

Appendix 10 – Gene variants' nomenclature

More details are available from CFTR2 database. www.cftr2.org. Table adapted from CFTR2 database.

Variant cDNA name (ordered 5' to 3')	Variant protein name	Variant legacy name	Variant final determination 11 March 2019
c.-9_14del23	No protein name	124del23bp	CF-causing
c.(?_1)_(53+1_54-1)del	p.Glu2GlyfsX17	CFTRdele1	CF-causing
c.1A>G	p.Met1Val	M1V	CF-causing
c.4C>T	p.Gln2X	Q2X	CF-causing
c.11C>A	p.Ser4X	S4X	CF-causing
c.14C>T	p.Pro5Leu	P5L	Varying clinical consequence
c.38C>T	p.Ser13Phe	S13F	CF-causing
c.44T>C	p.Leu15Pro	L15P	CF-causing
c.50delT	p.Phe17SerfsX8	182delT	CF-causing
c.(53+1_54-1)_(164+1_165-1)del	No protein name	CFTRdele2	CF-causing
c.(53+1_54-1)_(489+1_490-1)del	No protein name	CFTRdele2-4	CF-causing
c.53+1G>T	No protein name	185+1G->T	CF-causing
c.54-5940_273+10250del21kb	p.Ser18ArgfsX16	CFTRdele2,3	CF-causing
c.57G>A	p.Trp19X	W19X	CF-causing
c.79G>A	p.Gly27Arg	G27R	CF-causing
c.79G>T	p.Gly27X	G27X	CF-causing
c.88C>T	p.Gln30X	Q30X	CF-causing
c.91C>T	p.Arg31Cys	R31C	Non CF-causing
c.92G>T	p.Arg31Leu	R31L	Unknown
c.115C>T	p.Gln39X	Q39X	CF-causing
c.137C>A	p.Ala46Asp	A46D	CF-causing
c.164+1G>A	No protein name	296+1G->A	CF-causing

c.164+1G>T	No protein name	296+1G->T	CF-causing
c.(164+1_165-1)_(1584+1_1585-1)del(2619+1_2620-1)_(2988+1_2989-1)del	No protein name	CFTRdele3-10,14b-16	CF-causing
c.164+28A>G	No protein name	296+28A->G	Unknown
c.164+2T>C	No protein name	296+2T->C	CF-causing
c.164+3_164+4insT	No protein name	296+3insT	CF-causing
c.165-3C>T	No protein name	297-3C->T	CF-causing
c.165-1G>A	No protein name	297-1G->A	CF-causing
c.166G>A	p.Glu56Lys	E56K	CF-causing
c.169T>G	p.Trp57Gly	W57G	CF-causing
c.170G>A or c.171G>A	p.Trp57X	W57X	CF-causing
c.175dupA	p.Arg59LysfsX10	306insA	CF-causing
c.174_177delTAGA	p.Asp58GluufsX32	306delTAGA	CF-causing
c.178G>A	p.Glu60Lys	E60K	CF-causing
c.178G>T	p.Glu60X	E60X	CF-causing
c.200C>T	p.Pro67Leu	P67L	CF-causing
c.220C>T	p.Arg74Trp	R74W	Varying clinical consequence
c.223C>T	p.Arg75X	R75X	CF-causing
c.224G>A	p.Arg75Gln	R75Q	Non CF-causing
c.233dupT	p.Trp79LeufsX32	365-366insT	CF-causing
c.254G>A	p.Gly85Glu	G85E	CF-causing
c.262_263delTT	p.Leu88IleufsX22	394delTT	CF-causing
c.263T>A or c.263T>G	p.Leu88X	L88X	CF-causing
c.271G>A	p.Gly91Arg	G91R	CF-causing
c.(273+1_274-1)_(1116+1_1117-1)del	No protein name	CFTRdele4-7	CF-causing
c.(273+1_274-1)_(1679+1_1680-1)del	No protein name	CFTRdele4-11	CF-causing
c.(273+1_274-1)_(1116+1_1117-1)del(1584+1_1585-1)_(3468+1_3469-1)del	No protein name	CFTR50kdel	CF-causing
c.273+1G>A	No protein name	405+1G->A	CF-causing

