

## ACMG CF Mutation Panel - HGVS Nomenclature

<u>cDNA Sequence Change Nomenclature</u>	<u>Amini Acid Change Nomenclature</u>	<u>Colloquial Name</u>
c.1521_1523delCTT/	p.Phe508del	F508del
c.2988+1G>T (AJ575003.1:g.305G>T)		3120+G>T
c.489+1G>T (AJ574942.1:g.240G>T)		621+1G>T
c.350G>A	p.Arg117His	R117H
c.2052delA	p.Lys684AsnfsX38	2184delA
c.1766+1G>A (AJ574983.1:g.179G>A)		1898+1G>A
c.1519_1521delATC	p.Ile507del	dell507
c.1624G>T	p.Gly542X	G542X
c.1657C>T	p.Arg553X	R553X
c.1652G>A	p.Gly551Asp	G551D
c.2657+5G>A (AJ574995.1:g.216G>A)		2789+5G>A
c.3846G>A	p.Trp1282X	W1282X
c.3718-2477C>T (AY848832.1:g.40725C>T)		3849+10C>T
c.579+1G>T (AJ574943.1:g.261g>T)		711+1G>T
c.3484C>T	p.Arg1162X	R1162X
c.1585-1G>A (AJ574980.1:g.116G>A)		1717-1G>A
c.3909C>G	p.Asn1303Lys	N1303K
c.1040G>C	p.Arg347Pro	R347P
c.1000C>T	p.Arg334Trp	R334W
c.3437delC	p.Ala1146ValfsX2	3659delC
c.1364C>A	p.Ala455Glu	A455E
c.1679G>C	p.Arg560Thr	R560T
c.254G>A	p.Gly85Glu	G85E

## Other CF Mutations Nomenclature

<u>cDNA Sequence Change Nomenclature</u>	<u>Amini Acid Change Nomenclature</u>	<u>Colloquial Name</u>
c.3302T>A	p.Met1101Lys	M1101K
c.3277C>A	p.Tyr1092X	Y1092XC>A
c.1721C>A	p.Pro574His	P574H
c.532G>C	p.Gly178Arg	G178R
c.312delA	p.Arg104ArgfsX3	444delA
c.1680-1G>A (AJ574983.1:g.92G>A)		1812-1G>A
c.1040G>A	p.Arg347His	R347H
c.3744delA	p.Ser1248SerfsX11	3876delA
c.1647G>A	p.Ser549Asn	S549N
c.1646A>C	p.Ser549Arg	S549R
c.3773_3774insT	p.Leu1258PhefsX7	3905insT
c.1519A>G	p.Ile507Val	I507V
c.223C>T	p.Arg75X	R75X
c.178G>T	p.Glu60X	E60X
c.1652G>A	p.Gly551Asp	G551D
c.1516A>G	p.Ile506Val	I506V
c.3454G>C	p.Asp1152His	D1152H
c.3140-26A>G (AJ575014.1:g.322A>G)		3272-26A>G
c.366G>A	p.Tyr122X	Y122X
c.2047AA>G	p.Lys684SerfsX38	2183AA>G
c.2011delT	p.Leu671fsX1	2143delT
c.948delT	p.Phe316LeufsX12	1078delT
c.262delTT	p.Leu88IlefsX22	394delTT
c.443T>C	p.Ile148Thr	I148T
c.3703A>C	p.Ser1235Arg	S1235R
c.1523T>G	p.Phe508Cys	F508C
c.1558G>T	p.Val520Phe	V520F
c.1477C>T	p.Gln493X	Q493X
c.4046G>A	p.Gly1349Asp	G1349D

last updated 2-22-2008