

List of CFTR2 mutations**Date:** 13 August 2015**Number of patients in CFTR2:** 88,664

**Clinical and
Functional
Translation
of CFTR**

This detailed medical and genetics information is complicated and potentially confusing. We encourage you to discuss this information with your doctor, a genetic counselor, or a CF specialist. The information shown is for educational purposes only and is not intended for diagnostic use. You should not make any medical or reproductive decisions or change your health behavior based on this information without talking to your doctor.

Mutation cDNA name	Mutation protein name	Mutation legacy name	# alleles in CFTR2	Allele frequency in CFTR2 (of 141,341 identified mutations)*	Mutation final determination 20 Feb 2015 (previous version)	Mutation final determination 13 August 2015 (current version)	Change from previous version?
c.-9_14del23	No protein name	124del23bp	6	0.00004		CF-causing	Yes
c.(?_1)_(53+1_54-1)del	p.Glu2GlyfsX17	CFTRdelete1	6	0.00004		CF-causing	Yes
c.1A>G	p.Met1Val	M1V	26	0.00018	CF-causing	CF-causing	No
c.11C>A	p.Ser4X	S4X	14	0.00010		CF-causing	Yes
c.50delT	p.Phe17SerfsX8	182delT	9	0.00006		CF-causing	Yes
c.(53+1_54-1)_(164+1_165-1)del	No protein name	CFTRdelete2	41	0.00029		CF-causing	Yes
c.(53+1_54-1)_(489+1_490-1)del	No protein name	CFTRdelete2-4	4	0.00003		CF-causing	Yes
c.53+1G>T	No protein name	185+1G->T	8	0.00006		CF-causing	Yes
c.54-5940_273+10250 del21kb	p.Ser18ArgfsX16	CFTRdelete2,3	417	0.00295	CF-causing	CF-causing	No
c.79G>T	p.Gly27X	G27X	10	0.00007		CF-causing	Yes
c.91C>T	p.Arg31Cys	R31C	23	0.00016	Non CF-causing	Non CF-causing	No
c.115C>T	p.Gln39X	Q39X	47	0.00033	CF-causing	CF-causing	No
c.137C>A	p.Ala46Asp	A46D	2	0.00001	CF-causing	CF-causing	No
c.164+1G>A	No protein name	296+1G->A	3	0.00002		CF-causing	Yes
c.165-1G>A	No protein name	297-1G->A	5	0.00004	CF-causing	CF-causing	No
c.166G>A	p.Glu56Lys	E56K	5	0.00004	CF-causing	CF-causing	No
c.170G>A or c.171G>A	p.Trp57X	W57X	6	0.00004		CF-causing	Yes
c.174_175insA	p.Arg59LysfsX10	306insA	18	0.00013	CF-causing	CF-causing	No
c.178G>T	p.Glu60X	E60X	296	0.00209	CF-causing	CF-causing	No
c.200C>T	p.Pro67Leu	P67L	239	0.00169	CF-causing	CF-causing	No
c.220C>T	p.Arg74Trp	R74W	35	0.00025	Varying clinical consequence	Varying clinical consequence	No
c.223C>T	p.Arg75X	R75X	92	0.00065	CF-causing	CF-causing	No
c.224G>A	p.Arg75Gln	R75Q	71	0.00050	Non CF-causing	Non CF-causing	No
c.233dupT	p.Trp79LeufsX32	365-366insT	7	0.00005		CF-causing	Yes
c.254G>A	p.Gly85Glu	G85E	610	0.00432	CF-causing	CF-causing	No

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c.262_263delTT	p.Leu88IlefsX22	394delTT	304	0.00215	CF-causing	CF-causing	No
