

## DEVYSER CFTR ITALIA V2

**Table of reported primer site polymorphisms/mutations**

Normal allele Italia 1	Mutant allele Italia 2	Reported polymorphisms/mutations	CFTR gene location
<b>CFTRdele1-wt</b>	<b>CFTRdele1</b>	None	<b>Promotor / Intron 1</b>
<b>M1</b>	<b>M1V</b>	c.1A>C	<b>Exon 1</b>
		c.2T>A	
		c.2T>C	
		c.2T>G	
		c.3G>A	
		c.3G>T	
		c.4C>T	
		c.11C>A	
		c.14C>T	
		c.19G>T	
		c.24G>A	
<b>P5</b>	<b>P5L</b>	c.19G>T	<b>Exon 1</b>
		c.24G>A	
		c.28A>C	
		c.31G>A	
<b>CFTR-dele2-wt</b>	<b>CFTRdele2</b>	None	<b>Intron 1 / Intron 2</b>
<b>Dele2ins182-wt</b>	<b>Dele2ins182</b>	None	<b>Intron 1 / Intron 2</b>
<b>Q39</b>	<b>Q39X</b>	c.125C>T	<b>Exon 2</b>
		c.131A>T	
		c.131A>G	
		c.137C>A	
		c.137C>T	
<b>D110 (c.330C)</b>	<b>D110E</b>	c.310delA	<b>Exon 4</b>
		c.313delA	
		c.314T>A	
		c.319_326delGCTTCCTA	
		c.320C>G	
		c.320C>A	
		c.323C>T	
		c.325_327delTATinsG	
		c.325T>A	
		c.325T>C	
		c.326_327delAT	
		c.326A>G	
		c.327T>A	
		c.328delG	
		c.328G>A	
		c.328G>T	
c.328G>C			

Normal allele Italia 1	Mutant allele Italia 2	Reported polymorphisms/mutations	CFTR gene location
D110 (c.328G)	D110H	c.310delA	Exon 4
		c.313delA	
		c.314T>A	
		c.319_326delGCTTCCTA	
		c.320C>G	
		c.320C>A	
		c.323C>T	
		c.325_327delTATinsG	
		c.325T>A	
		c.325T>C	
		c.326_327delAT	
		c.326A>G	
		c.327T>A	
		c.328delG	
c.328G>A			
c.328G>T			
G178	G178R	c.508C>T	Exon 5
		c.508C>G	
		c.509G>A	
		c.518_522delATAAAA	
		c.523A>G	
		c.526delA	
		c.530T>C	
		c.531delT	
		c.531T>G	
711+5G	711+5G->A	c.558C>A	Exon 5 / Intron 5
		c.561C>A	
		c.563T>C	
		c.566A>G	
		c.567C>A	
		c.574G>A	
		c.574_576delGAT	
		c.575A>G	
		c.577G>A	
		c.577G>T	
		c.578_579+5delAAGTATG	
		c.579+1G>T	
		c.579+3A>C	
		c.579+3A>G	
c.579+3A>T			

Normal allele Italia 1	Mutant allele Italia 2	Reported polymorphisms/mutations	CFTR gene location
852del22-wt	852del22	c.713C>T <sup>1</sup>	Exon 6 / Intron 6
		c.714delT <sup>1</sup>	
		c.715G>A <sup>1</sup>	
		c.731T>A	
		c.741C>T	
		c.741C>G	
		c.742_743insTACA	
		c.743G>C	
		c.743+1G>A	
		c.743+1G>C	
		c.743+4G>T	
R352	R352Q	c.938C>A	Exon 8
		c.940G>C	
		c.941G>A	
		c.948delT	
		c.948T>G	
		c.954G>A	
		c.955T>G	
		c.1057C>T	
		c.1059A>C	
		c.1063C>T	
		c.1067G>C	
		c.1068G>A	
		c.1069delG	
		c.1075C>A	
c.1076A>G			
1259insA-wt	1259insA	c.1125A>G	Intron 8 / Exon 9
		c.1133A>G	
		c.1135G>A	
		c.1135G>T	
		c.1148T>C	
		c.1149G>A	
		c.1152delA	
c.1584+18672A	c.1584+18672A->G	none	Intron 11

