

Elucigene® CF-EU2v1

Table of CFTR gene small insertions and deletions – arranged by peak no.

CFTR gene position	CF-EU2 Peak No	Amplicon	Mutation (HGVS Nomenclature)	Size change of amplicon
exon 7	1	R347H	c.1053_1054delTC	-2
exon 7	1		c.1069delG	-1
exon 7	2	R347P	c.1053_1054delTC	-2
exon 7	2		c.1069delG	-1
intron 14b	3	2789+5G>A	c.2629_2631delTCT	-3
intron 14b	3		c.2634_2641delGGTTGTGC	-8
intron 14b	3		c.2655_2670delAAACACTCCTCTTCAA	-16
intron 16	4	3120+1G>A	c.2947_2948delTT	-2
intron 16	4		c.2968_2969insA	1
Intron 5	5	711+1 G>T	none	
exon 7	6	R334W	c.1006_1007insG	1
exon 7	6		c.1008_1019dup12	12
exon 7	6		c.1016_1018del	-3
exon 7	6		c.1018delA	-1
exon 7	6		c.1018_1019insTC	2
exon 7	6		c.1022_1023dupTC	2
exon 7	6		c.1029delC	-1
exon 7	6		c.1029_1030insG	1
exon 7	6		c.1053_1054delTC	-2
exon 7	6		c.1069delG	-1
exon 10	7	I507del	c.1482_1483delTT	-2
exon 10	7		c.1477_1478delCA	-2
exon 10	7		c.1469_1470delTC	-2
exon 10	7		c.1444_1445insT	1
exon 10	7		c.1439delG	-1
exon 10	7		c.1433_1434delCA	-2
intron 19	9	3849+10KbC>T	none	
exon 10	10	1677delTA	c.1528delG	-1
exon 10	10		c.1521_1523delCTT (F508del)	-3
exon 10	10		c.1519_1521delATC (I507del)	-3
exon 10	10		c.1482_1483delTT	-2
exon 10	10		c.1477_1478delCA	-2
exon 10	10		c.1469_1470delTC	-2
exon 10	10		c.1444_1445insT	1
exon 10	10		c.1439delG	-1
exon 10	10		c.1433_1434delCA	-2

exon 7	11	1078delT	c.980delT	-1
exon 7	11		c.987delA	-1
exon 7	11		c.1006_1007insG	1
exon 7	11		c.1016_1018del	-3
exon 7	11		c.1018delA	-1
exon 7	11		c.1018_1019insTC	2
exon 7	11		c.1008_1019dup12	12
exon 7	11		c.1022_1023dupTC	2
exon 7	11		c.1029delC	-1
exon 7	11		c.1029_1030insG	1
exon 7	11		c.1053_1054delTC	-2
exon 7	11		c.1069delG	-1
exon 10	12	V520F	c.1545_1546delTA (1677delTA)	-2
exon 10	12		c.1528delG	-1
exon 10	12		c.1521_1523delCTT (F508del)	-3
exon 10	12		c.1519_1521delATC (I507del)	-3
exon 10	12		c.1482_1483delTT	-2
exon 10	12		c.1477_1478delCA	-2
exon 10	12		c.1469_1470delTC	-2
exon 10	12		c.1444_1445insT	1
exon 10	12		c.1439delG	-1
exon 10	12		c.1433_1434delCA	-2
exon 6a	13	L206W	c.601delG	-1
exon 20	14	W1282X	c.3855delC	-1
exon 11	15	R560T	none	
exon 13	16	2347delG	c.2240_2247delCGATACTG	-8
exon 13	16		c.2248_2255del	-8
exon 13	16		c.2276_2277delCC	-2
exon 13	16		c.2277delC	-1
exon 13	16		c.2291delG	-1
exon 13	16		c.2324_2325delAC	-2
exon 13	16		c.2361_2362ins8	1
exon 13	16		c.2380delG	-1
exon 15	17	Q890X	c.2686_2687insT	1
exon 15	17		c.2687_2690delCTCAinsTGAGTACTATGAG	13
exon 15	17		c.2737_2738insG	1
exon 15	17		c.2764_2765insAG	2
exon 15	17		c.2775_2776delTT	-2
exon 15	17		c.2777delT	-1
exon 15	17		c.2810_2811insT	1
exon 15	17		c.2816_2817delATinsC	-1
exon 15	17		c.2825delT	-1
exon 11	18	R553X	c.1660_1661insA	1
exon 11	18		c.1670delC	-1
exon 11	18		c.1674delA	-1

