

Supplemental Table 1: CFTR mutation nomenclature (HG Variation Society guidelines and Legacy name)

HG Variation Society guidelines		Legacy name
Nucleotidic notation	Aminoacidic notation	
c.254G>A	p.Gly85Glu	G85E
c.350G>A	p.Arg117His	R117H
c.443T>C	p.Ile148Thr	I148T
c.489+1G>T		621+1G>T
c.579+1G>T		711+1G>T
c.837A>T	p.Glu279Asp	E279D
c.948delT	p.Phe316LeufsX12	1078delT
c.1000C>T	p.Arg334Trp	R334W
c.1521_1523delCTT	p.Phe508del	F508del; [delta]F508
c.1624G>T	p.Gly542*	G542X
c.1657C>T	p.Arg553*	R553X
c.1736A>G	p.Asp579Gly	D579G
c.2051_2052delAAinsG	p.Lys684SerfsX38	2183AA>G
c.2810_2811insT	p.Val938GlyfsX37	2942insT
c.2991G>C	p.Leu997Phe	L997F
c.3454G>C	p.Asp1152His	D1152H
c.3617C>G	p.Ser1206*	S1206X(C>G)
c.3808G>A	p.Asp1270Asn	D1270N
c.3884_3885insT	p.Ser1297PhefsX5	4016insT
c.3909C>G	p.Asn1303Lys	N1303K
c.[443T>C;3607_3072delATAGTG]	p.[Ile148Thr;Ile1023_Val1024del]	[I148T;3199del6]
c.[220C>T;601G>A;3808G>A]	p.[Arg74Trp;Val201Met;Asp1270Asn]	[R74W;V201M;D1270N]

c.[1210-34TG[12];1210-12T[5];2930C>T]		[(TG) ₁₂ T ₅ ;S977F]
c.[1210-34TG[12];1210-12T[5]]		(TG) ₁₂ T ₅
c.[220C>T;3808G>A]	p.[Arg74Trp;Asp1270Asn]	[R74W;D1270N]
c.[350G>T;2991G>C]	p.[Arg117Leu;Leu997Phe]	[R117L;L997F]