c.273+3A>C	No protein name	405+3A->C	CF-causing
c.274-2A>G	No protein name	406-2A->G	CF-causing
c.274-1G>A	No protein name	406-1G->A	CF-causing
c.274G>A	p.Glu92Lys	E92K	CF-causing
c.274G>T	p.Glu92X	E92X	CF-causing
c.292C>T	p.Gln98X	Q98X	CF-causing
c.293A>G	p.Gln98Arg	Q98R	CF-causing
c.296C>T	p.Pro99Leu	P99L	CF-causing
c.305T>G	p.Leu102Arg	L102R	CF-causing
c.310delA	p.Arg104GlufsX3	442delA	CF-causing
c.313delA	p.Ile105SerfsX2	444delA	CF-causing
c.325_327delTATinsG	p.Tyr109GlyfsX4	457TAT->G	CF-causing
c.328G>C	p.Asp110His	D110H	CF-causing
c.330C>A	p.Asp110Glu	D110E	Varying clinical consequence
c.349C>T	p.Arg117Cys	R117C	CF-causing
c.349C>G	p.Arg117Gly	R117G	Varying clinical consequence
c.350G>A	p.Arg117His	R117H	Varying clinical consequence
c.350G>T	p.Arg117Leu	R117L	Varying clinical consequence
c.[350G>A;1210-12T[5]]	p[.Arg117His; No protein name]	R117H;5T	CF-causing
c.[350G>A;1210-12T[7]]	p.[Arg117His; No protein name]	R117H;7T	Varying clinical consequence
c.377G>A	p.Gly126D	G126D	CF-causing
c.409delC	p.Leu137SerfsX16	541delC	CF-causing
c.413_415dupTAC	p.Leu138dup	L138ins	CF-causing
c.416A>G	p.His139Arg	H139R	CF-causing

c.442delA	p.Ile148LeufsX5	574delA	CF-causing
c.443T>C	p.Ile148Thr	I148T	Non CF-causing
c.470_483delTTAGTTTGATTTAT	p.Phe157X	602del14	CF-causing
c.481T>G	p.Tyr161Asp	Y161D	CF-causing
c.489+1G>T	No protein name	621+1G->T	CF-causing
c.489+3A>G	No protein name	621+3A->G	Varying clinical consequence
c.494T>C	p.Leu165Ser	L165S	CF-causing
c.509G>A	p.Arg170His	R170H	Non CF-causing
c.531delT	p.Ile177MetfsX12	663delT	CF-causing
c.532G>A	p.Gly178Arg	G178R	CF-causing
c.543_546delTAGT	p.Leu183PhefsX5	675del4	CF-causing
c.577G>T	p.Glu193X	E193X	CF-causing
c.579+1G>T	No protein name	711+1G->T	CF-causing
c.579+3A>G	No protein name	711+3A->G	CF-causing
c.579+5G>A	No protein name	711+5G->A	CF-causing
c.580-1G>T	No protein name	712-1G->T	CF-causing
c.595C>T	p.His199Tyr	H199Y	CF-causing
c.601G>A	p.Val201Met	V201M	Unknown
c.613C>T	p.Pro205Ser	P205S	CF-causing
c.617T>G	p.Leu206Trp	L206W	CF-causing
c.647G>A	p.Trp216X	W216X	CF-causing
c.658C>T	p.Gln220X	Q220X	CF-causing
c.680T>G	p.Leu227Arg	L227R	CF-causing
c.695T>A	p.Val232Asp	V232D	CF-causing
c.717delG	p.Leu240X	849delG	CF-causing
c.722_743delGGAGAATGATGATGAAGTACAG	p.Gly241GlufsX13	852del22	CF-causing
c.(743+1_744-1)_(1584+1_1585-1)dup	No protein name	CFTRdup6b-10	CF-causing
c.794T>G	p.Met265Arg	M265R	Varying clinical consequence