exon 11	19	G551D	c.1656delA	-1
exon 11	19		c.1660_1661insA	1
exon 11	19		c.1670delC	-1
exon 11	19		c.1674delA	-1
exon 11	20	S549N	c.1650delA	-1
exon 11	20		c.1652delG	-1
exon 11	20		c.1656delA	-1
exon 11	20		c.1660_1661insA	1
exon 11	20		c.1670delC	-1
exon 11	20		c.1674delA	-1
exon 11	21	S549R(T>G)	c.1650delA	-1
exon 11	21		c.1652delG	-1
exon 11	21		c.1656delA	-1
exon 11	21		c.1660_1661insA	1
exon 11	21		c.1670delC	-1
exon 11	21		c.1674delA	-1
exon 17b	22	M1101K	c.3315delG	-1
exon 17b	22		c.3364delA	-1
exon 11	23	G542X	c.1635_1640del	-6
exon 11	23		c.1642_1643delCT	-2
exon 11	23		c.1645_1648delAGTG	-4
exon 11	23		c.1650delA	-1
exon 11	23		c.1652delG	-1
exon 11	23		c.1656delA	-1
exon 11	23		c.1660_1661insA	1
exon 11	23		c.1670delC	-1
exon 11	23		c.1674delA	-1
exon 20	24	3905insT	c.3774_3775insG	1
exon 20	24		c.3780_3782delACT	-3
exon 20	24		c.3790_3799delGAAGGAGAAA	9
exon 20	24		c.3816_3817delGT	-2
exon 20	24		c.3829delA	-1
exon 20	24		c.3835_3836delTT	-2
exon 20	24		c.3855delC	-1
exon 17b	25	Y1092X(C>A)	c.3287delT	-1
exon 17b	25		c.3291delC	-1
exon 17b	25		c.3294delG	-1
exon 17b	25		c.3315delG	-1
exon 17b	25		c.3364delA	-1
exon 4	26		c.280_286delACCAAAG	-5
exon 20	26	S1251N	c.3766_3767insC	1
exon 20	26		c.3773_3774insT (3905insT)	1
exon 20	26		c.3774_3775insG	1
exon 20	26		c.3780_3782delACT	-3

exon 20	26		c.3790_3799delGAAGGAGAAA	9
exon 20	26		c.3816_3817delGT	-2
exon 20	26		c.3829delA	-1
exon 20	26		c.3835_3836delTT	-2
exon 20	26		c.3855delC	-1
exon 4	27	444delA	c.310delA	-1
exon 4	27		c.303_304insA	1
intron 11	28	1811+1.6kbA>G	none	
intron 10	29	1717-1 G>A	c.1610_1611delAC	-2
intron 10	29		c.1617_1618insTA	2
intron 10	29		c.1635_1640del	-6
intron 10	29		c.1642_1643delCT	-2
intron 10	29		c.1645_1648delAGTG	-4
intron 10	29		c.1650delA	-1
intron 10	29		c.1652delG	-1
intron 10	29		c.1656delA	-1
intron 10	29		c.1660_1661insA	1
intron 10	29		c.1670delC	-1
intron 10	29		c.1674delA	-1
exon 4	30	R117H	c.343_345del	-3
exon 4	30		c.328delG	-1
exon 4	30		c.326_327delAT	-2
exon 4	30		c.325_327delTATinsG	-2
exon 4	30		c.319_326delGCTTCCTA	-8
exon 4	30		c.313delA (444delA)	-1
exon 4	30		c.310delA	-1
exon 4	30		c.303_304insA	1
exon 4	30		c.280_286delACCAAAG	-5
exon 4	31	R117C	c.343_345del	-3
exon 4	31		c.328delG	-1
exon 4	31		c.326_327delAT	-2
exon 4	31		c.325_327delTATinsG	-2
exon 4	31		c.319_326delGCTTCCTA	-8
exon 4	31		c.313delA (444delA)	-1
exon 4	31		c.310delA	-1
exon 4	31		c.303_304insA	1
exon 4	31		c.280_286delACCAAAG	-5
exon 21	32	N1303K	c.3876delA	-1
exon 21	32		c.3882_3885delTATT	-4
exon 21	32		c.3883delA	-1
exon 21	32		c.3884_3885insT	1
exon 21	32		c.3890_3891insT	1
exon 21	32		c.3908delA	-1
exon 21	32		c.3908dupA	1
exon 4	33	Y122X	c.357delC	-1