c.(273+1_274-1)_ (1116+1_1117- 1)del	(No protein name)	CFTRdelete4-7	5	0.00004		CF-causing	Yes
c.(273+1_274- 1)_ (1116+1_1117- 1)del(1584+1_15 85- 1)_ (3468+1_3469- 1)del	(No protein name)	CFTR50kbdel	5	0.00004		CF-causing	Yes
c.273+1G>A	(No protein name)	405+1G->A	39	0.00028	CF-causing	CF-causing	No
c.273+3A>C	(No protein name)	405+3A->C	10	0.00007		CF-causing	Yes
c.274-1G>A	(No protein name)	406-1G->A	40	0.00028	CF-causing	CF-causing	No
c.274G>A	p.Glu92Lys	E92K	38	0.00027	CF-causing	CF-causing	No
c.274G>T	p.Glu92X	E92X	38	0.00027	CF-causing	CF-causing	No
c.292C>T	p.Gln98X	Q98X	23	0.00016	CF-causing	CF-causing	No
c.310delA	p.Arg104GlufsX3	442delA	3	0.00002		CF-causing	Yes
c.313delA	p.Ile105SerfsX2	444delA	14	0.00010	CF-causing	CF-causing	No
c.325_327delTATinsG	p.Tyr109GlyfsX4	457TAT->G	23	0.00016	CF-causing	CF-causing	No
c.349C>T	p.Arg117Cys	R117C	145	0.00103	CF-causing	CF-causing	No
c.350G>A	p.Arg117His	R117H	1852	0.01310	Varying clinical consequence	Varying clinical consequence	No
c.[350G>A;1210-12[5]]	p.[Arg117His;No protein name]	R117H;5T	102	0.00072	CF-causing	CF-causing	No
c.[350G>A;1210-12[7]]	p.[Arg117His;No protein name]	R117H;7T	125	0.00088	Varying clinical consequence	Varying clinical consequence	No
c.409delC	p.Leu137SerfsX16	541delC	10	0.00007		CF-causing	Yes
c.442delA	p.Ile148LeufsX5	574delA	27	0.00019	CF-causing	CF-causing	No
c.443T>C	p.Ile148Thr	I148T	146	0.00103	Non CF-causing	Non CF-causing	No
c.489+1G>T	(No protein name)	621+1G->T	1318	0.00932	CF-causing	CF-causing	No
c.489+3A>G	(No protein name)	621+3A->G	10	0.00007		Varying clinical consequence	Yes
c.531delT	p.Ile177MetfsX12	663delT	24	0.00017	CF-causing	CF-causing	No
c.532G>A	p.Gly178Arg	G178R	84	0.00059	CF-causing	CF-causing	No
c.543_546delTAGT	p.Leu183PhefsX5	675del4	3	0.00002	CF-causing	CF-causing	No
c.577G>T	p.Glu193X	E193X	18	0.00013		CF-causing	Yes
c.579+1G>T	(No protein name)	711+1G->T	271	0.00192	CF-causing	CF-causing	No

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c.579+3A>G	No protein name	711+3A->G	62	0.00044	CF-causing	CF-causing	No
c.579+5G>A	No protein name	711+5G->A	60	0.00042	CF-causing	CF-causing	No
c.580-1G>T	No protein name	712-1G->T	42	0.00030	CF-causing	CF-causing	No
c.595C>T	p.His199Tyr	H199Y	22	0.00016	CF-causing	CF-causing	No
c.613C>T	p.Pro205Ser	P205S	33	0.00023	CF-causing	CF-causing	No
c.617T>G	p.Leu206Trp	L206W	332	0.00235	CF-causing	CF-causing	No
c.658C>T	p.Gln220X	Q220X	70	0.00050	CF-causing	CF-causing	No
c.680T>G	p.Leu227Arg	L227R	29	0.00021	Unknown significance	CF-causing	Yes
c.720_741delAGG GAGAATGATGAT GAAGTAC	p.Gly241GlufsX13	852del22	13	0.00009	CF-causing	CF-causing	No
c.(743+1_744-1)_(1584+1_1585- No protein name	CFTRdup6b-10		16	0.00011		CF-causing	Yes
1)dup							