c.803delA	p.Asn268IlefsX17	935delA	CF-causing
c.825C>G	p.Tyr275X	Y275X	CF-causing
c.828C>A	p.Cys276X	C276X	CF-causing
c.850dupA	p.Met284AsnfsX3	977insA	CF-causing
c.861_865delCTTAA	p.Asn287LysfsX19	991del5	CF-causing
c.933C>G	p.Phe311Leu	F311L	CF-causing
c.948delT	p.Phe316LeufsX12	1078delT	CF-causing
c.958T>G	p.Leu320Val	L320V	Non CF-causing
c.987delA	p.Gly330GluX39	1119delA	CF-causing
c.988G>T	p.Gly330X	G330X	CF-causing
c.1000C>T	p.Arg334Trp	R334W	CF-causing
c.1001G>A	p.Arg334Gln	R334Q	Varying clinical consequence
c.1001G>T	p.Arg334Leu	R334L	CF-causing
c.1006_1007insG	p.Ile336SerfsX28	1138insG	CF-causing
c.1007T>A	p.Ile336Lys	I336K	CF-causing
c.1013C>T	p.Thr338Ile	T338I	CF-causing
c.1021T>C	p.Ser341Pro	S341P	CF-causing
c.1021_1022dupTC	p.Phe342HisfsX28	1154insTC	CF-causing
c.1029delC	p.Cys343X	1161delC	CF-causing
c.1037T>C	p.Leu346Pro	L346P	CF-causing
c.1040G>A	p.Arg347His	R347H	CF-causing
c.1040G>C	p.Arg347Pro	R347P	CF-causing
c.1046C>T	p.Ala349Val	A349V	Unknown significance
c.1054C>T	p.Arg352Trp	R352W	Varying clinical consequence
c.1055G>A	p.Arg352Gln	R352Q	CF-causing
c.[1075C>A;1079C>A]	p.[Gln359Lys;Thr360Lys]	Q359K/T360K	CF-causing
c.1081delT	p.Trp361GlyfsX8	1213delT	CF-causing
c.1116+1G>A	No protein name	1248+1G->A	CF-causing

c.1117-1G>A	No protein name	1249-1G->A	CF-causing
c.1130dupA	p.Gln378AlafsX4	1259insA	CF-causing
c.1155_1156dupTA	p.Asn386lfeFsX3	1288insTA	CF-causing
c.1202G>A or c.1203G>A	p.Trp401X	W401X	CF-causing
c.1209+1G>A	No protein name	1341+1G->A	CF-causing
c.1210-12T[5]	No protein name	5T	Varying clinical consequence
c.1210-33_1210-6GT[11]T[4]	No protein name	5T;TG11	Varying clinical consequence
c.1210-33_1210-6GT[12]T[4]	No protein name	5T;TG12	Varying clinical consequence
c.1210-33_1210-6GT[13]T[4]	No protein name	5T;TG13	Varying clinical consequence
c.1210-12T[7]	No protein name	7T	Non CF-causing
c.1210-12T[9]	No protein name	9T	Non CF-causing
c.1211delG	p.Gly404AspfsX38	1343delG	CF-causing
c.1240C>T	p.Gln414X	Q414X	CF-causing
c.1301_1307delCACTTCT	p.Ser434LeufsX6	1429del7	CF-causing
c.1327G>T	p.Asp443Tyr	D443Y	Varying clinical consequence
c.1327_1330dupGATA	p.Ile444ArgfsX3	1461ins4	CF-causing
c.1340delA	p.Lys447ArgfsX2	1471delA	CF-causing
c.1358T>C	p.Leu453Ser	L453S	CF-causing
c.1364C>A	p.Ala455Glu	A455E	CF-causing
c.1365_1366delGG	p.Val456CysfsX25	1497delGG	CF-causing
c.1367T>C	p.Val456Ala	V456A	CF-causing
c.1373delG	p.Gly458AspfsX11	1504delG	CF-causing
c.1393-1G>A	No protein name	1525-1G->A	CF-causing
c.1393-2A>G	No protein name	1525-2A->G	CF-causing
c.1397C>A or c.1397C>G	p.Ser466X	S466X	CF-causing

