

DEVYSER CFTR IBERIA - REPORTED PRIMER SITE POLYMORPHISMS/MUTATIONS

Normal allele Iberia 1	Mutant allele Iberia 2	Reported polymorphisms/mutations	CFTR gene location
712-1G	712-1G>T	none	Intron 5
H199	H199Y	c.592G>C	Exon 6
		c.592G>A	
		c.581G>T	
		c.580G>A	
		c.580-1G>T	
P205	P205S	c.609C>T	Exon 6
		c.609C>G	
		c.606G>A	
		c.601delG	
		c.601G>A	
		c.598T>A	
		c.597T>G	
		c.596A>G	
		c.595C>T	
		c.592G>C	
		c.592G>A	
		c.581G>T	
		c.580G>A	
		c.580-1G>T	
L206	L206W	c.614C>G	Exon 6
		c.614C>T	
		c.613C>T	
		c.609C>T	
		c.609C>G	
		c.606G>A	
		c.601delG	
		c.601G>A	
		c.598T>A	
		c.597T>G	
		c.596A>G	
		c.595C>T	
		c.592G>C	
		c.592G>A	
V232	V232D	c.680T>G	Exon 6
		c.675T>A	
		c.673T>C	

Normal allele Iberia 1	Mutant allele Iberia 2	Reported polymorphisms/mutations	CFTR gene location
1609CA	1609delCA	c.1482_1483delTT	Exon 11
		c.1484C>A	
		c.1486T>C	
		c.1487G>A	
		c.1489A>G	
		c.1494G>C	
		c.1495C>G	
		c.1501A>G	
S549	S549R(T>G)	c.1648G>A	Exon 12
		c.1648G>T	
		c.1650delA	
		c.1651G>A	
		c.1652delG	
		c.1652G>A	
		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	
1811+1.6kbA	1811+1.6kbA>G	None	Intron 12
1812-1G	1812-1G>A	c.1680A>C	Intron 12
		c.1681_1682insC	
		c.1682C>A	
		c.1684G>A	
		c.1684G>C	
		c.1687T>G	
		c.1687T>A	
		c.1687T>C	
		c.1688A>G	
		c.1690A>G	
		c.1692delA	
		c.1694A>G	
		c.1696G>A	
		c.1700A>G	
c.1703delT			
1898+3A	1898+3A>G	c.1766+3A>C	Intron 13
		c.1766+5G>T	
		c.1766+5G>A	
		c.1766+30G>A	

Normal allele Iberia 1	Mutant allele Iberia 2	Reported polymorphisms/mutations	CFTR gene location
R709	R709X	c.2106C>G	Exon 14
		c.2490+1G>T	
		c.2490+1G>A	
		c.2490+2_2490+7delTAGGTA	
		c.2490+2T>C	
		c.2490+3a>G	
		c.2490+5g>T	
		c.2490+14G>A	
		c.2490+20g>C	
Q890	Q890X	c.2658-1G>C	Exon 17
		c.2658-1G>T	
		c.2658-2A>G	
2869	2869insG	c.2719A>G	Exon 17
		c.2723C>A	
		c.2724C>T	
		c.2726G>T	
		c.2731A>C	
		c.2735C>T	
		c.2735C>A	
		c.2736G>A	
Q1100	Q1100P	c.3302T>A	Exon 20
		c.3302T>G	
		c.3304A>T	
		c.3306A>G	
		c.3308T>A	
		c.3310G>T	
		c.3310G>A	
		c.3314T>G	
		c.3315delG	
		c.3322G>C	
		c.3208C>T	
		c.3209G>C	
		c.3209G>A	
		c.3211C>T	
		c.3212A>C	
		c.3213G>T	
		c.3215C>T	
		c.3218A>G	
		c.3220T>C	
		c.3222T>A	

Normal allele Iberia 1	Mutant allele Iberia 2	Reported polymorphisms/mutations	CFTR gene location
M1101	M1101K	c.3302T>G	Exon 20
		c.3304A>T	
		c.3306A>G	
		c.3308T>A	
		c.3310G>T	
		c.3310G>A	
		c.3314T>G	
		c.3315delG	
		c.3322G>C	

References

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