exon 4	33		c.343_345del	-3
exon 4	33		c.328delG	-1
exon 4	33		c.326_327delAT	-2
exon 4	33		c.325_327delTATinsG	-2
exon 4	33		c.319_326delGCTTCCTA	-8
exon 4	33		c.313delA (444delA)	-1
exon 4	33		c.310delA	-1
exon 4	33		c.303_304insA	1
exon 4	33		c.280_286delACCAAAG	-5
exon 3	34	394delTT	None	
exon 3	35	G85E	c.262_263delTT (394delTT)	-2
exon 17b	36	R1066C	c.3199_3200delinsA	-1
exon 17b	36		c.3229_3230delCT	-2
exon 17b	36		c.3263dupA	1
exon 17b	36		c.3264delC	-1
exon 17b	36		c.3287delT	-1
exon 17b	36		c.3291delC	-1
exon 17b	36		c.3294delG	-1
exon 17b	36		c.3315delG	-1
exon 17b	36		c.3364delA	-1
intron 12	37	1898+1G>A	c.1681_1682insC	1
intron 12	37		c.1692delA	-1
intron 12	37		c.1703delT	-1
intron 12	37		c.1713_1714delAG	-2
intron 12	37		c.1738delG	-1
intron 12	37		c.1739_1740insT	1
exon 14a	38	W846X	c.2502delT	-1
exon 14a	38		c.2502_2503insT	1
exon 14a	38		c.2508delT	-1
exon 13	39	2184delA/2183AA>G	c.2051_2052delAA	-2
exon 13	39		c.2052delA (2184delA)	-1
exon 13	39		c.2052_2053insA	1
exon 13	39		c.2053_2054insC	1
exon 13	39		c.2061_2062insTTTT	4
exon 13	39		c.2083_2084insG	1
exon 13	39		c.2089_2090insA	1
exon 13	39		c.2157_2163delinsGT	-5
exon 13	39		c.2175_2176insA	1
exon 13	39		c.2203delA	-1
exon 13	39		c.2215delG (2347delG)	-1
exon 13	39		c.2240_2247delCGATACTG	-8
exon 13	39		c.2248_2255del	-8
exon 13	39		c.2276_2277delCC	-2
exon 13	39		c.2277delC	-1
exon 13	39		c.2291delG	-1
exon 13	39		c.2324_2325delAC	-2

exon 13	39		c.2361_2362ins8	1
exon 13	39		c.2380delG	-1
exon 18	40	D1152H	c.3371_3373delAAG	-3
exon 18	40		c.3400_3401delACinsGTA	1
exon 18	40		c.3418_3420delATG	-3
exon 18	40		c.3424_3425insAGTA	4
exon 18	40		c.3445delT	-1
exon 2	41	CFTRdele2,3	none	
exon 3	42	P67L	c.217delC	-1
exon 3	42		c.227_228insT	1
exon 3	42		c.228_229insT	1
exon 3	42		c.233delT	-1
exon 3	42		c.233_234insT	1
exon 3	42		c.234delC	-1
exon 3	42		c.247_248insT	1
exon 3	42		c.262_263delITT (394delITT)	-2
exon 13	43	2143delT	c.2013_2015delAGA	-3
exon 13	43		c.2044delA	-1
exon 13	43		c.2044_2045insC	1
exon 13	43		c.2051_2052delAAinsG (2183AA>G)	-1
exon 13	43		c.2051_2052delAA	-2
exon 13	43		c.2052delA (2184delA)	-1
exon 13	43		c.2052_2053insA	1
exon 13	43		c.2053_2054insC	1
exon 13	43		c.2061_2062insTTTT	4
exon 13	43		c.2083_2084insG	1
exon 13	43		c.2089_2090insA	1
exon 13	43		c.2157_2163delinsGT	-5
exon 13	43		c.2175_2176insA	1
exon 13	43		c.2203delA	-1
exon 13	43		c.2215delG (2347delG)	-1
exon 13	43		c.2240_2247delCGATACTG	-8
exon 13	43		c.2248_2255del	-8
exon 13	43		c.2276_2277delCC	-2
exon 13	43		c.2277delC	-1
exon 13	43		c.2291delG	-1
exon 13	43		c.2324_2325delAC	-2
exon 13	43		c.2361_2362ins8	1
exon 13	43		c.2380delG	-1
exon 3	44	E60X	c.217delC	-1
exon 3	44		c.227_228insT	1
exon 3	44		c.228_229insT	1
exon 3	44		c.233delT	-1
exon 3	44		c.233_234insT	1
exon 3	44		c.234delC	-1
exon 3	44		c.247_248insT	1
exon 3	44		c.262_263delITT (394delITT)	-2

