DEVYSER CFTR CORE PLUS 1.1

Table of reported primer site polymorphisms/mutations

Normal allele Plus 1	Mutant allele Plus 2	Reported polymorphisms/mutations	CFTR gene location
CFTRdele2,3_wt	CFTRdele2,3 (21kb)	None	Intron 1 - Exon 3
E60	E60X	c.165-10T>G	Exon 3
		c.165-3C>T	
		c.165-3C>A	
		c.165-2A>G	
		c.165-1G>A	
		c.166G>A	
		c.168delA	
		c.169T>G	
		c.169T>C	
		c.170G>A	
		c.171G>A	
		c.172G>A	
		c.173A>G	
		c.174_177delTAGA	
		c.174_175insA	
G85	G85E	c.250T>C	Exon 3
		c.227_228insT	
		c.228_229insT	
		c.233_234insT	
		c.233delT	
		c.234delC	
		c.235T>C	
		c.236G>A	
		c.244A>G	
		c.247_248insT	
		c.254G>T	
E92	E92X and E92K	c.274-13T>C	Exon 4
		c.274-10C>G	
		c.274-6T>C	
		c.274-5T>G	
		c.274-3T>C	
		c.274-2A>C	
		c.274-2A>G	
		c.274-1G>C	
		c.274-1G>T	
		c.274-1G>A	

Normal allele Plus 1	Mutant allele Plus 2	Reported polymorphisms/mutations	CFTR gene location
R117	R117H and R117C	c.327T>A	Exon 4
		c.328delG	
		c.328G>A	
		c.328G>C	
		c.328G>T	
		c.330C>A	
		c.331C>G	
		c.332C>T	
		c.333G>A	
		c.338A>T	
		c.340A>T	
		c.343_345del	
		c.346G>A	
		c.346G>C	
		c.349C>G	
		c.350G>C	
		c.350G>T	
621+1G	621+1G>T	c.459_476del18	Intron 4
		c.463G>A	
		c.472A>C	
		c.473_474insT	
		c.473G>A	
		c.473G>C	
		c.476T>C	
		c.480T>A	
		c.481T>A	
		c.481T>G	
		c.482A>C	
		c.484A>G	
		c.488A>G	
		c.488delA	
		c.489G>A	
711+1G	711+1G>T	c.558C>A	Intron 5
		c.561C>A	
		c.563T>A	
		c.566A>G	
		c.567C>A	
		c.574_576delGAC	
		c.574G>A	
		c.575A>G	
		c.577G>A	
		c.577G>T	
		c.578_579+5del7	

Normal allele Plus 1	Mutant allele Plus 2	Reported polymorphisms/mutations	CFTR gene location
1078	1078delT	c.925G>A	Exon 8
		c.926C>A	
		c.926C>G	
		c.926C>T	
		c.927C>T	
		c.927delC	
		c.933_935delCTT	
		c.933C>G	
		c.938C>A	
		c.941G>A	
		c.941G>T	
		c.948T>G	
		c.950T>C	
R334	R334W	c.974A>G	Exon 8
		c.980delT	
		c.980T>G	
		c.987delA	
		c.988G>T	
		c.992T>A	
		c.997C>T	
I336	I336K	c.987delA	Exon 8
		c.988G>T	
		c.992T>A	
		c.997C>T	
		c.1000C>T	
		c.1001C>T	
		c.1006_1007insG	
		c.1006A>C	
T338	T338I	c.992T>A	Exon 8
		c.997C>T	
		c.1000C>T	
		c.1001G>A	
		c.1001G>T	
		c.1006_1007insG	
		c.1006A>C	
		c.1007T>A	
		c.1012A>G	
R347	R347H and R347P	c.1022_1023insTC	Exon 8
		c.1029_1030insG	
		c.1029delC	
		c.1032T>A	
		c.1036C>T	
		c.1037T>C	
		c.1039C>T	
		c.1040G>T	