c.803delA	p.Asn268IlefsX17	935delA	21	0.00015		CF-causing	Yes
c.828C>A	p.Cys276X	C276X	14	0.00010	CF-causing	CF-causing	No
c.859_863delAAC TT	p.Asn287LysfsX19	991del5	13	0.00009		CF-causing	Yes
c.948delT	p.Phe316LeufsX12	1078delT	181	0.00128	CF-causing	CF-causing	No
c.987delA	p.Gly330GlufsX39	1119delA	3	0.00002		CF-causing	Yes
c.988G>T	p.Gly330X	G330X	23	0.00016	CF-causing	CF-causing	No
c.1000C>T	p.Arg334Trp	R334W	425	0.00301	CF-causing	CF-causing	No
c.1006_1007insG	p.Ile336SerfsX28	1138insG	23	0.00016		CF-causing	Yes
c.1007T>A	p.Ile336Lys	I336K	54	0.00038	CF-causing	CF-causing	No
c.1013C>T	p.Thr338Ile	T338I	52	0.00037	CF-causing	CF-causing	No
c.1021T>C	p.Ser341Pro	S341P	23	0.00016	CF-causing	CF-causing	No
c.1022_1023insTC	p.Phe342HisfsX28	1154insTC	210	0.00149	CF-causing	CF-causing	No
c.1029delC	p.Cys343X	1161delC	21	0.00015		CF-causing	Yes
c.1040G>A	p.Arg347His	R347H	194	0.00137	CF-causing	CF-causing	No
c.1040G>C	p.Arg347Pro	R347P	528	0.00374	CF-causing	CF-causing	No
c.1055G>A	p.Arg352Gln	R352Q	100	0.00071	CF-causing	CF-causing	No
c.[1075C>A;1079C>A]	p.[Gln359Lys;Thr360Lys]	Q359K/T360K	22	0.00016	Unknown significance	Unknown significance	No
c.1081delT	p.Trp361GlyfsX8	1213delT	16	0.00011	CF-causing	CF-causing	No
c.1116+1G>A	No protein name	1248+1G->A	27	0.00019	CF-causing	CF-causing	No
c.1117-1G>A	No protein name	1249-1G->A	6	0.00004		CF-causing	Yes
c.1127_1128insA	p.Gln378AlafsX4	1259insA	17	0.00012	CF-causing	CF-causing	No
c.1153_1154insTA	p.Asn386IlefsX3	1288insTA	18	0.00013	CF-causing	CF-causing	No

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c.1202G>A or c.1203G>A	p.Trp401X	W401X	13	0.00009	CF-causing	CF-causing	No
c.1209+1G>A	No protein name	1341+1G->A	11	0.00008	CF-causing	CF-causing	No
c.1210-12[5]	No protein name	5T	516	0.00365	Varying clinical consequence	Varying clinical consequence	No
c.[1210-12[5]];[1 210-34TG[11]]	No protein name	5T;TG11	35	0.00025		Varying clinical consequence	Yes
c.[1210-12[5]];[1 210-34TG[12]]	No protein name	5T;TG12	182	0.00129		Varying clinical consequence	Yes
c.[1210-12[5]];[1 210-34TG[13]]	No protein name	5T;TG13	43	0.00030		Varying clinical consequence	Yes
c.1210-12[7]	No protein name	7T	10	0.00007	Non CF-causing	Non CF-causing	No
c.1240C>T	p.Gln414X	Q414X	5	0.00004	CF-causing	CF-causing	No
c.1329_1330insAGAT	p.Ile444ArgfsX3	1461ins4	46	0.00033	CF-causing	CF-causing	No
c.1340delA	p.Lys447ArgfsX2	1471delA	25	0.00018	CF-causing	CF-causing	No
c.1364C>A	p.Ala455Glu	A455E	496	0.00351	CF-causing	CF-causing	No
c.1365_1366delG	p.Val456CysfsX25	1497delGG	6	0.00004		CF-causing	Yes
c.1393-1G>A	No protein name	1525-1G->A	70	0.00050	CF-causing	CF-causing	No
c.1393-2A>G	No protein name	1525-2A->G	8	0.00006		CF-causing	Yes
c.1397C>A or c.1397C>G	p.Ser466X	S466X	38	0.00027	CF-causing	CF-causing	No