c.[1397C>G;3209G>A]	p.[Ser466X;Arg1070Gln]	S466X;R1070Q	CF-causing
c.1400T>C	p.Leu467Pro	L467P	CF-causing
c.1408A>G	p.Met470Val	M470V	Non CF-causing
c.1418delG	p.Gly473GluX54	1548delG	CF-causing
c.1420G>A	p.Glu474Lys	E474K	CF-causing
c.1466C>A	p.Ser489X	S489X	CF-causing
c.1475C>T	p.Ser492Phe	S492F	CF-causing
c.1477_1478delCA	p.Gln493ValfsX10	1609delCA	CF-causing
c.1477C>T	p.Gln493X	Q493X	CF-causing
c.1487G>A	p.Trp496X	W496X	CF-causing
c.1505T>C	p.Ile502Thr	I502T	CF-causing
c.1519_1521delATC	p.Ile507del	I507del	CF-causing
c.1521_1523delCTT	p.Phe508del	F508del	CF-causing
c.[1521_1523delCTT;3080T>C]	p.[Phe508del;Ile1027Thr]	F508del;I1027T	CF-causing
c.1523T>G	p.Phe508Cys	F508C	Non CF-causing
c.1538A>G	p.Asp513Gly	D513G	CF-causing
c.1545_1546delTA	p.Tyr515X	1677delTA	CF-causing
c.1558G>T	p.Val520Phe	V520F	CF-causing
c.1572C>A	p.Cys524X	C524X	CF-causing
c.1573C>T	p.Gln525X	Q525X	CF-causing
c.1584+1G>A	No protein name	1716+1G->A	CF-causing
c.(1584+1_1585-1)_(1679+1_1680-1)del	No protein name	CFTRdele11	CF-causing
c.1585-1G>A	No protein name	1717-1G->A	CF-causing
c.1585-8G>A	No protein name	1717-8G->A	CF-causing
c.1624G>T	p.Gly542X	G542X	CF-causing
c.1645A>C or c.1647T>G or c.1647T>A	p.Ser549Arg	S549R	CF-causing
c.1646G>A	p.Ser549Asn	S549N	CF-causing
c.1648G>T	p.Gly550X	G550X	CF-causing
c.1650delA	p.Gly551ValfsX8	1782delA	CF-causing
c.1651G>A	p.Gly551Ser	G551S	CF-causing

c.1652G>A	p.Gly551Asp	G551D	CF-causing
c.1654C>T	p.Gln552X	Q552X	CF-causing
c.1657C>T	p.Arg553X	R553X	CF-causing
c.1670delC	p.Ser557PhefsX2	1802delC	CF-causing
c.1673T>C	p.Leu558Ser	L558S	CF-causing
c.1675G>A	p.Ala559Thr	A559T	CF-causing
c.1679+1G>C	No protein name	1811+1G->C	CF-causing
c.1679G>A	p.Arg560Lys	R560K	CF-causing
c.1679G>C	p.Arg560Thr	R560T	CF-causing
c.1679+1G>A	No protein name	1811+1G->A	CF-causing
c.1680-886A>G	No protein name	1811+1634A->G or 1811+1.6kbA->G	CF-causing
c.1680-877G>T	No protein name	1811+1643G->T	CF-causing
c.1680-1G>A	No protein name	1812-1G->A	CF-causing
c.1680A>C	p.Arg560Ser	R560S	CF-causing
c.1682C>A	p.Ala561Glu	A561E	CF-causing
c.1684G>A	p.Val562Ile	V562I	Non CF-causing
c.1687T>A	p.Tyr563Asn	Y563N	CF-causing
c.1687T>G	p.Tyr563Asp	Y563D	CF-causing
c.1692delA	p.Asp565MetfsX7	1824delA	CF-causing
c.1703delT	p.Leu568CysfsX4	1833delT	CF-causing
c.1705T>G	p.Tyr569Asp	Y569D	CF-causing
c.1721C>A	p.Pro574His	P574H	CF-causing
c.1724T>A	p.Phe575Tyr	F575Y	Varying clinical consequence
c.1727G>C	p.Gly576Ala	G576A	Non CF-causing
c.1736A>G	p.Asp579Gly	D579G	Varying clinical consequence
c.1753G>T	p.Glu585X	E585X	CF-causing

