val938GlyfsTer37	CF-causing
2954delT	c.2822delT #	p.Leu941GlnfsTer27	CF-causing
2957delT	c.2825delT	p.Ile942ThrfsTer26	CF-causing
S945L	c.2834C>T	p.Ser945Leu	CF-causing
2991del32	c.2859_2890del [8]	p.Leu953PhefsTer11	CF-causing
3007delG	c.2875delG	p.Ala959HisfsTer9	CF-causing
3028delA	c.2896delA	p.Thr966ArgfsTer2	CF-causing
L967S	c.2900T>C	p.Leu967Ser	Non CF-causing $
G970R	c.2908G>C	p.Gly970Arg	CF-causing
G970D	c.2909G>A	p.Gly970Asp	CF-causing
S977F	c.2930C>T	p.Ser977Phe	Varying clinical consequence
D979V	c.2936A>T	p.Asp979Val	CF-causing
3120G>A	c.2988G>A	p.?	CF-causing
3120+1G>A	c.2988+1G>A	p.?	CF-causing
3121-1G>A	c.2989-1G>A	p.?	CF-causing
3121-2A>G	c.2989-2A>G	p.?	CF-causing
3132delTG	c.3002_3003delTG	p.Val1001AspfsTer45	CF-causing
3143del9	c.3011_3019delCTATAGCAG [9]	p.Ala1004_Ala1006del	CF-causing
A1006E	c.3017C>A	p.Ala1006Glu	CF-causing
3171delC	c.3039delC	p.Tyr1014ThrfsTer9	CF-causing
3171insC	c.3039dupC	p.Tyr1014LeufsTer33	CF-causing
F1016S	c.3047T>C	p.Phe1016Ser	Varying clinical consequence
3199del6	c.3067_3072delATAGTG ^	p.Ile1023_Val1024del	CF-causing
Y1032C	c.3095A>G	p.Tyr1032Cys	Varying clinical consequence
Q1035X	c.3103C>T #	p.Gln1035Ter	CF-causing
T1036N	c.3107C>A	p.Thr1036Asn	CF-causing
Q1042X	c.3124C>T	p.Gln1042Ter	CF-causing
3271delGG	c.3139_3139+1delGG	p.? [10]	CF-causing
3272-26A>G	c.3140-26A>G	p.?	CF-causing
F1052V	c.3154T>G	p.Phe1052Val	Varying clinical consequence

[8] Full variant name is c.2859_2890delACATTCTGTTCTTCAAGCACCTATGTCAACCC

[9] Alternative historical names for this variant is c.3011_3019delCTATAGCAG.

[10] Alternative historical name for this variant is p.Gly1047GlnfsTer28

Legacy Name	cDNA	Predicted Protein	CFTR2 24Sept2021
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.3220T>C	p.Phe1074Leu	Varying clinical consequence
F1074L	c.3222T>G	p.Phe1074Leu	Varying clinical consequence
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
W1098R	c.3292T>C	p.Trp1098Arg	CF-causing
W1098X	c.3293G>A	p.Trp1098Ter	CF-causing
W1098X	c.3294G>A	p.Trp1098Ter	CF-causing
W1098C	c.3294G>C	p.Trp1098Cys	CF-causing
W1098C	c.3294G>T	p.Trp1098Cys	CF-causing
F1099L	c.3297C>A	p.Phe1099Leu	Varying clinical consequence
Q1100P	c.3299A>C [&]	p.Gln1100Pro	-
M1101K	c.3302T>A	p.Met1101Lys	CF-causing
M1101R	c.3302T>G	p.Met1101Arg	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	p.?	CF-causing
R1128X	c.3382A>T [#]	p.Arg1128Ter	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	p.?	CF-causing
3600+2insT	c.3468+2dupT	p.?	CF-causing
3600+5G>A	c.3468+5G>A	p.?	CF-causing
R1158X	c.3472C>T	p.Arg1158Ter	CF-causing
S1159P	c.3475T>C	p.Ser1159Pro	CF-causing
S1159F	c.3476C>T	p.Ser1159Phe	CF-causing
R1162X	c.3484C>T	p.Arg1162Ter	CF-causing
3659delC	c.3528delC	p.Lys1177SerfsTer15	CF-causing
3667ins4	c.3532_3535dupTCAA	p.Thr1179IlefsTer17	CF-causing

