List of CFTR2 mutations

Date: 10 April 2012

Number of patients in CFTR2: 35,312

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 70,777 identified mutations)*	Mutation final determination 10 April 2012 (current version)
c.1A>G	p.? (unknown)	M1V	9	0.00013	CF-causing
c.54-5940_273+10250del21kb	p.Ser18ArgfsX16	CFTRdele2,3	277	0.00391	CF-causing
c.91C>T	p.Arg31Cys	R31C	13	0.00018	Unknown significance
c.115C>T	p.Gln39X	Q39X	23	0.00032	CF-causing
c.178G>T	p.Glu60X	E60X	165	0.00233	CF-causing
c.200C>T	p.Pro67Leu	P67L	77	0.00109	CF-causing
c.220C>T	p.Arg74Trp	R74W	26	0.00037	Varying clinical consequence
c.223C>T	p.Arg75X	R75X	48	0.00068	CF-causing
c.224G>A	p.Arg75Gln	R75Q	28	0.00040	Non CF-causing
c.254G>A	p.Gly85Glu	G85E	316	0.00446	CF-causing
c.262_263delTT	p.Leu88IlefsX22	394delTT	154	0.00218	CF-causing
c.273+1G>A	No protein name	405+1G->A	20	0.00028	CF-causing
c.274-1G>A	No protein name	406-1G->A	21	0.00030	CF-causing
c.274G>A	p.Glu92Lys	E92K	14	0.00020	CF-causing
c.274G>T	p.Glu92X	E92X	22	0.00031	CF-causing
c.292C>T	p.Gln98X	Q98X	13	0.00018	CF-causing
c.325_327delTATinsG	p.Tyr109GlyfsX4	457TAT->G	10	0.00014	CF-causing
c.328G>C	p.Asp110His	D110H	33	0.00047	CF-causing
c.349C>T	p.Arg117Cys	R117C	67	0.00095	CF-causing
c.350G>A	p.Arg117His	R117H	808	0.01142	Varying clinical consequence
c.366T>A	p.Tyr122X	Y122X	78	0.00110	CF-causing
c.442delA	p.Ile148LeufsX5	574delA	20	0.00028	CF-causing
c.443T>C	p.Ile148Thr	I148T	99	0.00140	Non CF-causing
c.489+1G>T	No protein name	621+1G->T	817	0.01154	CF-causing
c.531delT	p.Ile177MetfsX12	663delT	12	0.00017	CF-causing
c.532G>A	p.Gly178Glu	G178R	50	0.00071	CF-causing
c.579+1G>T	No protein name	711+1G->T	166	0.00235	CF-causing
c.579+3A>G	No protein name	711+3A->G	22	0.00031	Unknown significance
c.579+5G>A	No protein name	711+5G->A	47	0.00066	Unknown significance
c.580-1G>T	No protein name	712-1G->T	16	0.00023	CF-causing
c.595C>T	p.His199Tyr	H199Y	11	0.00016	Unknown significance

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c.613C>T	p.Pro205Ser	P205S	12	0.00017	Unknown significance
c.617T>G	p.Leu206Trp	L206W	136	0.00192	CF-causing
c.658C>T	p.Gln220X	Q220X	35	0.00049	CF-causing
c.680T>G	p.Leu227Arg	L227R	15	0.00021	Unknown significance
c.720_741delAGGGAGAATGATGATGAAGTAC	p.Gly241GlufsX13	852del22	11	0.00016	CF-causing
c.948delT	p.Phe316LeufsX12	1078delT	105	0.00148	CF-causing
c.988G>T	p.Gly330X	G330X	12	0.00017	CF-causing
c.1000C>T	p.Arg334Trp	R334W	174	0.00246	CF-causing
c.1007T>A	p.Ile336Lys	I336K	29	0.00041	CF-causing
c.1013C>T	p.Thr338Ile	T338I	55	0.00078	CF-causing
c.1021T>C	p.Ser341Pro	S341P	9	0.00013	CF-causing
c.1022_1023insTC	p.Phe342HisfsX28	1154insTC	101	0.00143	CF-causing
c.1040G>A	p.Arg347His	R347H	91	0.00129	CF-causing
c.1040G>C	p.Arg347Pro	R347P	234	0.00331	CF-causing
c.1055G>A	p.Arg352Gln	R352Q	49	0.00069	CF-causing
c.[1075C>A;1079C>A]	p.[Gln359Lys;Thr360Lys]	Q359K/T360K	14	0.00020	Unknown significance
c.1081delT	p.Trp361GlyfsX8	1213delT	9	0.00013	CF-causing
c.1116+1G>A	No protein name	1248+1G->A	13	0.00018	CF-causing
c.1127_1128insA	p.Gln378AlafsX4	1259insA	11	0.00016	CF-causing
c.1202G>A or c.1203G>A	p.Trp401X	W401X	14	0.00020	CF-causing
c.1209+1G>A	No protein name	1341+1G->A	9	0.00013	CF-causing
c.1210−12[5]	No protein name	5T	164	0.00232	Varying clinical consequence
c.1210−12[7]	No protein name	7T	11	0.00016	Non CF-causing
c.1329_1330insAGAT	p.Ile444ArgfsX3	1461ins4	16	0.00023	CF-causing
c.1364C>A	p.Ala455Glu	A455E	219	0.00309	CF-causing
c.1393-1G>A	No protein name	1525-1G->A	25	0.00035	CF-causing
c.1397C>A or c.1397C>G	p.Ser466X	S466X	20	0.00028	CF-causing
c.1400T>C	p.Leu467Pro	L467P	16	0.00023	CF-causing
c.1408A>G	p.Met470Val	M470V	41	0.00058	Non CF-causing
c.1418delG	p.Gly473GlufsX54	1548delG	9	0.00013	CF-causing
c.1466C>A	p.Ser489X	S489X	27	0.00038	CF-causing
c.1475C>T	p.Ser492Phe	S492F	16	0.00023	CF-causing
c.1477C>T	p.Gln493X	Q493X	168	0.00237	CF-causing
c.1519_1521delATC	p.Ile507del	I507del	319	0.00451	CF-causing
c.1521_1523delCTT	p.Phe508del	F508del	49740	0.70277	CF-causing
c.1545_1546delTA	p.Tyr515X	1677delTA	51	0.00072	CF-causing
c.1558G>T	p.Val520Phe	V520F	73	0.00103	CF-causing
c.1573C>T	p.Gln525X	Q525X	11	0.00016	CF-causing
c.1585-8G>A	No protein name	1717-8G->A	9	0.00013	Unknown significance