c.1763A>T	p.Glu588Val	E588V	Varying clinical consequence
c.1766+1G>A	No protein name	1898+1G->A	CF-causing
c.1766+1G>C	No protein name	1898+1G->C	CF-causing
c.1766+1G>T	No protein name	1898+1G->T	CF-causing
c.(1766+1_1767-1)_(2619+1_2620-1)del	No protein name	CFTRdele13,14a	CF-causing
c.1766+3A>G	No protein name	1898+3A->G	CF-causing
c.1766+5G>T	No protein name	1898+5G->T	CF-causing
c.1792_1798delAAAACTA	p.Lys598GlyfsX11	1924del7	CF-causing
c.1826A>G	p.His609Arg	H609R	CF-causing
c.1837G>A	p.Ala613Thr	A613T	CF-causing
c.1841A>G	p.Asp614Gly	D614G	Varying clinical consequence
c.1865G>A	p.Gly622Asp	G622D	Varying clinical consequence
c.1882G>C or c.1882G>A	p.Gly628Arg	G628R	CF-causing
c.1923_1931del9insA	p.Ser641ArgfsX5	2055del9->A	CF-causing
c.1943delA	p.Asp648ValfsX15	2075delA	CF-causing
c.1973_1985del13insAGAAA	p.Arg658LysfsX4	2105- 2117del13insAGAAA	CF-causing
c.1986_1989delAACT	p.Thr663ArgfsX8	2118del4	CF-causing
c.2002C>T	p.Arg668Cys	R668C	Non CF-causing
c.2012delT	p.Leu671X	2143delT	CF-causing
c.2017G>T	p.Gly673X	G673X	CF-causing
c.2051_2052delAAinsG	p.Lys684SerfsX38	2183AA->G	CF-causing
c.2052dupA	p.Gln685ThrfsX4	2184insA	CF-causing
c.2052delA	p.Lys684AsnfsX38	2184delA	CF-causing
c.2053dupC	p.Gln685ProfsX84	2185insC	CF-causing
c.2053C>T	p.Gln685X	Q685X	CF-causing
c.2125C>T	p.Arg709X	R709X	CF-causing

c.2128A>T	p.Lys710X	K710X	CF-causing
c.2143C>T	p.Gln715X	Q715X	CF-causing
c.2158C>T	p.Gln720X	Q720X	CF-causing
c.2175dupA	p.Glu726ArgfsX4	2307insA	CF-causing
c.2195T>G	p.Leu732X	L732X	CF-causing
c.2215delG	p.Val739TyrfsX16	2347delG	CF-causing
c.2241_2248delGATACTGC	p.Ile748SerfsX28	2372del8	CF-causing
c.2249C>T	p.Pro750Leu	P750L	Varying clinical consequence
c.2260G>A	p.Val754Met	V754M	Non CF-causing
c.2290C>T	p.Arg764X	R764X	CF-causing
c.2353C>T	p.Arg785X	R785X	CF-causing
c.2374C>T	p.Arg792X	R792X	CF-causing
c.2421A>G	p.Ile807Met	I807M	Non CF-causing
c.2423_2424dupAT	p.Ser809IlefsX13	2556insAT	CF-causing
c.2453delT	p.Leu818TrpfsX3	2585delT	CF-causing
c.2463_2464delTG	p.Ser821ArgfsX4	2594delGT	CF-causing
c.2464G>T	p.Glu822X	E822X	CF-causing
c.2490+1G>A	No protein name	2622+1G->A	CF-causing
c.2491G>T	p.Glu831X	E831X	CF-causing
c.2506G>T	p.Asp836Tyr	D836Y	Non CF-causing
c.2537G>A or c.2538G>A	p.Trp846X	W846X	CF-causing
c.2547C>A	p.Tyr849X	Y849X	CF-causing
c.2551C>T	p.Arg851X	R851X	CF-causing
c.2562T>C or c.2562T>G or c.2562T>A	p.Thr854Thr	T854T	Non CF-causing
c.2583delT	p.Phe861LeufsX3	2711delT	CF-causing
c.2589_2599delAATTTGGTGCT	p.Ile864SerfsX28	2721del11	CF-causing
c.2601dupA	p.Val868SerfsX28	2732insA	CF-causing
c.(2619+1_2620-1)_(3367+1_3368-1)del	No protein name	CFTRdele14b-17b	CF-causing
c.2620-26A>G	No protein name	2752-26A->G	Unknown significance

