

Supplementary Table 3a: Variants previously reported to alter splicing of the CFTR gene as established by functional studies.

HGVS nucleotide nomenclature name	Legacy name	Quantity of normal CFTR mRNA (% of WT)	RNA source	Reference
c.1679+1.6kbA>G	1811+1.6kbA>G	1-3	Nasal epithelium	17, 18
c.1766+3A>G	1898+3A>G	0	Minigene construct	18
c.2657+5G>A	2789+5G>A	4	Nasal epithelium	19
c.3140-26A>G	3272-26A>G	5	Nasal epithelium	20, 21
c.3717+12191C>T	3849+10kbC>T	8	Nasal epithelium	8

Supplementary Table 3b: Comparison of CFTR RNA splicing variants studied *in vitro* by minigene with *in vivo* results.

HGVS nucleotide nomenclature name	Legacy name	CFTR Minigene analysis		CFTR <i>in vivo</i> analysis	Reference
		Transcript	Protein (band C)	Transcript	
c.579+1G>T	711+1G>T	Exon 5 skipping	Not detected	Exon 5 skipping	23, 24
c.1585-1G>A	1717-1G>A	(i) Exon 11 skipping (ii) c.1717delG	Not detected	Exon 11 skipping	25
c.2657+5G>A	2789+5G>A	(i) WT transcript (ii) Exon 14b skipping	<10% of wild type	(i) WT transcript (ii) Exon 14b skipping	19

Supplementary Table 3c: Relative amount of properly spliced RNA transcript and fully processed protein generated by CFTR minigenes transfected into two human cell lines.

HGVS nucleotide nomenclature name	Legacy name	Correctly spliced CFTR transcript (% of respective WT minigene \pm S.D.)		Processed CFTR protein C-band (% of respective WT minigene \pm S.D.)	
		HEK	CFBE41o-	HEK	CFBE41o-
c.579+3A>G	711+3A>G	0	ND	0	ND
c.579+5G>A	711+5G>A	0	ND	0	ND
c.1585-8G>A	1717-8G>A	0	0	0	0
c.2657+2_2657+3insA	2789+2insA	71 \pm 7.3	80 \pm 8.3	76 \pm 10.7	84 \pm 6.3
c.2988G>A	3120G>A	0	0	0	0

ND indicates not done.