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c.1585-1G>A	No protein name	1717-1G->A	635	0.00897	CF-causing
c.1624G>T	p.Gly542X	G542X	1856	0.02622	CF-causing
c.1645A>C or c.1647T>G	p.Ser549Arg	S549R	48	0.00068	CF-causing
c.1646G>A	p.Ser549Asn	S549N	91	0.00129	CF-causing
c.1652G>A	p.Gly551Asp	G551D	1427	0.02016	CF-causing
c.1654C>T	p.Gln552X	Q552X	28	0.00040	CF-causing
c.1657C>T	p.Arg553X	R553X	645	0.00911	CF-causing
c.1673T>C	p.Leu558Ser	L558S	15	0.00021	Unknown significance
c.1675G>A	p.Ala559Thr	A559T	46	0.00065	CF-causing
c.1679+1.6kbA>G	No protein name	1811+1.6kbA->G	36	0.00051	CF-causing
c.1679G>A	p.Arg560Lys	R560K	9	0.00013	CF-causing
c.1679G>C	p.Arg560Thr	R560T	198	0.00280	CF-causing
c.1680-1G>A	No protein name	1812-1G->A	19	0.00027	CF-causing
c.1705T>G	p.Tyr569Asp	Y569D	11	0.00016	Unknown significance
c.1727G>C	p.Gly576Ala	G576A	42	0.00059	Varying clinical consequence
c.1736A>G	p.Asp579Gly	D579G	24	0.00034	Varying clinical consequence
c.1753G>T	p.Glu585X	E585X	41	0.00058	CF-causing
c.1766+1G>A	No protein name	1898+1G->A	245	0.00346	CF-causing
c.1766+3A>G	No protein name	1898+3A->G	11	0.00016	Unknown significance
c.1841A>G	p.Asp614Gly	D614G	11	0.00016	Unknown significance
c.2002C>T	p.Arg668Cys	R668C	49	0.00069	Varying clinical consequence
c.2012delT	p.Leu671X	2143delT	77	0.00109	CF-causing
c.2051_2052delAAinsG	p.Lys684SerfsX38	2183AA->G	292	0.00413	CF-causing
c.2051_2052delAAinsG	p.Lys684SerfsX38	2183delAA->G	85	0.00120	CF-causing
c.2052_2053insA	p.Gln685ThrfsX4	2184insA	135	0.00191	CF-causing
c.2052delA	p.Lys684AsnfsX38	2184delA	119	0.00168	CF-causing
c.2125C>T	p.Arg709X	R709X	24	0.00034	CF-causing
c.2128A>T	p.Lys710X	K710X	26	0.00037	CF-causing
c.2175_2176insA	p.Glu726ArgfsX4	2307insA	31	0.00044	CF-causing
c.2195T>G	p.Leu732X	L732X	15	0.00021	CF-causing
c.2215delG	p.Val739TyrfsX16	2347delG	29	0.00041	CF-causing
c.2260G>A	p.Val754Met	V754M	9	0.00013	Non CF-causing
c.2290C>T	p.Arg764X	R764X	15	0.00021	CF-causing
c.2453delT	p.Leu818TrpfsX3	2585delT	11	0.00016	CF-causing
c.2464G>T	p.Glu822X	E822X	25	0.00035	CF-causing
c.2490+1G>A	No protein name	2622+1G->A	29	0.00041	CF-causing
c.2491G>T	p.Glu831X	E831X	21	0.00030	CF-causing
c.2537G>A or c.2538G>A	p.Trp846X	W846X	37	0.00052	CF-causing
c.2551C>T	p.Arg851X	R851X	15	0.00021	CF-causing