c.2645G>A	p.Trp882X	W882X	CF-causing
c.2657+2_2657+3insA	No protein name	2789+2insA	Unknown significance
c.2657+5G>A	No protein name	2789+5G->A	CF-causing
c.2658-1G>C	No protein name	2790-1G->C	CF-causing
c.2668C>T	p.Gln890X	Q890X	CF-causing
c.2735C>A	p.Ser912X	S912X	CF-causing
c.2735C>T	p.Ser912Leu	S912L	Unknown significance
c.2737_2738insG	p.Tyr913X	2869insG	CF-causing
c.2739T>A	p.Tyr913X	Y913X	CF-causing
c.2763_2764dupAG	p.Val922GlufsX2	2896insAG	CF-causing
c.2780T>C	p.Leu927Pro	L927P	CF-causing
c.2810dupT	p.Val938GlyfsX37	2942insT	CF-causing
c.2825delT	p.Ile942ThrfsX26	2957delT	CF-causing
c.2834C>T	p.Ser945Leu	S945L	CF-causing
c.2859_2890delACATTCTGTTCTTCAAGCACCTATGTCAACCC	p.Leu953PhefsX11	2991del32	CF-causing
c.2875delG	p.Ala959HisfsX9	3007delG	CF-causing
c.2896delA	p.Thr966ArgfsX2	3028delA	CF-causing
c.2900T>C	p.Leu967Ser	L967S	Varying clinical consequence
c.2908G>C	p.Gly970Arg	G970R	CF-causing
c.(2908+1_2909-1)_(3367+1_3368-1)del	No protein name	CFTRdele16-17b	CF-causing
c.2909G>A	p.Gly970Asp	G970D	CF-causing
c.2930C>T	p.Ser977Phe	S977F	Varying clinical consequence
c.2936A>T	p.Asp979Val	D979V	CF-causing
c.2988G>A	No protein name	3120G->A	CF-causing
c.(2988+1_2989-1)_(3367+1_3368-1)del	No protein name	CFTRdele17a,17b	CF-causing
c.(2988+1_2989-1)_(3468+1_3469-1)del	No protein name	CFTRdele17a-18	CF-causing
c.2988+1G>A	No protein name	3120+1G->A	CF-causing
c.2989-1G>A	No protein name	3121-1G->A	CF-causing

c.2989-2A>G	No protein name	3121-2A->G	CF-causing
c.2989-977_3367+248del	No protein name	3121-977_3499+248del2515	CF-causing
c.2991G>C	p.Leu997Phe	L997F	Non CF-causing
c.3002_3003delTG	p.Val1001AspfsX45	3132delTG	CF-causing
c.3017C>A	p.Ala1006Glu	A1006E	CF-causing
c.3011_3019delCTATAGCAG or c.3009_3017delAGCTATAGC	p.Ala1004_Ala1006del	3143del9	CF-causing
c.3039delC	p.Tyr1014ThrfsX9	3171delC	CF-causing
c.3039dupC	p.Tyr1014LeufsX33	3171insC	CF-causing
c.3041A>G	p.Tyr1014Cys	Y1014C	Unknown significance
c.3047T>C	p.Phe1016Ser	F1016S	Varying clinical consequence
c.3080T>C	p.Ile1027Thr	I1027T	Non CF-causing
c.3095A>G	p.Tyr1032Cys	Y1032C	Varying clinical consequence
c.3107C>A	p.Thr1036Asn	T1036N	CF-causing
c.3124C>T	p.Gln1042X	Q1042X	CF-causing
c.3139_3139+1delGG	p.Gly1047GlnfsX28	3271delGG	CF-causing
c.3140-26A>G	No protein name	3272-26A->G	CF-causing
c.3154T>G	p.Phe1052Val	F1052V	Varying clinical consequence
c.3158C>T	p.Thr1053Ile	T1053I	Non CF-causing
c.3160C>G	p.His1054Asp	H1054D	CF-causing
c.3181G>C	p.Gly1061Arg	G1061R	CF-causing
c.3194T>C	p.Leu1065Pro	L1065P	CF-causing
c.3196C>T	p.Arg1066Cys	R1066C	CF-causing
c.3197G>A	p.Arg1066His	R1066H	CF-causing
c.3205G>A	p.Gly1069Arg	G1069R	Varying clinical consequence

