

DEVYSER CFTR CORE

Table of reported insertions and deletions

Core 1 fragment	CFTR gene location	cDNA	Reported insertion/deletion	Calculated size change
G85	Exon 3	c.254G	c.262_263delTT	-2
			c.264_268delATATT	-5
CFTRdele2,3_wt	Intron 1 - Exon 3	c.273+10250_10251TT	c.273+10255delC	-1
R117	Exon 4	c.349C and c.350G	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
621+1G	Intron 4	c.489+1G	None	None
711+1G	Intron 5	c.579+1G	None	None
1078	Exon 8	c.948T	c.1006_1007insG	1
			c.1016_1018del	-3
			c.1018_1019insTC	2
			c.1018delA	-1
			c.1022_1023insTC	2
			c.1029_1030insG	1
			c.1029delC	-1
			c.1053_1054delTC	-2
			c.1069delG	-1
			c.1081delT	-1
			c.1083delG	-1
			c.1093_1094delCT	-2
			c.1111_1112ins6	6
			c.1111dupA	1
			c.980delT	-1
			c.987delA	-1

Core 1 fragment	CFTR gene location	cDNA	Reported insertion/deletion	Calculated size change
R334	Exon 8	c.1000C	c.1006_1007insG	1
			c.1016_1018del	-3
			c.1018_1019insTC	2
			c.1018delA	-1
			c.1022_1023insTC	2
			c.1029_1030insG	1
			c.1029delC	-1
			c.1053_1054delTC	-2
			c.1069delG	-1
			c.1081delT	-1
			c.1083delG	-1
			c.1093_1094delCT	-2
			c.1111_1112ins6	6
			c.1111dupA	1
I336	Exon 8	c.1007T	c.1016_1018del	-3
			c.1018_1019insTC	2
			c.1018delA	-1
			c.1022_1023insTC	2
			c.1029_1030insG	1
			c.1029delC	-1
			c.1053_1054delTC	-2
			c.1069delG	-1
			c.1081delT	-1
			c.1083delG	-1
			c.1093_1094delCT	-2
			c.1111_1112ins6	6
			c.1111dupA	1
			T338	Exon 8
c.1018_1019insTC	2			
c.1018delA	-1			
c.1022_1023insTC	2			
c.1029_1030insG	1			
c.1029delC	-1			
c.1053_1054delTC	-2			
c.1069delG	-1			
c.1081delT	-1			
c.1083delG	-1			
c.1093_1094delCT	-2			
c.1111_1112ins6	6			
c.1111dupA	1			
R347	Exon 8	c.1040G		
			c.1069delG	-1
			c.1081delT	-1
			c.1083delG	-1
			c.1093_1094delCT	-2
			c.1111_1112ins6	6
			c.1111dupA	1

Core 1 fragment	CFTR gene location	cDNA	Reported insertion/deletion	Calculated size change
I507	Exon 11	c.1519_1521ATC	c.1408_1417delATGATTATGG	-10
			c.1418delG	-1
			c.1433_1434delCA	-2
			c.1439delG	-1
			c.1444_1445insT	1
			c.1465_1468delTCAT	-4
			c.1469_1470delTC	-2
			c.1469delT	-1
			c.1477_1478delCA	-2
			c.1482_1483delTT	-2
			F508	Exon 11
c.1418delG	-1			
c.1433_1434delCA	-2			
c.1439delG	-1			
c.1444_1445insT	1			
c.1465_1468delTCAT	-4			
c.1469_1470delTC	-2			
c.1469delT	-1			
c.1477_1478delCA	-2			
c.1482_1483delTT	-2			
c.1519_1521delATC	-3			
1677	Exon 11	c.1545_1546TA	c.1408_1417delATGATTATGG	-10
			c.1418delG	-1
			c.1433_1434delCA	-2
			c.1439delG	-1
			c.1444_1445insT	1
			c.1465_1468delTCAT	-4
			c.1469_1470delTC	-2
			c.1469delT	-1
			c.1477_1478delCA	-2
			c.1482_1483delTT	-2
			c.1519_1521delATC	-3
c.1521_1523delCTT	-3			
c.1528delG	-1			
1717-1G	Intron 11	c.1585-1G	None	None
G542	Exon 12	c.1624G	c.1610_1611delAC	-2
			c.1617_1618insTA	2
G551	Exon 12	c.1652G	c.1610_1611delAC	-2
			c.1617_1618insTA	2
			c.1635_1640del	-6
			c.1642_1643delCT	-2
			c.1645_1648delAGTG	-4
			c.1650delA	-1
R553	Exon 12	c.1657C	c.1652delG	-1
			c.1610_1611delAC	-2
			c.1617_1618insTA	2
			c.1635_1640del	-6
			c.1642_1643delCT	-2
			c.1645_1648delAGTG	-4
			c.1650delA	-1
c.1656delA	-1			

