

## DEVYSER CFTR IBERIA

Table of reported insertions and deletions

Iberia 1 fragment	CFTR gene location	cDNA	Reported insertion/deletion	Calculated size change
712-1G	Intron 5	c.580-1G	c.601delG	-1
			c.650_659delAGTTGTTACA	-10
			c.708delIT	-1
			c.714delIT	-1
			c.720_741delAGGGAGAATGATGATG AAGTAC	-22
			c.742_743insTACA	+4
H199	Exon 6	c.595C	c.601delG	-1
			c.650_659delAGTTGTTACA	-10
			c.708delIT	-1
			c.714delIT	-1
			c.720_741delAGGGAGAATGATGATG AAGTAC	-22
			c.742_743insTACA	+4
P205	Exon 6	c.613C	c.650_659delAGTTGTTACA	-10
			c.708delIT	-1
			c.714delIT	-1
			c.720_741delAGGGAGAATGATGATG AAGTAC	-10
			c.742_743insTACA	+4
L206	Exon 6	c.617T	c.650_659delAGTTGTTACA	-10
			c.708delIT	-1
			c.714delIT	-1
			c.720_741delAGGGAGAATGATGATG AAGTAC	-22
			c.742_743insTACA	+4
V232	Exon 6	c.695T	c.708delIT	-1
			c.714delIT	-1
			c.720_741delAGGGAGAATGATGATG AAGTAC	-22
			c.742_743insTACA	+4
1609CA	Exon 11	c.1477_1478CA	c.1469_1470delITC	-2
			c.1469delIT	-1
			c.1444_1445insT	+1
			c.1439delG	-1
			c.1433_1434delICA	-2
			c.1418delG	-1
			c.1408_1417delIATGATTATGG	-10

Iberia 1 fragment	CFTR gene location	cDNA	Reported insertion/deletion	Calculated size change
S549	Exon 12	c.1647T	c.1645_1648delAGTG	-4
			c.1642_1643delCT	-2
			c.1635_1640del	-6
			c.1617_1618insTA	+2
			c.1610_1611delAC	-2
1811+1,6kbA	Intron 12	c.1679+1,6kbA	none	
1812-1G	Intron 12	c.1680-1G	none	
1898+3A	Exon 14	c.1766+3A	c.1681_1682insC	+1
			c.1692delA	-1
			c.1703delT	-1
			c.1713_1714delAG	-2
			c.1738delG	-1
			c.1739_1740insT	+1
R709	Exon 14	c.2125C	c.2157_2163delinsGT	-4
			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.2453delT	-1
			c.2472delT	-1
2869	Exon 17	c.2737	c.2757_2758insATTAT*	+5
			c.2764_2765insAG	+2
			c.2775_2776delTT	-2
			c.2777delT	-1
			c.2789delG	-1
			c.2803_2813delCTACCACTGGT	-11
			c.2805_2810delinsTCAGA	-1
			c.2810_2811insT	+1
			c.2816_2817delATinsC	-1
			c.2825delT	-1
			c.2859_2890delACATTCTGTTCTTCAA GCACCTATGTCAACCC	-32
			c.2875delG	-1
			c.2896delA	-1

Iberia 1 fragment	CFTR gene location	cDNA	Reported insertion/deletion	Calculated size change
Q890	Exon 17	c.2668C	c.2686_2687insT	+1
			c.2687_2690delCTCAinsTGAGTACTATGAG	+9
			c.2705delG	-1
			c.2737_2738insG	+1
			c.2757_2758insATTAT*	+5
			c.2764_2765insAG	+2
			c.2775_2776delTT	-2
			c.2777delT	-1
			c.2789delG	-1
			c.2803_2813delCTACCACTGGT	-11
			c.2805_2810delinsTCAGA	-1
			c.2810_2811insT	+1
			c.2816_2817delATinsC	-1
			c.2825delT	-1
			c.2859_2890delACATTCTGTTCTTCAAGCACCTATGTCAACCC	-32
c.2875delG	-1			
c.2896delA	-1			
Q1100	Exon 20	c.3299A	c.3229_3230delCT	-2
			c.3263dupA	+1
			c.3264delC	-1
			c.3287delT	-1
			c.3291delC	-1
			c.3294delG	-1
M1101	Exon 20	c.3302T	c.3229_3230delCT	-2
			c.3263dupA	+1
			c.3264delC	-1
			c.3287delT	-1
			c.3291delC	-1

## References

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

\*External finding reported from customer site