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c.2583delT	p.Phe861LeufsX3	2711delT	19	0.00027	CF-causing
c.2657+2_2657+3insA	No protein name	2789+2insA	25	0.00035	Unknown significance
c.2657+5G>A	No protein name	2789+5G->A	538	0.00760	CF-causing
c.2668C>T	p.Gln890X	Q890X	21	0.00030	CF-causing
c.2780T>C	p.Leu927Pro	L927P	15	0.00021	CF-causing
c.2834C>T	p.Ser945Leu	S945L	63	0.00089	CF-causing
c.2875delG	p.Ala959HisfsX9	3007delG	28	0.00040	CF-causing
c.2908G>C	p.Gly970Arg	G970R	9	0.00013	CF-causing
c.2930C>T	p.Ser977Phe	S977F	9	0.00013	Varying clinical consequence
c.2988G>A	No protein name	3120G->A	40	0.00057	Unknown significance
c.2988+1G>A	No protein name	3120+1G->A	266	0.00376	CF-causing
c.2989-1G>A	No protein name	3121-1G->A	9	0.00013	CF-causing
c.2991G>C	p.Leu997Phe	L997F	28	0.00040	Varying clinical consequence
c.3080T>C	p.Ile1027Thr	I1027T	51	0.00072	Non CF-causing
c.3140-26A>G	No protein name	3272-26A->G	188	0.00266	CF-causing
c.3154T>G	p.Phe1052Val	F1052V	13	0.00018	Varying clinical consequence
c.3194T>C	p.Leu1065Pro	L1065P	25	0.00035	CF-causing
c.3196C>T	p.Arg1066Cys	R1066C	122	0.00172	CF-causing
c.3197G>A	p.Arg1066His	R1066H	30	0.00042	CF-causing
c.3205G>A	p.Gly1069Arg	G1069R	9	0.00013	Varying clinical consequence
c.3208C>T	p.Arg1070Trp	R1070W	13	0.00018	Varying clinical consequence
c.3209G>A	p.Arg1070Gln	R1070Q	21	0.00030	Varying clinical consequence
c.3230T>C	p.Leu1077Pro	L1077P	48	0.00068	CF-causing
c.3266G>A	p.Trp1089X	W1089X	42	0.00059	CF-causing
c.3276C>A or c.3276C>G	p.Tyr1092X	Y1092X	143	0.00202	CF-causing
c.3302T>A	p.Met1101Lys	M1101K	152	0.00215	CF-causing
c.3310G>T	p.Glu1104X	E1104X	13	0.00018	CF-causing
c.3454G>C	p.Asp1152His	D1152H	196	0.00277	Varying clinical consequence
c.3472C>T	p.Arg1158X	R1158X	93	0.00131	CF-causing
c.3484C>T	p.Arg1162X	R1162X	346	0.00489	CF-causing
c.3485G>T	p.Arg1162Leu	R1162L	9	0.00013	Unknown significance
c.3528delC	p.Lys1177SerfsX15	3659delC	248	0.00350	CF-causing
c.3587C>G	p.Ser1196X	S1196X	14	0.00020	CF-causing
c.3611G>A or c.3612G>A	p.Trp1204X	W1204X	14	0.00020	CF-causing
c.3659delC	p.Thr1220LysfsX8	3791delC	15	0.00021	CF-causing
c.3700A>G	p.Ile1234Val	I1234V	18	0.00025	Varying clinical consequence

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c.3705T>G	p.Ser1235Arg	S1235R	54	0.00076	Non CF-causing
c.3717+12191C>T	No protein name	3849+10kbC->T	524	0.00740	CF-causing
c.3731G>A	p.Gly1244Glu	G1244E	49	0.00069	CF-causing
c.3744delA	p.Lys1250ArgfsX9	3876delA	35	0.00049	CF-causing
c.3752G>A	p.Ser1251Asn	S1251N	85	0.00120	CF-causing
c.3773_3774insT	p.Leu1258PhefsX7	3905insT	135	0.00191	CF-causing
c.3808G>A	p.Asp1270Asn	D1270N	32	0.00045	Varying clinical consequence
c.3846G>A	p.Trp1282X	W1282X	1056	0.01492	CF-causing
c.3873+1G>A	No protein name	4005+1G->A	20	0.00028	CF-causing
c.3884_3885insT	p.Ser1297PhefsX5	4016insT	37	0.00052	CF-causing
c.3909C>G	p.Asn1303Lys	N1303K	1242	0.01755	CF-causing
c.3937C>T	p.Gln1313X	Q1313X	26	0.00037	CF-causing
c.3964-78_4242+577del	NULL	CFTRdele22,23	17	0.00024	CF-causing
c.4077_4080delTGTTinsAA	No protein name	4209TGTT->AA	9	0.00013	CF-causing
c.4251delA	p.Glu1418ArgfsX14	4382delA	29	0.00041	CF-causing

*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.