List of CFTR2 mutations

Date: 27 February 2015

Number of patients in CFTR2: 39,696

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 22 July 2013 (previous version)	Mutation final determination 20 Feb 2015 (current version)	Change from previous version?
c.1A>G	p.? (unknown)	M1V	9	0.00013	CF-causing	CF-causing	No
c.54-5940_273+10250del21kb	p.Ser18ArgfsX16	CFTRdele2,3	277	0.00391	CF-causing	CF-causing	No
c.91C>T	p.Arg31Cys	R31C	13	0.00018	Non CF-causing	Non CF-causing	No
c.115C>T	p.Gln39X	Q39X	23	0.00032	CF-causing	CF-causing	No
c.137C>A	p.Ala46Asp	A46D	6	0.00008	CF-causing	CF-causing	No
c.165-1G>A	No protein name	297-1G->A	3	0.00004	CF-causing	CF-causing	No
c.166G>A	p.Glu56Lys	E56K	5	0.00007	CF-causing	CF-causing	No
c.174_175insA	p.Arg59LysfsX10	306insA	6	0.00008	CF-causing	CF-causing	No
c.178G>T	p.Glu60X	E60X	165	0.00233	CF-causing	CF-causing	No
c.200C>T	p.Pro67Leu	P67L	77	0.00109	CF-causing	CF-causing	No
c.220C>T	p.Arg74Trp	R74W	26	0.00037	Varying clinical consequence	Varying clinical consequence	No
c.223C>T	p.Arg75X	R75X	48	0.00068	CF-causing	CF-causing	No
c.224G>A	p.Arg75Gln	R75Q	28	0.00040	Non CF-causing	Non CF-causing	No
c.254G>A	p.Gly85Glu	G85E	316	0.00446	CF-causing	CF-causing	No
c.262_263delTT	p.Leu88IlefsX22	394delTT	154	0.00218	CF-causing	CF-causing	No
c.273+1G>A	No protein name	405+1G->A	20	0.00028	CF-causing	CF-causing	No
c.274-1G>A	No protein name	406-1G->A	21	0.00030	CF-causing	CF-causing	No
c.274G>A	p.Glu92Lys	E92K	14	0.00020	CF-causing	CF-causing	No
c.274G>T	p.Glu92X	E92X	22	0.00031	CF-causing	CF-causing	No
c.292C>T	p.Gln98X	Q98X	13	0.00018	CF-causing	CF-causing	No
c.313delA	p.Ile105SerfsX2	444delA	7	0.00010	CF-causing	CF-causing	No
c.325_327delTATinsG	p.Tyr109GlyfsX4	457TAT->G	10	0.00014	CF-causing	CF-causing	No
c.328G>C	p.Asp110His	D110H	33	0.00047	CF-causing	CF-causing	No
c.349C>T	p.Arg117Cys	R117C	67	0.00095	CF-causing	CF-causing	No
c.350G>A	p.Arg117His	R117H	808	0.01142	Varying clinical consequence	Varying clinical consequence	No
c.[350G>A;1210-12[5]]	p.[Arg117His;No protein name]	R117H with 5T	NA	NA		CF-causing	Yes
c.[350G>A;1210-12[7]]	p.[Arg117His;No protein name]	R117H with 7T	NA	NA		Varying clinical consequence	Yes
c.366T>A	p.Tyr122X	Y122X	78	0.00110	CF-causing	CF-causing	No
c.442delA	p.Ile148LeufsX5	574delA	20	0.00028	CF-causing	CF-causing	No
c.443T>C	p.Ile148Thr	I148T	99	0.00140	Non CF-causing	Non CF-causing	No
c.489+1G>T	No protein name	621+1G->T	817	0.01154	CF-causing	CF-causing	No
c.531delT	p.Ile177MetfsX12	663delT	12	0.00017	CF-causing	CF-causing	No
c.532G>A	p.Gly178Glu	G178R	50	0.00071	CF-causing	CF-causing	No
c.543_546delTAGT	p.Leu183PhefsX5	675del4	3	0.00004	CF-causing	CF-causing	No
c.579+1G>T	No protein name	711+1G->T	166	0.00235	CF-causing	CF-causing	No