Normal allele Italia 1	Mutant allele Italia 2	Reported polymorphisms/mutations	CFTR gene location
S549	S549R(A->C)	c.1645_1648delAGTG	Exon 12
		c.1646G>T	
		c.1646G>A	
		c.1647T>G	
		c.1648G>A	
		c.1648G>T	
		c.1650delA	
		c.1651G>A	
		c.1652delG	
		c.1652G>A	
		c.1654C>A	
		c.1654C>T	
		c.1656delA	
		c.1657C>T	
		c.1657C>G	
		c.1658G>A	
		c.1660_1661insA	
c.1663A>G			
c.1666A>G			
c.1670delC			
Q552	Q552X	c.1654C>A	Exon 12
		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	
		c.1675G>A	
		c.1675G>C	
		c.1676C>A	
c.1676C>T			

Normal allele Italia 1	Mutant allele Italia 2	Reported polymorphisms/mutations	CFTR gene location
D579	D579G	c.1680-108T>C	Intron 12 / Exon 13
		c.1736A>C	
		c.1738delG	
		c.1739_1740insT	
		c.1744A>T	
		c.1745C>G	
		c.1753G>T	
		c.1756A>G	
		c.1759T>A	
		c.1763A>T	
c.1745C>G			
E585	E585X	c.1680-108T>C	Exon 13 / Intron 13
		c.1756A>G	
		c.1759T>A	
		c.1763A>T	
		c.1766G>T	
		c.1766G>A	
		c.1766+1G>A	
		c.1766+1G>T	
		c.1766+1G>C	
		c.1766+2T>C	
		c.1766+3A>G	
		c.1766+3A>C	
		c.1766+5G>T	
c.1766+5G>A			
1898+3A	1898+3A->G	c.1680-108T>C	Intron 13
		c.1677+30G>A	
		c.1766+3A>C	
		c.1766+5G>T	
		c.1766+5G>A	
2184delA-wt	2184delA	c.2036G>A	Exon 14 / Intron 14
		c.2044_2045insC	
		c.2044delA	
		c.2048A>G	
		c.2051_2052delAAinsG	
		c.2051_2052delAA	
		c.2052A>G	
		c.2052_2053insA	
		c.2052dupA	
		c.2053C>T	
		c.2490+14G>A	
		c.2490+20G>C	
Dele17a_18-wt and Dele14b_17b-wt	Dele17a_18 and Dele14b_17b	None	Intron 19

Normal allele Italia 1	Mutant allele Italia 2	Reported polymorphisms/mutations	CFTR gene location
R1066	R1066H	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>T	
		c.3196C>A	
		c.3196C>G	
c.3197G>T			
D1152	D1152H	c.3435G>A	Exon 21
		c.3439G>A	
		c.3443A>G	
		c.3444C>A	
		c.3445delT	
R1158	R1158X	c.3469-111G>C	Intron 21 / Exon 22
		c.3472C>A	
		c.3475T>C	
		c.3476C>T	
		c.3481A>C	
		c.3484C>T	
		c.3485_3486delGA	
		c.3485G>T	
		c.3490_3491insT	
		c.3494A>C	
G1244	G1244E	c.3731G>T	Exon 23
		c.3735A>G	
		c.3737C>T	
		c.3739G>A	
		c.3739G>C	
		c.3744delA	
		c.3745G>A	
		c.3747delG	
		c.3746G>A	
		c.3754A>C	
		c.3757G>A	

Normal allele Italia 1	Mutant allele Italia 2	Reported polymorphisms/mutations	CFTR gene location
4016insT-wt	4016insT	c.3874-200G>A	Intron 23 / Exon 24
		c.3890_3891insT	
		c.3893G>C	
		c.3893G>T	
		c.3895delA	
		c.3896C>T	
		c.3897A>G	
		c.3898T>C	
		c.3905A>G	
		c.3907A>C	
		c.3908delA	
		c.3908A>T	
		c.3908dupA	
		c.3909_3914del6insTGT	
c.3909C>G			
CFTRdele22,23-wt	CFTRdele22,23	c.3964-3C>G	Intron 24 / Exon 25
		c.3964-1G>A	
		c.3971T>C	
		c.3976delT	
		c.3982_3984delATAinsTT	
Dele22_24-wt	Dele22_24	None	Intron 24
G1349	G1349D	c.4036_4042del	Exon 25
		c.4035_4038dupCCTA	
		c.4039_4040insA	
		c.4040_4041delIGC	
		c.4042delC	
		c.4045G>A	
4382delA -wt	4382delA	c.4243-15C>T	Intron 26 / Exon 27
		c.4243-3T>A	
		c.4243-2A>C	
		c.4243-1G>C	
		c.4243-7delT	
		c.*33_63del31	

## References

Cystic Fibrosis Mutation Database: [www.genet.sickkids.on.ca/cftr/app](http://www.genet.sickkids.on.ca/cftr/app)

<sup>1</sup> Only in mutation oligo