c.1400T>C	p.Leu467Pro	L467P	38	0.00027	CF-causing	CF-causing	No
c.1408A>G	p.Met470Val	M470V	217	0.00154	Non CF-causing	Non CF-causing	No
c.1418delG	p.Gly473GlufsX54	1548delG	11	0.00008	CF-causing	CF-causing	No
c.1466C>A	p.Ser489X	S489X	58	0.00041	CF-causing	CF-causing	No
c.1475C>T	p.Ser492Phe	S492F	24	0.00017	CF-causing	CF-causing	No
c.1477_1478delCA	p.Gln493ValfsX10	1609delCA	21	0.00015		CF-causing	Yes
c.1477C>T	p.Gln493X	Q493X	292	0.00207	CF-causing	CF-causing	No
c.1519_1521delATC	p.Ile507del	I507del	647	0.00458	CF-causing	CF-causing	No
c.1521_1523delCTT	p.Phe508del	F508del	98735	0.69856	CF-causing	CF-causing	No
c.1545_1546delTA	p.Tyr515X	1677delTA	95	0.00067	CF-causing	CF-causing	No
c.1558G>T	p.Val520Phe	V520F	156	0.00110	CF-causing	CF-causing	No
c.1572C>A	p.Cys524X	C524X	8	0.00006		CF-causing	Yes
c.1573C>T	p.Gln525X	Q525X	8	0.00006	CF-causing	CF-causing	No
c.(1584+1_1585-1)_(_1679+1_1680- No protein name	CFTRdel11		4	0.00003		CF-causing	Yes
c.1585-1G>A	No protein name	1717-1G->A	1216	0.00860	CF-causing	CF-causing	No
c.1585-8G>A	No protein name	1717-8G->A	27	0.00019	CF-causing	CF-causing	No

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c.1624G>T	p.Gly542X	G542X	3594	0.02543	CF-causing	CF-causing	No
c.1645A>C or c.1647T>G	p.Ser549Arg	S549R	70	0.00050	CF-causing	CF-causing	No
c.1646G>A	p.Ser549Asn	S549N	203	0.00144	CF-causing	CF-causing	No
c.1648G>T	p.Gly550X	G550X	6	0.00004		CF-causing	Yes
c.1650delA	p.Gly551ValfsX8	1782delA	4	0.00003	CF-causing	CF-causing	No
c.1651G>A	p.Gly551Ser	G551S	19	0.00013	CF-causing	CF-causing	No
c.1652G>A	p.Gly551Asp	G551D	2984	0.02111	CF-causing	CF-causing	No
c.1654C>T	p.Gln552X	Q552X	36	0.00025	CF-causing	CF-causing	No
c.1657C>T	p.Arg553X	R553X	1323	0.00936	CF-causing	CF-causing	No
c.1673T>C	p.Leu558Ser	L558S	34	0.00024	Unknown significance	Unknown significance	No
c.1675G>A	p.Ala559Thr	A559T	91	0.00064	CF-causing	CF-causing	No
c.1679+1.6kbA>G	No protein name	1811+1.6kbA->G	71	0.00050	CF-causing	CF-causing	No
c.1679+1G>C	No protein name	1811+1G->C	17	0.00012	CF-causing	CF-causing	No
c.1679G>A	p.Arg560Lys	R560K	15	0.00011	CF-causing	CF-causing	No
c.1679G>C	p.Arg560Thr	R560T	343	0.00243	CF-causing	CF-causing	No
c.1680-1G>A	No protein name	1812-1G->A	32	0.00023	CF-causing	CF-causing	No
c.1682C>A	p.Ala561Glu	A561E	16	0.00011	CF-causing	CF-causing	No
c.1692delA	p.Asp565MetfsX7	1824delA	5	0.00004	CF-causing	CF-causing	No
c.1705T>G	p.Tyr569Asp	Y569D	45	0.00032	Unknown significance	CF-causing	Yes
c.1727G>C	p.Gly576Ala	G576A	74	0.00052	Non CF-causing	Non CF-causing	No
c.1736A>G	p.Asp579Gly	D579G	53	0.00037	Varying clinical consequence	Varying clinical consequence	No
c.1753G>T	p.Glu585X	E585X	109	0.00077	CF-causing	CF-causing	No