Core 1 fragment	CFTR gene location	cDNA	Reported insertion/deletion	Calculated size change
R560	Exon 12	c.1679G	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
			c.1656delA	-1
			c.1660_1661insA	1
			c.1670delC	-1
			c.1674delA	-1
1898+1G	Intron 13	c.1766+1G	c.1681_1682insC	1
			c.1692delA	-1
			c.1703delT	-1
			c.1713_1714delAG	-2
			c.1738delG	-1
			c.1739_1740insT	1
2143	Exon 14	c.2012T	c.2052_2053insA	1
			c.2052dupA	1
			c.2013_2015delAGA	-3
			c.2044_2045insC	1
			c.2044delA	-1
			c.2051_2052delAA	-2
			c.2051_2052delAAinsG	-1
			c.2052delA	-1
			c.2053_2054insC	1
			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_2255delCCTCGCAT	-8
			c.2276_2277delCC	-2
			c.2277delC	-1
			c.2290delCGAAGGAGGCA	-11
			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			

Core 1 fragment	CFTR gene location	cDNA	Reported insertion/deletion	Calculated size change
2183_2184AA	Exon 14	c.2051_2052AA	c.2053_2054insC	1
			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_2255delCCTCGCAT	-8
			c.2276_2277delCC	-2
			c.2277delC	-1
			c.2290delCGAAGGAGGCA	-11
			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
			2789+5G	Intron 16
3120+1G	Intron 18	c.2988+1G	c.2947_2948delTT	-2
			c.2968_2969insA	1
3272-26A	Intron 19	c.3140-26A	c.3161delA	-1
			c.3180delA	-1
			c.3188_3189insCTATG	5
			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
			c.3367+24_c.3367+27delGCTA*	-4
			c.3367+26_c.3367+30del*	-4
			L1065	Exon 20
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			
c.3367+24_c.3367+27delGCTA*	-4			
c.3367+26_c.3367+30del*	-4			

Core 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
			c.3367+24_c.3367+27delGCTA*	-4
			c.3367+26_c.3367+30del*	-4
L1077	Exon 20	c.3230T	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
			c.3367+24_c.3367+27delGCTA*	-4
			c.3367+26_c.3367+30del*	-4
Y1092	Exon 20	c.3276C	c.3287delT	-1
			c.3291delC	-1
			c.3294delG	-1
			c.3315delG	-1
			c.3331_3332delinsA	-1
			c.3364delA	-1
			c.3367+24_c.3367+27delGCTA	-4
			c.3367+26_c.3367+30del	-4
R1162	Exon 22	c.3484C	c.3490_3491insT	-1
			c.3659delC	-1
			c.3485_3486delGA	-2
			c.3497delT	-1
			c.3528delC	-1
			c.3530delA	-1
			c.3535_3536insTCAA	4
			c.3535_3538delACCA	-4
			c.3540delA	-1
			c.3592delG	-1
			c.3598delAinsTCT	2
			c.3600delA	-1
			c.3605delA	-1
			c.3618_3619delAG	-2
			c.3623delG	-1
			c.3657_3658insA	1
c.3691delT	-1			
c.3708delT	-1			

Core 1 fragment	CFTR gene location	cDNA	Reported insertion/deletion	Calculated size change
3659	Exon 22	c.3528C	c.3530delA	-1
			c.3535_3536insTCAA	4
			c.3535_3538delACCA	-4
			c.3540delA	-1
			c.3592delG	-1
			c.3598delAinsTCT	2
			c.3600delA	-1
			c.3605delA	-1
			c.3618_3619delAG	-2
			c.3623delG	-1
			c.3657_3658insA	1
			c.3659delC	-1
			c.3691delT	-1
			c.3708delT	-1
3849+10kbC	Intron 22	c.3717+12191C	None	None
W1282	Exon 23	c.3846G	c.3855delC	-1
			c.3873+121delTT	-2
N1303	Exon 24	c.3909C	c.3874-103delT	-1
			c.3874-19_3874-17delCTT	-3
			c.3874-46_3874-42delTATTT	-5
			c.3874-61_3874-48del	-14
			c.3876delA	-1
			c.3882_3885delTATT	-4
			c.3883delA	-1
			c.3884_3885insT	1
			c.3890_3891insT	1
			c.3895delA	-1
			c.3908delA	-1
		c.3908dupA	1	

References

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

*External finding reported by customer site