Normal allele Plus 1	Mutant allele Plus 2	Reported polymorphisms/mutations	CFTR gene location
A455	A455E	c.1343T>G	Exon 10
		c.1353_1354insT	
		c.1355A>C	
		c.1359_1361delGTT	
I507	I507del	c.1517T>C	Exon 11
		c.1517T>G	
		c.1518C>G	
		c.1521_1523delCTT	
		c.1521C>G	
		c.1523T>C	
		c.1523T>G	
		c.1528delG	
F508	F508del	c.1523A>G	Exon 11
		c.1523T>C	
		c.1526G>A	
		c.1528delG	
		c.1532C>G	
		c.1538A>G	
		c.1543T>C	
		c.1549T>C	
1677	1677delTA	c.1543T>C	Exon 11
		c.1546A>G	
		c.1550A>G	
		c.1555A>G	
		c.1558G>A	
		c.1558G>T	
		c.1561A>C	
		c.1561A>T	
		c.1567G>T	
1717-1G	1717-1G>A	c.1585G>C	Intron 11
		c.1586A>G	
		c.1601C>A	
		c.1606A>G	
G542	G542X	c.1625G>A	Exon 12
		c.1630G>A	
		c.1631G>T	
		c.1632T>G	
		c.1635_1640del	
		c.1641A>T	
		c.1642_1643del	
		c.1643T>A	
		c.1645A>C	
G551	G551D	c.1652delG	Exon 12
		c.1652delG	
		c.1654C>A	
		c.1654C>T	
		c.1656delA	
		c.1657C>G	
		c.1657C>T	
		c.1658G>A	
		c.1660_1661insA	
		c.1663A>G	
		c.1666A>G	
		c.1670delC	
		c.1673T>C	
		c.1674delA	

Normal allele Plus 1	Mutant allele Plus 2	Reported polymorphisms/mutations	CFTR gene location
R553	R553X	c.1657C>G	Exon 12
		c.1658G>A	
		c.1660_1661insA	
		c.1663A>G	
		c.1666A>G	
		c.1670delC	
		c.1673T>C	
		c.1674delA	
		c.1675G>A	
		c.1675G>C	
		c.1676C>A	
		c.1676C>T	
		c.1678A>G	
R560	R560T	c.1679+11A>G	Exon 12
		c.1679+16T>C	
		c.1679+18G>A	
		c.1679+1G>A	
		c.1679+1G>C	
		c.1679+24G>A	
		c.1679+2T>C	
		c.1679+5A>G	
		c.1679+9C>G	
		c.1679G>A	
1898+1G	1898+1G>A	c.1680-108T>C	Intron 13
		c.1766+1G>C	
		c.1766+1G>T	
		c.1766+2T>C	
		c.1766+3A>C	
2043	2043delT	c.1915G>T	Exon 14
		c.1919_1920delTT	
		c.1920_1921dupTA	
		c.1923_1931del9insA	
		c.1932C>G	
2143	2143delT	c.1990G>T	Exon 14
		c.1993A>T	
		c.2002C>T	
		c.2009_2010insA	