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c.579+3A>G	No protein name	711+3A->G	22	0.00031	CF-causing	CF-causing	No
c.579+5G>A	No protein name	711+5G->A	47	0.00066	CF-causing	CF-causing	No
c.580-1G>T	No protein name	712-1G->T	16	0.00023	CF-causing	CF-causing	No
c.595C>T	p.His199Tyr	H199Y	11	0.00016	CF-causing	CF-causing	No
c.613C>T	p.Pro205Ser	P205S	12	0.00017	CF-causing	CF-causing	No
c.617T>G	p.Leu206Trp	L206W	136	0.00192	CF-causing	CF-causing	No
c.658C>T	p.Gln220X	Q220X	35	0.00049	CF-causing	CF-causing	No
c.680T>G	p.Leu227Arg	L227R	15	0.00021	Unknown significance	Unknown significance	No
c.720_741delAGGGAGAATGATGATGAAGTAC	p.Gly241GlufsX13	852del22	11	0.00016	CF-causing	CF-causing	No
c.828C>A	p.Cys276X	C276X	7	0.00010	CF-causing	CF-causing	No
c.948delT	p.Phe316LeufsX12	1078delT	105	0.00148	CF-causing	CF-causing	No
c.988G>T	p.Gly330X	G330X	12	0.00017	CF-causing	CF-causing	No
c.1000C>T	p.Arg334Trp	R334W	174	0.00246	CF-causing	CF-causing	No
c.1007T>A	p.Ile336Lys	I336K	29	0.00041	CF-causing	CF-causing	No
c.1013C>T	p.Thr338Ile	T338I	55	0.00078	CF-causing	CF-causing	No
c.1021T>C	p.Ser341Pro	S341P	9	0.00013	CF-causing	CF-causing	No
c.1022_1023insTC	p.Phe342HisfsX28	1154insTC	101	0.00143	CF-causing	CF-causing	No
c.1040G>A	p.Arg347His	R347H	91	0.00129	CF-causing	CF-causing	No
c.1040G>C	p.Arg347Pro	R347P	234	0.00331	CF-causing	CF-causing	No
c.1055G>A	p.Arg352Gln	R352Q	49	0.00069	CF-causing	CF-causing	No
c.[1075C>A;1079C>A]	p.[Gln359Lys;Thr360Lys]	Q359K/T360K	14	0.00020	Unknown significance	Unknown significance	No
c.1081delT	p.Trp361GlyfsX8	1213delT	9	0.00013	CF-causing	CF-causing	No
c.1116+1G>A	No protein name	1248+1G->A	13	0.00018	CF-causing	CF-causing	No
c.1127_1128insA	p.Gln378AlafsX4	1259insA	11	0.00016	CF-causing	CF-causing	No
c.1153_1154insAT	p.Asn386IlefsX3	1288insTA	7	0.00010	CF-causing	CF-causing	No
c.1202G>A or c.1203G>A	p.Trp401X	W401X	14	0.00020	CF-causing	CF-causing	No
c.1209+1G>A	No protein name	1341+1G->A	9	0.00013	CF-causing	CF-causing	No
c.1210−12[5]	No protein name	5T	164	0.00232	Varying clinical consequence	Varying clinical consequence	No
c.1210−12[7]	No protein name	7T	11	0.00016	Non CF-causing	Non CF-causing	No
c.1240C>T	p.Gln414X	Q414X	5	0.00007	CF-causing	CF-causing	No
c.1329_1330insAGAT	p.Ile444ArgfsX3	1461ins4	16	0.00023	CF-causing	CF-causing	No
c.1340delA	p.Lys447ArgfsX2	1471delA	7	0.00010	CF-causing	CF-causing	No
c.1364C>A	p.Ala455Glu	A455E	219	0.00309	CF-causing	CF-causing	No
c.1393-1G>A	No protein name	1525-1G->A	25	0.00035	CF-causing	CF-causing	No
c.1397C>A or c.1397C>G	p.Ser466X	S466X	20	0.00028	CF-causing	CF-causing	No
c.1400T>C	p.Leu467Pro	L467P	16	0.00023	CF-causing	CF-causing	No
c.1408A>G	p.Met470Val	M470V	41	0.00058	Non CF-causing	Non CF-causing	No