c.(1766+1_1767-1)_2619+1_2620-1del	No protein name	CFTRdele13,14a	4	0.00003		CF-causing	Yes
c.1766+1G>A	No protein name	1898+1G->A	421	0.00298	CF-causing	CF-causing	No
c.1766+1G->C	No protein name	1898+1G->C	6	0.00004	CF-causing	CF-causing	No
c.1766+3A>G	No protein name	1898+3A->G	27	0.00019	CF-causing	CF-causing	No
c.1792_1798delAAACTA	p.Lys598GlyfsX11	1924del7	7	0.00005		CF-causing	Yes
c.1841A>G	p.Asp614Gly	D614G	17	0.00012	Unknown significance	Varying clinical consequence	Yes
c.1923_1931del9insA	p.Ser641ArgfsX5	2055del9->A	23	0.00016	CF-causing	CF-causing	No
c.1973_1985del13insAGAAA	p.Arg658LysfsX4	2105-2117del13insAGA AA	27	0.00019	CF-causing	CF-causing	No
c.1986_1989delA	p.Thr663ArgfsX8	2118del4	16	0.00011	CF-causing	CF-causing	No
c.2002C>T	p.Arg668Cys	R668C	68	0.00048	Non CF-causing	Non CF-causing	No
c.2012delT	p.Leu671X	2143delT	94	0.00067	CF-causing	CF-causing	No

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c.2017G>T	p.Gly673X	G673X	12	0.00008		CF-causing	Yes
c.2051_2052delA AinsG	p.Lys684SerfsX38	2183AA->G or 2183delAA->G	522	0.00369	CF-causing	CF-causing	No
c.2052_2053insA	p.Gln685ThrfsX4	2184insA	324	0.00229	CF-causing	CF-causing	No
c.2052delA	p.Lys684AsnfsX38	2184delA	255	0.00180	CF-causing	CF-causing	No
c.2053_2054insC	p.Gln685ProfsX84	2185insC	4	0.00003		CF-causing	Yes
c.2053C>T	p.Gln685X	Q685X	4	0.00003		CF-causing	Yes
c.2125C>T	p.Arg709X	R709X	62	0.00044	CF-causing	CF-causing	No
c.2128A>T	p.Lys710X	K710X	50	0.00035	CF-causing	CF-causing	No
c.2175_2176insA	p.Glu726ArgfsX4	2307insA	62	0.00044	CF-causing	CF-causing	No
c.2195T>G	p.Leu732X	L732X	32	0.00023	CF-causing	CF-causing	No
c.2215delG	p.Val739TyrfsX16	2347delG	40	0.00028	CF-causing	CF-causing	No
c.2260G>A	p.Val754Met	V754M	23	0.00016	Non CF-causing	Non CF-causing	No
c.2290C>T	p.Arg764X	R764X	31	0.00022	CF-causing	CF-causing	No
c.2353C>T	p.Arg785X	R785X	32	0.00023	CF-causing	CF-causing	No
c.2374C>T	p.Arg792X	R792X	13	0.00009	CF-causing	CF-causing	No
c.2424_2425insAT	p.Ser809IlefsX13	2556insAT	4	0.00003	CF-causing	CF-causing	No
c.2453delT	p.Leu818TrpfsX3	2585delT	23	0.00016	CF-causing	CF-causing	No
c.2462_2463delGT	p.Ser821ArgfsX4	2594delGT	10	0.00007	CF-causing	CF-causing	No
c.2464G>T	p.Glu822X	E822X	21	0.00015	CF-causing	CF-causing	No
c.2490+1G>A	No protein name	2622+1G->A	85	0.00060	CF-causing	CF-causing	No
c.2491G>T	p.Glu831X	E831X	27	0.00019	CF-causing	CF-causing	No
c.2537G>A	p.Trp846X	W846X	54	0.00038	CF-causing	CF-causing	No
c.2547C>A	p.Tyr849X	Y849X	13	0.00009	CF-causing	CF-causing	No
c.2551C>T	p.Arg851X	R851X	26	0.00018	CF-causing	CF-causing	No
c.2583delT	p.Phe861LeufsX3	2711delT	34	0.00024	CF-causing	CF-causing	No
c.2589_2599delA ATTTGGTGCT	p.Ile864SerfsX28	2721del11	7	0.00005		CF-causing	Yes