Normal allele Plus 1	Mutant allele Plus 2	Reported polymorphisms/mutations	CFTR gene location
2183_2184AA	2184AA>G and 2184insA	c.2036G>A	Exon 14
		c.2044_2045insC	
		c.2044delA	
		c.2048A>G	
		c.2051_2052delAA	
		c.2052A>G	
	2184delA	c.2048A>G	
		c.2051_2052delAAinsG	
		c.2051_2052delAA	
		c.2052A>G	
		c.2052_2053insA	
		c.2053_2054insC	
		c.2053C>T	
		c.2057C>T	
		c.2061_2062insTTTT	
		c.2052A>T	
		c.2065C>T	
		c.2068A>G	
2789+5G	2789+5G>A	c.2645G>A	Intron 16
		c.2655_2670del16	
		c.2657+2_2657+3insA	
		c.2657+3delG	
3120+1G	3120+1G>A	c.2909-1G>A	Intron 18
		c.2909delG	
		c.2909G>A	
		c.2916_2917delTCinsAT	
		c.2918T>A	
		c.2918T>C	
		c.2924_2925delGA	
		c.2929T>C	
		c.2930C>G	
		c.2930C>T	
		c.2932A>T	
		c.2988+2T>C	
		c.2988+30T>C	
3272-26A	3272-26A>G	c.3140-33A>G	Intron 19
		c.3140-42G>T	
L1065	L1065P	c.3176T>G	Exon 20
		c.3176T>G	
		c.3177A>G	
		c.3177A>G	
		c.3179A>C	
		c.3179A>C	
		c.3180delA	
		c.3181G>C	
		c.3186A>G	
		c.3188_3189insCTATG	
		c.3189G>A	
		c.3193C>T	
		c.3194T>G	

Normal allele Plus 1	Mutant allele Plus 2	Reported polymorphisms/mutations	CFTR gene location
R1066	R1066C	c.3176T>G	Exon 20
		c.3177A>G	
		c.3179A>C	
		c.3180delA	
		c.3181G>C	
		c.3186A>G	
		c.3188_3189insCTATG	
		c.3189G>A	
		c.3193C>T	
		c.3194T>C	
		c.3194T>G	
		c.3196C>A	
		c.3196C>G	
L1077	L1077P	c.3208C>T	Exon 20
		c.3209G>C	
		c.3211C>T	
		c.3212A>C	
		c.3213G>T	
		c.3215C>T	
		c.3218A>G	
		c.3220T>C	
		c.3222T>A	
		c.3229_3230delCT	
Y1092	Y1092X (C>A)	c.3256A>G	Exon 20
		c.3257C>T	
		c.3259G>C	
		c.3262A>G	
		c.3263dupA	
		c.3264delC	
		c.3266G>A	
		c.3274T>C	
		c.3275A>G	
		c.3276C>G	
M1101	M1101K	c.3285A>T	Exon 20
		c.3287delT	
		c.3287T>G	
		c.3291delC	
		c.3292T>C	
		c.3294delG	
		c.3293G>T	
		c.3293G>A	
		c.3294G>A	
		c.3294G>C	
		c.3297C>A	
		c.3298C>A	
		c.3299A>C	
		c.3302T>G	
R1162	R1162X	c.3469-2A>G	Exon 22
		c.3472C>A	
		c.3472C>T	
		c.3475T>C	
		c.3476C>T	
		c.3481A>C	
3659	3659delC	None	Exon 22
3849+10kbC	3849+10kbC>T	None	Intron 22

Normal allele Plus 1	Mutant allele Plus 2	Reported polymorphisms/mutations	CFTR gene location
3905	3905insT	c.3752G>A	Exon 22
		c.3754A>C	
		c.3759G>A	
		c.3759G>C	
		c.3761T>G	
		c.3763T>C	
		c.3764C>A	
		c.3764C>T	
		c.3766_3767insC	
		c.3767C>T	
		c.3771T>G	
W1282	W1282X	c.3822G>A	Exon 23
		c.3825T>C	
		c.3829delA	
		c.3835_3836delTT	
		c.3841C>T	
		c.3844T>C	
		c.3844T>G	
		c.3846G>T	
N1303	N1303K	c.3874-200G>A	Exon 24
		c.3909_3914del6insTGT	
		c.3915T>A	
		c.3916_3917insCC	
		c.3918C>T	
		c.3920A>G	
		c.3921T>A	
		c.3922G>T	
		c.3925C>G	
		c.3925C>T	
		c.3927G>A	
		c.3929G>A	
		c.3931A>G	
		c.3932G>A	

References

Cystic Fibrosis Mutation Database: www.genet.sickkids.on/cftr/app