c.1418delG	p.Gly473GlufsX54	1548delG	9	0.00013	CF-causing	CF-causing	No
c.1466C>A	p.Ser489X	S489X	27	0.00038	CF-causing	CF-causing	No
c.1475C>T	p.Ser492Phe	S492F	16	0.00023	CF-causing	CF-causing	No
c.1477C>T	p.Gln493X	Q493X	168	0.00237	CF-causing	CF-causing	No
c.1519_1521delATC	p.Ile507del	I507del	319	0.00451	CF-causing	CF-causing	No
c.1521_1523delCTT	p.Phe508del	F508del	49740	0.70277	CF-causing	CF-causing	No

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c.1545_1546delTA	p.Tyr515X	1677delTA	51	0.00072	CF-causing	CF-causing	No
c.1558G>T	p.Val520Phe	V520F	73	0.00103	CF-causing	CF-causing	No
c.1573C>T	p.Gln525X	Q525X	11	0.00016	CF-causing	CF-causing	No
c.1585-8G>A	No protein name	1717-8G->A	9	0.00013	CF-causing	CF-causing	No
c.1585-1G>A	No protein name	1717-1G->A	635	0.00897	CF-causing	CF-causing	No
c.1624G>T	p.Gly542X	G542X	1856	0.02622	CF-causing	CF-causing	No
c.1645A>C or c.1647T>G	p.Ser549Arg	S549R	48	0.00068	CF-causing	CF-causing	No
c.1646G>A	p.Ser549Asn	S549N	91	0.00129	CF-causing	CF-causing	No
c.1650delA	p.Gly551ValfsX8	1782delA	5	0.00007	CF-causing	CF-causing	No
c.1651G>A	p.Gly551Ser	G551S	8	0.00011	CF-causing	CF-causing	No
c.1652G>A	p.Gly551Asp	G551D	1427	0.02016	CF-causing	CF-causing	No
c.1654C>T	p.Gln552X	Q552X	28	0.00040	CF-causing	CF-causing	No
c.1657C>T	p.Arg553X	R553X	645	0.00911	CF-causing	CF-causing	No
c.1673T>C	p.Leu558Ser	L558S	15	0.00021	Unknown significance	Unknown significance	No
c.1675G>A	p.Ala559Thr	A559T	46	0.00065	CF-causing	CF-causing	No
c.1679G>A	p.Arg560Lys	R560K	9	0.00013	CF-causing	CF-causing	No
c.1679G>C	p.Arg560Thr	R560T	198	0.00280	CF-causing	CF-causing	No
c.1679+1G>C	No protein name	1811+1G->C	6	0.00008	CF-causing	CF-causing	No
c.1679+1.6kbA>G	No protein name	1811+1.6kbA->G	36	0.00051	CF-causing	CF-causing	No
c.1680-1G>A	No protein name	1812-1G->A	19	0.00027	CF-causing	CF-causing	No
c.1682C>A	p.Ala561Glu	A561E	8	0.00011	CF-causing	CF-causing	No
c.1692delA	p.Asp565MetfsX7	1824delA	3	0.00004	CF-causing	CF-causing	No
c.1705T>G	p.Tyr569Asp	Y569D[§]	11	0.00016	Unknown significance	Unknown significance	No
c.1727G>C	p.Gly576Ala	G576A	42	0.00059	Non CF-causing	Non CF-causing	No
c.1736A>G	p.Asp579Gly	D579G	24	0.00034	Varying clinical consequence	Varying clinical consequence	No
c.1753G>T	p.Glu585X	E585X	41	0.00058	CF-causing	CF-causing	No
c.1766+1G>A	No protein name	1898+1G->A	245	0.00346	CF-causing	CF-causing	No
c.1766+1G>C	No protein name	1898+1G->C	3	0.00004	CF-causing	CF-causing	No
c.1766+3A>G	No protein name	1898+3A->G	11	0.00016	CF-causing	CF-causing	No
c.1841A>G	p.Asp614Gly	D614G	11	0.00016	Unknown significance	Unknown significance	No
c.1923_1931del9insA	p.Ser641ArgfsX5	2055del9->A	8	0.00011	CF-causing	CF-causing	No
c.1973_1985del13insAGAAA	p.Arg658LysfsX4	2105-2117del13insAGAAA	8	0.00011	CF-causing	CF-causing	No