c.2600_2601insA	p.Val868SerfsX28	2732insA	5	0.00004		CF-causing	Yes
c.(2619+1_2620-1)_ (3367+1_3368-1) del	No protein name	CFTRdele14b-17b	19	0.00013		CF-causing	Yes
c.2620-26A>G	No protein name	2752-26A->G	9	0.00006		Varying clinical consequence	Yes
c.2657+2_2657+3 insA	No protein name	2789+2insA	84	0.00059	Unknown significance	Unknown significance	No
c.2657+5G>A	No protein name	2789+5G->A	998	0.00706	CF-causing	CF-causing	No
c.2658-1G>C	No protein name	2790-1G->C	6	0.00004	CF-causing	CF-causing	No
c.2668C>T	p.Gln890X	Q890X	46	0.00033	CF-causing	CF-causing	No

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c.2735C>A	p.Ser912X	S912X	16	0.00011	CF-causing	CF-causing	No	
c.2737_2738insG	p.Tyr913X	2869insG	9	0.00006	CF-causing	CF-causing	No	
c.2739T>A	p.Tyr913X	Y913X	12	0.00008	CF-causing	CF-causing	No	
c.2764_2765insAG	p.Val922GlufsX2	2896insAG	6	0.00004	CF-causing	CF-causing	No	
c.2780T>C	p.Leu927Pro	L927P	30	0.00021	CF-causing	CF-causing	No	
c.2810_2811insT	p.Val938GlyfsX37	2942insT	8	0.00006		CF-causing	Yes	
c.2825delT	p.Ile942ThrfsX26	2957delT	7	0.00005		CF-causing	Yes	
c.2834C>T	p.Ser945Leu	S945L	166	0.00117	CF-causing	CF-causing	No	
c.2859_2890delA	CATTCTGTTCTTC	p.Leu953PhefsX1	2991del32	10	0.00007		CF-causing	Yes
AAGCACCTATGTC 1								
AACCC								
c.2875delG	p.Ala959HisfsX9	3007delG	40	0.00028	CF-causing	CF-causing	No	
c.2896delA	p.Thr966ArgfsX2	3028delA	3	0.00002		CF-causing	Yes	
c.2908G>C	p.Gly970Arg	G970R	12	0.00008	CF-causing	CF-causing	No	
c.(2908+1_2909-1)_ (3367+1_3368- No protein name	CFTRdel16-17b		13	0.00009		CF-causing	Yes	
1)del								
c.2930C>T	p.Ser977Phe	S977F	13	0.00009	Varying clinical consequence	Varying clinical consequence	No	
c.2988G>A	No protein name	3120G->A	85	0.00060	CF-causing	CF-causing	No	
c.(2988+1_2989-1)_ (3367+1_3368- No protein name	CFTRdel17a,17b		21	0.00015		CF-causing	Yes	
1)del								
c.(2988+1_2989-1)_ (3468+1_3469- No protein name	CFTRdel17a-18		51	0.00036		CF-causing	Yes	
1)del								
c.2988+1G>A	No protein name	3120+1G->A	471	0.00333	CF-causing	CF-causing	No	
c.2989-1G>A	No protein name	3121-1G->A	20	0.00014	CF-causing	CF-causing	No	
c.2989-2A>G	No protein name	3121-2A->G	7	0.00005		CF-causing	Yes	
c.2989-977_3367+248del	No protein name	3121-977_3499+248del	2	0.00001	CF-causing	CF-causing	No	
2515								
c.2991G>C	p.Leu997Phe	L997F	109	0.00077	Non CF-causing	Non CF-causing	No	
c.3002_3003delT	p.Val1001AspfsX4	3132delTG	4	0.00003	CF-causing	CF-causing	No	
G 5								
c.3039delC	p.Tyr1014ThrfsX9	3171delC	5	0.00004		CF-causing	Yes	
c.3080T>C	p.Ile1027Thr	I1027T	93	0.00066	Non CF-causing	Non CF-causing	No	
c.3140-26A>G	No protein name	3272-26A->G	463	0.00328	CF-causing	CF-causing	No	
