Liste des mutations du gène CFTR recherchées par la trousse Elucigene® CF-EU2v1

Marqueur n°	Nomenclature traditionnelle	Nomenclature HGVS	Exon /Intron
1	R347H	c.1040G>A	exon 7
2	R347P	c.1040G>C	exon 7
3	2789+5 G>A	c.2657+5G>A	intron 14 b
4	3120+1 G>A	c.2988+1G>A	intron 16
5	711+1 G>T	c.579+1G>T	intron 5
6	R334W	c.1000C>T	exon 7
7	I507del	c.1519_1521del	exon 10
8	F508del	c.1521_1523del	exon 10
9	3849+10kb C>T	c.3718-2477C>T	intron 19
10	1677delTA	c.1545_1546del	exon 10
11	1078delT	c.948del	exon 7
12	V520F	c.1558G>T	exon 10
13	L206W	c.617T>G	exon 6a
14	W1282X	c.3846G>A	exon 20 / intron 20
15	R560T	c.1679G>C	exon 11
16	2347delG	c.2215del	exon 13
17	Q890X	c.2668C>T	exon 15
18	R553X	c.1657C>T	exon 11
19	G551D	c.1652G>A	exon 11
20	S549R(T>G)	c.1647T>G	exon 11
21	S549N	c.1646G>A	exon 11
22	M1101K	c.3302T>A	exon 17b
23	G542X	c.1624G>T	exon 11
24	3905insT	c.3773dup	exon 20 / intron 20
25	Y1092X(C>A)	c.3276C>A	exon 17b
26	S1251N	c.3752G>A	exon 20 / intron 20
27	444delA	c.313del	exon 4
28	1811+1.6kb A>G	c.1680-886A>G	intron 11
29	1717-1 G>A	c.1585-1G>A	exon 11
30	R117H	c.350G>A	exon 4
31	R117C	c.349C>T	exon 4
32	N1303K	c.3909C>G	intron 20 / exon 21
33	Y122X	c.366T>A	exon 4
34	394delTT	c.262_263del	exon 3
35	G85E	c.254G>A	exon 3
36	R1066C	c.3196C>T	exon 17b
37	1898+1 G>A	c.1766+1G>A	exon 12
38	W846X	c.2538G>A	exon 14a
39	2184delA	c.2052del	exon 13
40	D1152H	c.3454G>C	exon 18
41	CFTRdel2_3	c.54-5940_273+1025del	intron 1 /exon 3
42	P67L	c.200C>T	exon 3
43	2143delT	c.2012del	exon 13
44	E60X	c.178G>T	exon 3
45	3659delC	c.3528del	exon 19
46	3272-26A>G	c.3140-26A>G	exon 17b
47	621+1G	c.489+1G>T	exon 4
48	A455E	c.1364C>A	exon 9 / intron 9
49	R1162X	c.3484C>T	exon 19
50	R1158X	c.3472C>T	exon 19
Variant 5T*	IVS8-5T		
Variant 7T	IVS8-7T		
Variant 9T	IVS8-9T		