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Cystic Fibrosis Mutation Nomenclature

The mutation nomenclature for *CFTR* mutations has been changed to comply with guidelines from the Human Genome Variation Society (www.hgvs.org). These state that nucleotide number 1 should correspond to the A of the ATG translation initiation codon (previously the A of the ATG was numbered 133). The description of all variants is preceded by a letter indicating the type of reference sequence used; 'c.' relates to a coding DNA sequence and 'p.' a protein sequence.

The table below details the old and new nomenclature for the 23 *CFTR* mutations identified by the current test kit.

Old Nomenclature	New nomenclature	
	DNA level	Protein Level
ΔF508	c.1521_1523delCTT	p.Phe508del
ΔI507	c.1519_1521delATC	p.lle507del
R117H	c.350G>A	p.Arg117His
G542X	c.1624G>T	p.Gly542X
G551D	c.1652G>A	p.Gly551Asp
R553X	c.1657C>T	p.Arg553X
R560T	c.1679G>C	p.Arg560Thr
3659delC	c.3528delC	p.Thr1176fs
W1282X	c.3846G>A	p.Trp1282X
N1303K	c.3909C>G	p.Asn1303Lys
G85E	c.254G>A	p.Gly85Glu
A455E	c.1364C>A	p.Ala455Glu
R334W	c.1000C>T	p.Arg334Trp
R347P	c.1040G>C	p.Arg347Pro
1717-1G>A	c.1585-1G>A	-
621+1G>T	c.489+1G>T	-
3849+10kbC>T	c.3717+10kbC>T	-
711+1G>T	c.579+1G>T	-
1898+1G>A	c.1766+1G>A	-
2789+5G>A	c.2657+5G>A	-
R1162X	c.3484C>T	p.Arg1162X
3120+1G>A	c.2988+1G>A	-
2184delA	c.2052delA	p.Lys684fs

Reference sequence accession number: NM_000492.3