Legacy Name	cDNA	Predicted Protein	CFTR2 24Sept2021
S1196X	c.3587C>G	p.Ser1196Ter	CF-causing
3732delA	c.3600delA [#]	p.Asp1201MetfsTer10	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	p.?	CF-causing
3849+4A>G	c.3717+4A>G	p.?	CF-causing
3849+5G>A	c.3717+5G>A	p.?	CF-causing
3849+40A>G	c.3717+40A>G	p.?	CF-causing
3849+10kbC>T	c.3718-2477C>T	p.?	CF-causing
3850-1G>A	c.3718-1G>A	p.?	CF-causing
3850-3T>G	c.3718-3T>G	p.?	CF-causing
V1240G	c.3719T>G	p.Val1240Gly	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
G1249R	c.3745G>A	p.Gly1249Arg	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
I1269N	c.3806T>A	p.Ile1269Asn	CF-causing
D1270N	c.3808G>A	p.Asp1270Asn	Varying clinical consequence
W1274X	c.3822G>A [#]	p.Trp1274Ter	CF-causing
W1282X	c.3846G>A	p.Trp1282Ter	CF-causing
W1282X;R1283M	c.3846_3848delinsAAT [11]	p.Trp1282Ter	CF-causing
R1283M	c.3848G>T	p.Arg1283Met	CF-causing
Q1291R	c.3872A>G	p.Gln1291Arg	Varying clinical consequence
Q1291H	c.3873G>C	p.Gln1291His	Varying clinical consequence
4005+1G>A	c.3873+1G>A	p.?	CF-causing
4005+2T>C	c.3873+2T>C	p.?	CF-causing
4010del4	c.3883_3886delATTT	p.Ile1295PhefsTer32	CF-causing
4015delA	c.3883delA	p.Ile1295PhefsTer33	CF-causing
4016insT (4021dupT)	c.3889dupT	p.Ser1297PhefsTer5	CF-causing
4022insT	c.3891dupT	p.Gly1298TrpfsTer4	CF-causing

[11] Alternative historical name for this variant is c.[3846G>A;3848G>T], p.[Trp1282Ter;Arg1283Met]

Legacy Name	cDNA	Predicted Protein	CFTR2 24Sept2021
4040delA	c.3908delA	p.Asn1303ThrfsTer25	CF-causing
N1303K	c.3909C>G	p.Asn1303Lys	CF-causing
W1310X	c.3929G>A [#]	p.Trp1310Ter	CF-causing
Q1313X	c.3937C>T	p.Gln1313Ter	CF-causing
L1324P	c.3971T>C	p.Leu1324Pro	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.Val1360ThrfsTer3	CF-causing
4218insT	c.4086dupT	p.Lys1363Ter	CF-causing
I1366N	c.4097T>A	p.Ile1366Asn	CF-causing
E1371X	c.4111G>T	p.Glu1371Ter	CF-causing
H1375P	c.4124A>C	p.His1375Pro	CF-causing
4259del5	c.4127_4131delTGGAT	p.Leu1376SerfsTer8	CF-causing
4271delC	c.4139delC [&]	p.Thr1380AsnfsTer4	-
Q1382X	c.4144C>T	p.Gln1382Ter	CF-causing
4279insA	c.4147dupA	p.Ile1383AsnfsTer3	CF-causing
4326delTC	c.4197_4198delCT [12]	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	p.?	CF-causing
4374+1G>A	c.4242+1G>A	p.?	CF-causing
4382delA	c.4251delA	p.Glu1418ArgfsTer14	CF-causing
4428insGA	c.4300_4301dupAG	p.Ser1435GlyfsTer14	CF-causing
S1455X	c.4364C>G	p.Ser1455Ter	Varying clinical consequence
Q1476X	c.4426C>T	p.Gln1476Ter	Varying clinical consequence
L1480P	c.4439T>C	p.Leu1480Pro	Varying clinical consequence
CFTRdele2,3	c.54-5940_273+10250del	p.? [13]	CF-causing
CFTRdele22,23	c.3964-78_4242+577del	p.?	CF-causing

HGVS nomenclature version 20.05

- Reference sequences: coding variants NM_000492.3, NP_000483.3
- Reference sequences: non-coding variants NC_000007.13 (NM_000492.3)

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

[12] This variant was originally entered into the SeqNext mutation Database as c.4196_4197delTC, it was corrected on April 12, 2018.

[13] Alternative historical name for this variant is p.Ser18ArgfsX16