c.3208C>T	p.Arg1070Trp	R1070W	Varying clinical consequence
c.3209G>A	p.Arg1070Gln	R1070Q	Varying clinical consequence
c.3217dupT	p.Tyr1073LeufsX3	3349insT	CF-causing
c.3222T>A or c.3220T>C or c.3222T>G	p.Phe1074Leu	F1074L	Varying clinical consequence
c.3230T>C	p.Leu1077Pro	L1077P	CF-causing
c.3266G>A	p.Trp1089X	W1089X	CF-causing
c.3276C>A or c.3276C>G	p.Tyr1092X	Y1092X	CF-causing
c.3292T>C	p.Trp1098Arg	W1098R	CF-causing
c.3293G>A or c.3294G>A	p.Trp1098X	W1098X	CF-causing
c.3294G>C or c.3294G>T	p.Trp1098Cys	W1098C	CF-causing
c.3297C>A	p.Phe1099Leu	F1099L	Varying clinical consequence
c.3302T>A	p.Met1101Lys	M1101K	CF-causing
c.3302T>G	p.Met1101Arg	M1101R	CF-causing
c.3304A>T	p.Arg1102X	R1102X	CF-causing
c.3310G>T	p.Glu1104X	E1104X	CF-causing
c.3353C>T	p.Ser1118Phe	S1118F	CF-causing
c.(3367+1+3368-1)_(3468+1_3469-1)del	No protein name	CFTRdele18	CF-causing
c.3368-2A>G	No protein name	3500-2A->G	CF-causing
c.3435G>A	p.Trp1145X	W1145X	CF-causing
c.3454G>C	p.Asp1152His	D1152H	Varying clinical consequence
c.3458T>A	p.Val1153Glu	V1153E	Varying clinical consequence
c.3468G>A	No protein name	3600G->A	CF-causing
c.(3468+1_3469-1)_(3717+1_3718-1)del	No protein name	CFTRdele19	CF-causing
c.(3468+1_3469-1)_(3963+1_3964-1)del	No protein name	CFTRdele19-21	CF-causing

c.3468+2dupT	No protein name	3600+2insT	CF-causing
c.3468+5G>A	No protein name	3600+5G->A	CF-causing
c.3472C>T	p.Arg1158X	R1158X	CF-causing
c.3475T>C	p.Ser1159Pro	S1159P	CF-causing
c.3476C>T	p.Ser1159Phe	S1159F	CF-causing
c.3484C>T	p.Arg1162X	R1162X	CF-causing
c.3485G>T	p.Arg1162Leu	R1162L	Non CF-causing
c.3528delC	p.Lys1177SerfsX15	3659delC	CF-causing
c.3532_3535dupTCAA	p.Thr1179IlefsX17	3667ins4	CF-causing
c.3587C>G	p.Ser1196X	S1196X	CF-causing
c.3605delA	p.Asp1202AlafsX9	3737delA	CF-causing
c.3611G>A or c.3612G>A	p.Trp1204X	W1204X	CF-causing
c.3659delC	p.Thr1220LysfsX8	3791delC	CF-causing
c.366T>A	p.Tyr122X	Y122X	CF-causing
c.3691delT	p.Ser1231ProfsX4	3821delT	CF-causing
c.3700A>G	p.Ile1234Val	I1234V	CF-causing
c.3705T>G	p.Ser1235Arg	S1235R	Non CF-causing
c.3717G>A	No protein name	3849G->A	CF-causing
c.3717+4A>G	No protein name	3849+4A->G	CF-causing
c.3717+5G>A	No protein name	3849+5G->A	CF-causing
c.3717+40A>G	No protein name	3849+40A->G	CF-causing
c.3718-2477C>T	No protein name	3849+10kbC->T	CF-causing
c.3718-1G>A	No protein name	3850-1G->A	CF-causing
c.3718-3T>G	No protein name	3850-3T->G	CF-causing
c.3719T>G	p.Val1240Gly	V1240G	CF-causing
c.3731G>A	p.Gly1244Glu	G1244E	CF-causing
c.3737C>T	p.Thr1246Ile	T1246I	Varying clinical consequence
c.3744delA	p.Lys1250ArgfsX9	3876delA	CF-causing
c.3745G>A	p.Gly1249Arg	G1249R	CF-causing

