Table 1.S: Analysis of CFTR Missense Mutations

	CFTR2 variant annotation (as of 31 July 2020)	n alleles in CFTR2	Sweat chloride, mEq/L[a]	% Patients who are PI[a,b]
Non-CF	-	-	Less than 30	Less than 1%
P67L	CF-causing	239	60	33
G85E	CF-causing	610	100	86
E92K	CF-causing	38	89	54
R117H;9T[c]	-	-	-	-
R117H;7T	Varying clinical consequences	125	43	15
R117H;5T	CF-causing	102	74	31
R334W	CF-causing	425	99	40
R347P	CF-causing	528	100	67
A455E	CF-causing	496	84	34
S492F	CF-causing	24	79	48
V520F	CF-causing	156	105	96
R560K	CF-causing	15	110	100
R560T	CF-causing	343	103	98
S549R	CF-causing	93	105	98
D614G	Varying clinical consequences	17	60	29
L1065P	CF-causing	43	103	79
R1066C	CF-causing	220	104	98
L1077P	CF-causing	96	101	94
M1101K	CF-causing	238	106	70
N1303K	CF-causing	2226	105	98

[a]Clinical data shown from individuals in CFTR2 who have variant of interest and a PI-CF-causing variant

[b]PI; pancreatic insufficient

[c]Not annotated on cftr2.org