c.1986_1989delAACT	p.Thr663ArgfsX8	2118del4	5	0.00007	CF-causing	CF-causing	No
c.2002C>T	p.Arg668Cys	R668C	49	0.00069	Non CF-causing	Non CF-causing	No
c.2012delT	p.Leu671X	2143delT	77	0.00109	CF-causing	CF-causing	No
c.2051_2052delAAinsG	p.Lys684SerfsX38	2183AA->G[‡]	292	0.00413	CF-causing	CF-causing	No
c.2051_2052delAAinsG	p.Lys684SerfsX38	2183delAA->G[‡]	85	0.00120	CF-causing	CF-causing	No

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c.2052_2053insA	p.Gln685ThrfsX4	2184insA	135	0.00191	CF-causing	CF-causing	No
c.2052delA	p.Lys684AsnfsX38	2184delA	119	0.00168	CF-causing	CF-causing	No
c.2125C>T	p.Arg709X	R709X	24	0.00034	CF-causing	CF-causing	No
c.2128A>T	p.Lys710X	K710X	26	0.00037	CF-causing	CF-causing	No
c.2175_2176insA	p.Glu726ArgfsX4	2307insA	31	0.00044	CF-causing	CF-causing	No
c.2195T>G	p.Leu732X	L732X	15	0.00021	CF-causing	CF-causing	No
c.2215delG	p.Val739TyrfsX16	2347delG	29	0.00041	CF-causing	CF-causing	No
c.2260G>A	p.Val754Met	V754M	9	0.00013	Non CF-causing	Non CF-causing	No
c.2290C>T	p.Arg764X	R764X	15	0.00021	CF-causing	CF-causing	No
c.2353C>T	p.Arg785X	R785X	9	0.00013	CF-causing	CF-causing	No
c.2374C>T	p.Arg792X	R792X	7	0.00010	CF-causing	CF-causing	No
c.2424_2425insAT	p.Ser809IlefsX13	2556insAT	3	0.00004	CF-causing	CF-causing	No
c.2453delT	p.Leu818TrpfsX3	2585delT	11	0.00016	CF-causing	CF-causing	No
c.2462_2463delGT	p.Ser821ArgfsX4	No legacy name	10	0.00014	CF-causing	CF-causing	No
c.2464G>T	p.Glu822X	E822X	25	0.00035	CF-causing	CF-causing	No
c.2490+1G>A	No protein name	2622+1G->A	29	0.00041	CF-causing	CF-causing	No
c.2491G>T	p.Glu831X	E831X	21	0.00030	CF-causing	CF-causing	No
c.2537G>A or c.2538G>A	p.Trp846X	W846X	37	0.00052	CF-causing	CF-causing	No
c.2547C>A	p.Tyr849X	Y849X	4	0.00006	CF-causing	CF-causing	No
c.2551C>T	p.Arg851X	R851X	15	0.00021	CF-causing	CF-causing	No
c.2583delT	p.Phe861LeufsX3	2711delT	19	0.00027	CF-causing	CF-causing	No
c.2657+2_2657+3insA	No protein name	2789+2insA	25	0.00035	Unknown significance	Unknown significance	No
c.2657+5G>A	No protein name	2789+5G->A	538	0.00760	CF-causing	CF-causing	No
c.2658-1G>C	No protein name	2790-1G->C	3	0.00004	CF-causing	CF-causing	No
c.2668C>T	p.Gln890X	Q890X	21	0.00030	CF-causing	CF-causing	No
c.2735C>A	p.Ser912X	S912X	7	0.00010	CF-causing	CF-causing	No
c.2737_2738insG		2869insG	5	0.00007	CF-causing	CF-causing	No
c.2739T>A	p.Tyr913X	Y913X	3	0.00004	CF-causing	CF-causing	No
c.2764_2765insAG	p.Val922GlufsX2	2896insAG	5	0.00007	CF-causing	CF-causing	No
c.2780T>C	p.Leu927Pro	L927P	15	0.00021	CF-causing	CF-causing	No
c.2834C>T	p.Ser945Leu	S945L	63	0.00089	CF-causing	CF-causing	No
c.2875delG	p.Ala959HisfsX9	3007delG	28	0.00040	CF-causing	CF-causing	No
