Referenční sekvence - CFTR

Název varianty dle tradiční nomenklatury	Název varianty dle NM_000492.4
711+ 1G>T	c.579+1G>T
2043delG	c.1911delG
1677delTA	c.1545_1546delTA
W1282X	c.3846G>A
R1283M	c.3848G>T
K710X[2]	c.2128A>T
3849+10kbC>T	c.3718-2477C>T
I507_F508-wt[3]	c.1519_1523ATCTT
2789+5G>A	c.2657+5G>A
M1101K	c.3302T>A
G85E	c.254G>A
3905insT	c.3773_3774insT
1525-1G>A	c.1393-1G>A
2184delA	c.2052delA
3659delC	c.3528delC
N1303K	c.3909C>G
2184insA	c.2052_2053insA
1812-1G->A	c.1680-1G>A
CFTRdele2,3	c.54-5940_273+10250del21kb
2143delT	c.2012delT
Y569D	c.1705T>G
R1162X	c.3484C>T
A561E[2]	c.1682C>A
S1251N[2]	c.3752G>A
P67L	c.200C>T
R1158X	c.3472C>T
1609delCA	c.1477_1478delCA
Q493X	c.1477C>T
E60X	c.178G>T
1898+1G>A	c.1766+1G>A
1898+5G>T[2]	c.1766+5G>T
I507del	c.1519_1521delATC
F508del	c.1521_1523delCTT

Název varianty dle tradiční nomenklatury	Název varianty dle NM_000492.4
V520F	c.1558G>T
394delTT	c.262_263delTT
D1152H	c.3454G>C
V232D[2]	c.695T>A
L218X	c.653T>A
621+2T>C[2]	c.489+2T>C
1717-1G>A	c.1585-1G>A
L206W	c.617T>G
E92X	c.274G>T
3120+1G>A	c.2988+1G>A
G542X	c.1624G>T
S549N	c.1646G>A
G551D	c.1652G>A
712-1G>T	c.580-1G>T
R553X	c.1657C>T
3272-26A>G	c.3140-26A>G
R560T	c.1679G>C
2183AA>G	c.2051_2052delAAinsG
R117H	c.350G>A
R117C	c.349C>T
1811+1.6kbA>G	c.1680-886A>G
2869insG[2]	c.2737_2738insG
Y122X	c.366T>A
Q890X[2]	c.2668C>T
R1066C	c.3196C>T
R347H	c.1040G>A
R347P	c.1040G>C
1161delC[2]	c.1029delC
1154ins TC	c.1022_1023insTC
E92K	c.274G>A
I336K	c.1007T>A
R334W	c.1000C>T
Y1092X (C>A)	c.3276C>A
621+1G>T	c.489+1G>T

Název varianty dle tradiční nomenklatury	Název varianty dle NM_000492.4
1078delT	c.948delT
A455E	c.1364C>A
5T (TG9-13)	c.[1210−12[5];1210- 34TG[9-13]]
7T	c.1210-12[7]
9T	c.1210-12[9]