exon 19	45	3659delC	c.3530delA	-1
exon 19	45		c.3535_3538delACCA	-4
exon 19	45		c.3535_3536insTCAA	4
exon 19	45		c.3540delA	-1
exon 19	45		c.3592delG	-1
exon 19	45		c.3605delA	-1
exon 19	45		c.3618_3619delAG	-2
exon 19	45		c.3623delG	-1
exon 19	45		c.3657_3658insA	1
exon 19	45		c.3659delC	-1
exon 19	45		c.3691delT	-1
exon 19	45		c.3691delT	-1
intron 17a	46	3272-26A>G	c.3161delA	-1
intron 17a	46		c.3188_3189insCTATG	5
intron 17a	46		c.3199_3200delinsA	-1
intron 17a	46		c.3229_3230delCT	-2
intron 17a	46		c.3263dupA	1
intron 17a	46		c.3264delC	-1
intron 17a	46		c.3287delT	-1
intron 17a	46		c.3291delC	-1
intron 17a	46		c.3294delG	-1
intron 17a	46		c.3315delG	-1
intron 17a	46		c.3364delA	-1
intron 4	47	621+1G>T	.473_474insT	1
intron 4	47		c.459_476delAATAGCTATGTTTAGTTT	-18
intron 4	47		c.444_445InsCTA	3
intron 4	47		c.442delA	-1
intron 4	47		c.433delC	-1
intron 4	47		c.429delT	-1
intron 4	47		c.424delA	-1
intron 4	47		c.420_421insA	1
intron 4	47		c.415_416insTA	2
intron 4	47		c.415_416insGA	2
intron 4	47		c.414_415insCTA	3
intron 4	47		c.412_413insACT	3
intron 4	47		c.409delC	-1
intron 4	47		c.409_412delCTCC	-4
intron 4	47		c.393delT	-1
intron 4	47		c.387delT	-1
intron 4	47		c.357delC	-1
intron 4	47		c.343_345del	-3
intron 4	47		c.328delG	-1
intron 4	47		c.326_327delAT	-2
intron 4	47		c.325_327delTATinsG	-2
intron 4	47		c.319_326delGCTTCCTA	-8
intron 4	47		c.313delA (444delA)	-1
intron 4	47		c.310delA	-1
intron 4	47		c.303_304insA	1
intron 4	47		c.280_286delACCAAAG	-5

exon 9	48	A455E	c.1365_1366delGG	-2
exon 9	48		c.1373delG	-1
exon 9	48		c.1392+60insA	1
exon 19	49	R1162X	c.3485_3486delGA	-2
exon 19	49		c.3490_3491insT	1
exon 19	49		c.3497delT	-1
exon 19	49		c.3528delC (3659delC)	-1
exon 19	49		c.3530delA	-1
exon 19	49		c.3535_3538delACCA	-4
exon 19	49		c.3535_3536insTCAA	4
exon 19	49		c.3540delA	-1
exon 19	49		c.3592delG	-1
exon 19	49		c.3605delA	-1
exon 19	49		c.3618_3619delAG	-2
exon 19	49		c.3623delG	-1
exon 19	49		c.3657_3658insA	1
exon 19	49		c.3659delC	-1
exon 19	49		c.3691delT	-1
exon 19	49		c.3691delT	-1
exon 19	50	R1158X	c.3485_3486delGA	-2
exon 19	50		c.3490_3491insT	1
exon 19	50		c.3497delT	-1
exon 19	50		c.3528delC (3659delC)	-1
exon 19	50		c.3530delA	-1
exon 19	50		c.3535_3538delACCA	-4
exon 19	50		c.3535_3536insTCAA	4
exon 19	50		c.3540delA	-1
exon 19	50		c.3592delG	-1
exon 19	50		c.3605delA	-1
exon 19	50		c.3618_3619delAG	-2
exon 19	50		c.3623delG	-1
exon 19	50		c.3657_3658insA	1
exon 19	50		c.3659delC	-1
exon 19	50		c.3691delT	-1
exon 19	50		c.3691delT	-1