c.3154T>G	p.Phe1052Val	F1052V	34	0.00024	Varying clinical consequence	Varying clinical consequence	No	

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c.3160C>G	p.His1054Asp	H1054D	11	0.00008	CF-causing	CF-causing	No
c.3181G>C	p.Gly1061Arg	G1061R	23	0.00016	CF-causing	CF-causing	No
c.3194T>C	p.Leu1065Pro	L1065P	43	0.00030	CF-causing	CF-causing	No
c.3196C>T	p.Arg1066Cys	R1066C	220	0.00156	CF-causing	CF-causing	No
c.3197G>A	p.Arg1066His	R1066H	100	0.00071	CF-causing	CF-causing	No
c.3205G>A	p.Gly1069Arg	G1069R	10	0.00007	Varying clinical consequence	Varying clinical consequence	No
c.3208C>T	p.Arg1070Trp	R1070W	36	0.00025	Varying clinical consequence	Varying clinical consequence	No
c.3209G>A	p.Arg1070Gln	R1070Q	19	0.00013	Varying clinical consequence	Varying clinical consequence	No
c.3222T>A	p.Phe1074Leu	F1074L	11	0.00008	CF-causing	Varying clinical consequence	Yes
c.3230T>C	p.Leu1077Pro	L1077P	96	0.00068	CF-causing	CF-causing	No
c.3266G>A	p.Trp1089X	W1089X	67	0.00047	CF-causing	CF-causing	No
c.3276C>A or c.3276C>G	p.Tyr1092X	Y1092X	204	0.00144	CF-causing	CF-causing	No
c.328G>C	p.Asp110His	D110H	58	0.00041	CF-causing	CF-causing	No
c.3293G>A or c.3294G>A	p.Trp1098X	W1098X	9	0.00006		CF-causing	Yes
c.3302T>A	p.Met1101Lys	M1101K	238	0.00168	CF-causing	CF-causing	No
c.3304A>T	p.Arg1102X	R1102X	5	0.00004		CF-causing	Yes
c.3310G>T	p.Glu1104X	E1104X	23	0.00016	CF-causing	CF-causing	No
c.3368-2A>G	No protein name	3500-2A->G	13	0.00009		CF-causing	Yes
c.3454G>C	p.Asp1152His	D1152H	570	0.00403	Varying clinical consequence	Varying clinical consequence	No
c.(3468+1_3469-1)_(_3717+1_3718- No protein name		CFTRdelete19	9	0.00006		CF-causing	Yes
1)del							
c.3472C>T	p.Arg1158X	R1158X	177	0.00125	CF-causing	CF-causing	No
c.3484C>T	p.Arg1162X	R1162X	649	0.00459	CF-causing	CF-causing	No
c.3485G>T	p.Arg1162Leu	R1162L	16	0.00011	Non CF-causing	Non CF-causing	No
c.3528delC	p.Lys1177SerfsX15	3659delC	536	0.00379	CF-causing	CF-causing	No
c.3535_3536insT CAA	p.Thr1179IlefsX17	3667ins4	10	0.00007	CF-causing	CF-causing	No
c.3587C>G	p.Ser1196X	S1196X	19	0.00013	CF-causing	CF-causing	No
c.3605delA	p.Asp1202AlafsX9	3737delA	5	0.00004	CF-causing	CF-causing	No
c.3611G>A or c.3612G>A	p.Trp1204X	W1204X	29	0.00021	CF-causing	CF-causing	No
c.3659delC	p.Thr1220LysfsX8	3791delC	20	0.00014	CF-causing	CF-causing	No
c.3667T>A	p.Tyr122X	Y122X	88	0.00062	CF-causing	CF-causing	No
c.3691delT	p.Ser1231ProfsX4	3821delT	5	0.00004	CF-causing	CF-causing	No
c.3700A>G	p.Ile1234Val	I1234V	33	0.00023	CF-causing	CF-causing	No
c.3705T>G	p.Ser1235Arg	S1235R	107	0.00076	Non CF-causing	Non CF-causing	No
c.3717+12191C>T	No protein name	3849+10kbC->T	1143	0.00809	CF-causing	CF-causing	No
c.3717+4A>G	No protein name	3849+4A->G	14	0.00010		CF-causing	Yes