c.2908G>C	p.Gly970Arg	G970R	9	0.00013	CF-causing	CF-causing	No
c.2930C>T	p.Ser977Phe	S977F	9	0.00013	Varying clinical consequence	Varying clinical consequence	No
c.2988G>A	No protein name	3120G->A	40	0.00057	CF-causing	CF-causing	No
c.2988+1G>A	No protein name	3120+1G->A	266	0.00376	CF-causing	CF-causing	No
c.2989-977_3367+248del	No protein name	3121-977_3499+248del2515	3	0.00004	CF-causing	CF-causing	No
c.2989-1G>A	No protein name	3121-1G->A	9	0.00013	CF-causing	CF-causing	No
c.2991G>C	p.Leu997Phe	L997F	28	0.00040	Non CF-causing	Non CF-causing	No
c.3002_3003delTG	p.Val1001AspfsX45	3132delTG	5	0.00007	CF-causing	CF-causing	No
c.3080T>C	p.Ile1027Thr	I1027T	51	0.00072	Non CF-causing	Non CF-causing	No
c.3140-26A>G	No protein name	3272-26A->G	188	0.00266	CF-causing	CF-causing	No

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c.3154T>G	p.Phe1052Val	F1052V	13	0.00018	Varying clinical consequence	Varying clinical consequence	No
c.3160C>G	p.His1054Asp	H1054D	8	0.00011	CF-causing	CF-causing	No
c.3181G>C	p.Gly1061Arg	G1061R	5	0.00007	CF-causing	CF-causing	No
c.3194T>C	p.Leu1065Pro	L1065P	25	0.00035	CF-causing	CF-causing	No
c.3196C>T	p.Arg1066Cys	R1066C	122	0.00172	CF-causing	CF-causing	No
c.3197G>A	p.Arg1066His	R1066H	30	0.00042	CF-causing	CF-causing	No
c.3205G>A	p.Gly1069Arg	G1069R	9	0.00013	Varying clinical consequence	Varying clinical consequence	No
c.3208C>T	p.Arg1070Trp	R1070W	13	0.00018	Varying clinical consequence	Varying clinical consequence	No
c.3209G>A	p.Arg1070Gln	R1070Q	21	0.00030	Varying clinical consequence	Varying clinical consequence	No
c.3222T>A	p.Phe1074Leu	F1074L	5	0.00007	CF-causing	CF-causing	No
c.3230T>C	p.Leu1077Pro	L1077P	48	0.00068	CF-causing	CF-causing	No
c.3266G>A	p.Trp1089X	W1089X	42	0.00059	CF-causing	CF-causing	No
c.3276C>A or c.3276C>G	p.Tyr1092X	Y1092X	143	0.00202	CF-causing	CF-causing	No
c.3302T>A	p.Met1101Lys	M1101K	152	0.00215	CF-causing	CF-causing	No
c.3310G>T	p.Glu1104X	E1104X	13	0.00018	CF-causing	CF-causing	No
c.3454G>C	p.Asp1152His	D1152H	196	0.00277	Varying clinical consequence	Varying clinical consequence	No
c.3472C>T	p.Arg1158X	R1158X	93	0.00131	CF-causing	CF-causing	No
c.3484C>T	p.Arg1162X	R1162X	346	0.00489	CF-causing	CF-causing	No
c.3485G>T	p.Arg1162Leu	R1162L	9	0.00013	Non CF-causing	Non CF-causing	No
c.3528delC	p.Lys1177SerfsX15	3659delC	248	0.00350	CF-causing	CF-causing	No
c.3535_3536insTCAA	p.Thr1179IlefsX17	3667ins4	7	0.00010	CF-causing	CF-causing	No
c.3587C>G	p.Ser1196X	S1196X	14	0.00020	CF-causing	CF-causing	No
c.3605delA	p.Asp1202AlafsX9	3737delA	5	0.00007	CF-causing	CF-causing	No
c.3611G>A or c.3612G>A	p.Trp1204X	W1204X	14	0.00020	CF-causing	CF-causing	No
c.3659delC	p.Thr1220LysfsX8	3791delC	15	0.00021	CF-causing	CF-causing	No
c.3691delT	p.Ser1231ProfsX4	3821delT	7	0.00010	CF-causing	CF-causing	No
