

DEVYSER CFTR UK

Table of reported insertions and deletions

UK 1 fragment	CFTR gene location	cDNA	Reported insertion/deletion	Calculated size change
E60	Exon 3	c.178G	c.198_202delTCCTA	-5
			c.217delC	-1
			c.227_228insT	+1
			c.228_229insT	+1
			c.233delT	-1
			c.233_234insT	+1
			c.234delC	-1
			c.247_248insT	+1
			c.262_263delTT	-2
			c.264_268delATATT	-5
P67	Exon 3	c.200C	c.217delC	-1
			c.227_228insT	+1
			c.228_229insT	+1
			c.233delT	-1
			c.233_234insT	+1
			c.234delC	-1
			c.247_248insT	+1
			c.262_263delTT	-2
c.264_268delATATT	-5			
Q98	Exon 4	c.292c	None	None
621+2T	Intron 4	c.489+2T	None	None
L218	Exon 6	c.653T	c.708delT	-1
			c.714delT	-1
			c.720_741delIAGGGAGAATGATG ATGAAGTAC	-22
			c.742_743insTACA	+4
1161C	Exon 8	c.1029C	None	None
1154-wt	Exon 8	c.1022_1023	c.1029delC	-1
			c.1029_1030insG	+1
			c.1053_1054delTC	-2
			c.1069delG	-1
			c.1081delT	-1
			c.1083delG	-1
			c.1093_1094delCT	-2
c.1111_1112ins6	+6			
1525-1G	Intron 10	c.1393-1G	None	None
Q493	Exon 11	c.1477C	None	None
V520	Exon 11	c.1558G	None	None
S549	Exon 12	c.1646G	None	None
Y569	Exon 13	c.1705T	None	None
S589	Exon 13	c.1766G	None	None

UK 1 fragment	CFTR gene location	cDNA	Reported insertion/deletion	Calculated size change
1898+5G	Intron 13	c.1766+5G	None	None
D1152	Exon 21	c.3454G	None	None
R1158	Exon 22	c.3472C	None	None
R1283	Exon 22	c.3848G	None	None

References

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