

DEVYSER CFTR ITALIA V2

Table of reported insertions and deletions

Italia 1 fragment	CFTR gene location	cDNA	Reported insertion/deletion	Calculated size change
CFTRdele1-wt	Exon 1	-	c.-9_14del23	-23
			c.35_36insTATCA	5
			c.42delA	-1
			c.43delC	-1
			c.43_44insT	1
			c.49_50dupTT	2
			c.50delT	-1
M1	Exon 1	c.1A	None	None
P5	Exon 1	c.14C	c.-9_14del23	-23
CFTR-dele2-wt	-	-	None	None
Dele2ins182-wt	-	-	None	None
Q39	Exon 2	c.115C	c.80delG	-1
			c.105_106insA	1
			c.111_112delAT	-2
			c.112_113delTA	-2
D110 (c.328G)	Exon 4	c.328G	c.343_345del	-3
			c.357delC	-1
			c.379_381dupTTA	3
			c.387delT	-1
			c.393delT	-1
			c.405_406dupAC	2
			c.409_412delCTCC	-4
			c.409delC	-1
			c.412_413insACT	3
			c.414_415insCTA	3
			c.415_416insGA	2
			c.415_416insTA	2
			c.420_421insA	1
			c.424delA	-1
			c.429delT	-1
			c.433delC	-1
			c.442delA	-1
			c.444_445InsCTA	3
			c.459_476delAATAGCTATGTTTAGTTT	-18
			c.473_474insT	1
c.488delA	-1			

Italia 1 fragment	CFTR gene location	cDNA	Reported insertion/deletion	Calculated size change
D110 (c.330C)	Exon 4	c.330C	c.343_345del	-3
			c.357delC	-1
			c.379_381dupTTA	3
			c.387delT	-1
			c.393delT	-1
			c.405_406dupAC	2
			c.409_412delCTCC	-4
			c.409delC	-1
			c.412_413insACT	3
			c.414_415insCTA	3
			c.415_416insGA	2
			c.415_416insTA	2
			c.420_421insA	1
			c.424delA	-1
			c.429delT	-1
			c.433delC	-1
			c.442delA	-1
			c.444_445insCTA	3
			c.459_476delAATAGCTATGTTTAGTTT	-18
			c.473_474insT	1
c.488delA	-1			
G178	Exon 5	c.532G	c.543_546delTAGT	-4
			c.550delC	-1
			c.574_576delGAT	-3
			c.578_579+5delAAGTATG	-7
711+5G	Intron 5	c.579+5G	None	None
852del22-wt	Exon 6	-	c.601delG	-1
			c.642_643insT	1
			c.650_659del10	-10
			c.708delT	-1
			c.714delT	-1
R352	Exon 8	c.1055G	c.980delT	-1
			c.987delA	-1
			c.1006_1007insG	1
			c.1016_1018del	-2
			c.1018delA	-1
			c.1018_1019insTC	2
			c.1008_1019dup12	12
			c.1022_1023insTC	2
			c.1029delC	-1
			c.1029_1030insG	1
c.1053_1054delTC	-2			
1259insA	Intron 8	-	c.1117-30_1117-29delTA	-2
			c.1117-27_1117-26delTA	-2
c.1584+18672A	Intron 11	c.1585-9412A	None	None
S549	Exon 12	c.1645A	c.1610_1611delAC	-2
			c.1617_1618insTA	2
			c.1635_1640del	-6
			c.1642_1643delCT	-2

Italia 1 fragment	CFTR gene location	cDNA	Reported insertion/deletion	Calculated size change
Q552	Exon 12	c.1654C	c.1610_1611delAC	-2
			c.1617_1618insTA	2
			c.1635_1640del	-6
			c.1642_1643delCT	-2
			c.1645_1648delAGTG	-4
			c.1650delA	-1
			c.1652delG	-1
D579	Exon 13	c.1736A	c.1681_1682insC	1
			c.1692delA	-1
			c.1703delT	-1
			c.1713_1714delAG	-2
E585	Exon 13	c.1753G	c.1681_1682insC	1
			c.1692delA	-1
			c.1703delT	-1
			c.1713_1714delAG	-2
			c.1738delG	-1
			c.1739_1740insT	1
1898+3A	Intron 13	c.1766+3A	c.1681_1682insC	1
			c.1692delA	-1
			c.1703delT	-1
			c.1713_1714delAG	-2
			c.1738delG	-1
2184delA-wt	Exon 14	-	c.2061_2062insTTTT	4
			c.2083_2084insG	1
			c.2089_2090insA	1
			c.2157_2163delinsGT	-5
			c.2175_2176insA	1
			c.2203delA	-1
			c.2215delG	-1
			c.2240_2247delCGATACTG	-8
			c.2248_2255del	-8
			c.2276_2277delCC	-2
			c.2277delC	-1
			c.2291delG	-1
			c.2324_2325delAC	-2
			c.2361_2362ins8	8
			c.2380delG	-1
			c.2390_2391insC	1
			c.2424_2425insAT	2
			c.2433_2437delinsATA	-2
			c.2434_2435insT	1
			c.2446delA	-1
c.2453delT	-1			
c.2472delT	-1			
c.2490+2_2490+7delTAGGTA	-6			
c.2290_2230del CGAAGGAGGCA	-11			
Dele17a_18-wt and Dele14b_17b-wt	Intron 19	-	None	None

Italia 1 fragment	CFTR gene location	cDNA	Reported insertion/deletion	Calculated size change
R1066	Exon 20	c.3197G	c.3199_3200delinsA	-1
			c.3229_3230delCT	-2
			c.3263dupA	1
			c.3264delC	-1
			c.3287delT	-1
			c.3291delC	-1
			c.3294delG	-1
			c.3315delG	-1
			c.3331_3332delinsA	-1
			c.3364delA	-1
D1152	Intron 21	c.3454G	c.3468+2_3468+3insT	1
R1158	Exon 22	c.3472C	None	None
G1244	Exon 23	c.3731G	None	None
4016insT-wt	Exon 24	-	c.3874-103delT	-1
			c.3874-61_3874-48del	-14
			c.3874-46_3874-42delTATTT	-5
			c.3874-19_3874-17delCTT	-3
			c.3876delA	-1
			c.3882_3885delTATT	-4
			c.3883delA	-1
CFTRdele22,23-wt	Intron 24	-	None	None
Dele22_24-wt	Intron 24	-	c.3964-3891_3964-3888delTGTT*	-4
G1349	Exon 25	c.4046G	c.4071_4073delTAGinsAA	-1
			c.4077_4080delTGTTinsAA	-2
			c.4086_4087insT	1
			c.4105_4110delinsAGAA	-2
			c.4111_4113dupGAA	3
			c.4127_4131delTGGAT	-5
4382delA-wt	Exon 27	-	c.4296_4297insGA	2

References

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

*External finding reported by customer site