c.3700A>G	p.Ile1234Val	I1234V	18	0.00025	Unknown significance	CF-causing	Yes
c.3705T>G	p.Ser1235Arg	S1235R	54	0.00076	Non CF-causing	Non CF-causing	No
c.3717+12191C>T	No protein name	3849+10kbC->T	524	0.00740	CF-causing	CF-causing	No
c.3718-1G>A	No protein name	3850-1G->A	6	0.00008	CF-causing	CF-causing	No
c.3731G>A	p.Gly1244Glu	G1244E	49	0.00069	CF-causing	CF-causing	No
c.3744delA	p.Lys1250ArgfsX9	3876delA	35	0.00049	CF-causing	CF-causing	No
c.3752G>A	p.Ser1251Asn	S1251N	85	0.00120	CF-causing	CF-causing	No
c.3763T>C	p.Ser1255Pro	S1255P	3	0.00004	CF-causing	CF-causing	No
c.3764C>A	p.Ser1255X	S1255X	5	0.00007	CF-causing	CF-causing	No
c.3773_3774insT	p.Leu1258PhefsX7	3905insT	135	0.00191	CF-causing	CF-causing	No
c.3808G>A	p.Asp1270Asn	D1270N	32	0.00045	Varying clinical consequence	Varying clinical consequence	No
c.3846G>A	p.Trp1282X	W1282X	1056	0.01492	CF-causing	CF-causing	No
c.3873+1G>A	No protein name	4005+1G->A	20	0.00028	CF-causing	CF-causing	No

Mutation cDNA name	Mutation protein name	Mutation legacy name	# alleles in CFTR2	Allele frequency in CFTR2 (of 70,777 identified mutations)*	Mutation final determination 22 July 2013 (current version)	Mutation final determination 20 Feb 2015 (current version)	Change from previous version?
c.3883delA	p.Ile1295PhefsX33	4015delA	3	0.00004	CF-causing	CF-causing	No
c.3884_3885insT	p.Ser1297PhefsX5	4016insT	37	0.00052	CF-causing	CF-causing	No
c.3909C>G	p.Asn1303Lys	N1303K	1242	0.01755	CF-causing	CF-causing	No
c.3937C>T	p.Gln1313X	Q1313X	26	0.00037	CF-causing	CF-causing	No
c.3964-78_4242+577del	NULL	CFTRdele22,23	17	0.00024	CF-causing	CF-causing	No
c.4046G>A	p.Gly1349Asp	G1349D	5	0.00007	CF-causing	CF-causing	No
c.4077_4080delTGTTinsAA	No protein name	4209TGTT->AA	9	0.00013	CF-causing	CF-causing	No
c.4111G>T	p.Glu1371X	E1371X	4	0.00006	CF-causing	CF-causing	No
c.4196_4197delTC	p.Cys1400X	4326delTC	7	0.00010	CF-causing	CF-causing	No
c.4234C>T	p.Gln1412X	Q1412X	3	0.00004	CF-causing	CF-causing	No
c.4242+1G>T	No protein name	4374+1G->T	9	0.00013	CF-causing	CF-causing	No
c.4251delA	p.Glu1418ArgfsX14	4382delA	29	0.00041	CF-causing	CF-causing	No
c.4296_4297insGA	p.Ser1435GlyfsX14	4428insGA	4	0.00006	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.

‡These mutations were combined into the single mutation 2183AA->G as of 11 October 2013

§This mutation was incorrectly listed in the 22 July 2013 version of this PDF as a mutation of "Varying clinical consequence," though its characterization on the website when searched was "Unknown significance." The characterization on the website of "Unknown significance" is the correct characterization. This recent version of the PDF has been modified to reflect the correct website characterization.