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c.3718-1G>A	No protein name	3850-1G->A	12	0.00008	CF-causing	CF-causing	No
c.3718-3T>G	No protein name	3850-3T->G	6	0.00004		CF-causing	Yes
c.3731G>A	p.Gly1244Glu	G1244E	106	0.00075	CF-causing	CF-causing	No
c.3744delA	p.Lys1250ArgfsX9	3876delA	91	0.00064	CF-causing	CF-causing	No
c.3747delG	p.Lys1250ArgfsX9	3878delG	9	0.00006		CF-causing	Yes
c.3752G>A	p.Ser1251Asn	S1251N	120	0.00085	CF-causing	CF-causing	No
c.3761T>G	p.Leu1254X	L1254X	24	0.00017		CF-causing	Yes
c.3763T>C	p.Ser1255Pro	S1255P	10	0.00007	CF-causing	CF-causing	No
c.3764C>A	p.Ser1255X	S1255X	16	0.00011	CF-causing	CF-causing	No
c.3773_3774insT	p.Leu1258PhefsX7	3905insT	210	0.00149	CF-causing	CF-causing	No
c.3808G>A	p.Asp1270Asn	D1270N	55	0.00039	Varying clinical consequence	Varying clinical consequence	No
c.3846G>A	p.Trp1282X	W1282X	1722	0.01218	CF-causing	CF-causing	No
c.3873+1G>A	No protein name	4005+1G->A	21	0.00015	CF-causing	CF-causing	No
c.3882_3885delATT	p.Ile1295PhefsX32	4010del4	10	0.00007		CF-causing	Yes
c.3883delA	p.Ile1295PhefsX33	4015delA	5	0.00004	CF-causing	CF-causing	No
c.3884_3885insT	p.Ser1297PhefsX5	4016insT	75	0.00053	CF-causing	CF-causing	No
c.3899dupT	p.Ser1297PhefsX5	4021dupT	14	0.00010		CF-causing	Yes
c.3908delA	p.Asn1303ThrfssX25	4040delA	9	0.00006		CF-causing	Yes
c.3909C>G	p.Asn1303Lys	N1303K	2226	0.01575	CF-causing	CF-causing	No
c.3937C>T	p.Gln1313X	Q1313X	30	0.00021	CF-causing	CF-causing	No
c.(3963+1_3964-1)_(?1_?)del	No protein name	CFTRdele22-24	40	0.00028		CF-causing	Yes
c.3964-78_4242+577del	No protein name	CFTRdele22,23	40	0.00028	CF-causing	CF-causing	No
c.4046G>A	p.Gly1349Asp	G1349D	22	0.00016	CF-causing	CF-causing	No
c.4077_4080delTGTinsAA	No protein name	4209TGTT->AA	18	0.00013	CF-causing	CF-causing	No
c.4086_4087insT	p.Lys1363X	4218insT	5	0.00004		CF-causing	Yes
c.4111G>T	p.Glu1371X	E1371X	19	0.00013	CF-causing	CF-causing	No
c.4144C>T	p.Gln1382X	Q1382X	5	0.00004		CF-causing	Yes
c.4147_4148insA	p.Ile1383AsnfsX3	4279insA	5	0.00004		CF-causing	Yes
c.4196_4197delTC	p.Cys1400X	4326delTC	14	0.00010	CF-causing	CF-causing	No
c.4231C>T	p.Gln1411X	Q1411X	6	0.00004		CF-causing	Yes
c.4234C>T	p.Gln1412X	Q1412X	4	0.00003	CF-causing	CF-causing	No
c.4242+1G>T	No protein name	4374+1G->T	15	0.00011	CF-causing	CF-causing	No

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c.4251delA	p.Glu1418ArgfsX14	4382delA	63	0.00045	CF-causing	CF-causing	No
c.4296_4297insGA	p.Ser1435GlyfsX14	4428insGA	4	0.00003	CF-causing	CF-causing	No

*Represents the allele frequency within the CFTR2 database. This is subject to regional and ethnic variability of mutation distribution and may differ from the worldwide frequency.