c.3747delG	p.Lys1250ArgfsX9	3878delG	CF-causing
c.3752G>A	p.Ser1251Asn	S1251N	CF-causing
c.3761T>G	p.Leu1254X	L1254X	CF-causing
c.3763T>C	p.Ser1255Pro	S1255P	CF-causing
c.3764C>A	p.Ser1255X	S1255X	CF-causing
c.3773dupT	p.Leu1258PhefsX7	3905insT	CF-causing
c.3806T>A	p.Ile1269Asn	I1269N	CF-causing
c.3808G>A	p.Asp1270Asn	D1270N	Varying clinical consequence
c.3846G>A	p.Trp1282X	W1282X	CF-causing
c.3848G>T	p.Arg1283Met	R1283M	CF-causing
c.3872A>G	p.Gln1291Arg	Q1291R	Varying clinical consequence
c.3873+1G>A	No protein name	4005+1G->A	CF-causing
c.(3873+1_3874-1)_(3963+1_3964-1)del	No protein name	CFTRdele21	CF-causing
c.3873+2T>C	No protein name	4005+2T->C	CF-causing
c.3883_3886delATTT	p.Ile1295PhefsX32	4010del4	CF-causing
c.3883delA	p.Ile1295PhefsX33	4015delA	CF-causing
c.3889dupT	p.Ser1297PhefsX5	4016insT	CF-causing
c.3891dupT	p.Gly1298TrpfsX4	4022insT	CF-causing
c.3908delA	p.Asn1303ThrfsX25	4040delA	CF-causing
c.3909C>G	p.Asn1303Lys	N1303K	CF-causing
c.3937C>T	p.Gln1313X	Q1313X	CF-causing
c.(3963+1_3964-1)(*1_?)del	No protein name	CFTRdele22-24	CF-causing
c.3964-78_4242+577del	No protein name	CFTRdele22,23	CF-causing
c.3971T>C	p.Leu1324Pro	L1324P	CF-causing
c.3988C>T	p.Gln1330X	Q1330X	CF-causing
c.4004T>C	p.Leu1335Pro	L1335P	CF-causing
c.4036_4042del	p.Leu1346MetfsX6	4168delCTAAGCC	CF-causing
c.4046G>A	p.Gly1349Asp	G1349D	CF-causing

c.4077_4080delTGTTinsAA	No protein name	4209TGTT->AA	CF-causing
c.4086dupT	p.Lys1363X	4218insT	CF-causing
c.4111G>T	p.Glu1371X	E1371X	CF-causing
c.4124A>C	p.His1375Pro	H1375P	CF-causing
c.4127_4131delTGGAT	p.Leu1376SerfsX8	4259del5	CF-causing
c.4144C>T	p.Gln1382X	Q1382X	CF-causing
c.4147dupA	p.Ile1383AsnfsX3	4279insA	CF-causing
c.4197_4198delCT	p.Cys1400X	4326delTC	CF-causing
c.4231C>T	p.Gln1411X	Q1411X	CF-causing
c.4234C>T	p.Gln1412X	Q1412X	CF-causing
c.4242+1G>T	No protein name	4374+1G->T	CF-causing
c.4242+1G>A	No protein name	4374+1G->A	CF-causing
c.4251delA	p.Glu1418ArgfsX14	4382delA	CF-causing
c.4300_4301dup	p.Ser1435GlyfsX14	4428insGA	CF-causing