

Supplemental Table 2: Synopsis of CF phenotypic expression in patients bearing different CFTR genotypes. CFTR-RD: CFTR related disorders; PS: pancreatic sufficiency; PI: pancreatic insufficiency

CFTR genotype	N	Phenotypic expression
p.[Arg74Trp;Val201Met;Asp1270Asn];[class I-II mutation]	5	CF with PS (4 cases); CFTR-RD (1 case)
p.[Arg74Trp;Val201Met;Asp1270Asn];[class IV-V mutation]	3	CFTR-RD (3 cases)
p.[Arg74Trp;Asp1270Asn];[class IV-V mutation]	1	CFTR-RD
p.[Arg74Trp;Asp1270Asn];[class I-II mutation]	1	Healthy
p.[Asp1270Asn];[class I-II mutation]	2	Healthy (2 cases)
p.[Ile148Thr;Ile1023_Val1024del];[class I-II mutation]	5	CF with PI (5 cases)
p.[Ile148Thr];[class I-II mutation]	4	Healthy (4 cases)

p.[Arg117Leu;Leu997Phe];p.[Arg117Leu;Leu997Phe]	2	CF with PS (1 case); CFTR-RD (1 case)
p.[Arg117Leu;Leu997Phe];[class I-II mutation]	4	CF with PS (4 cases)
p.[Leu997Phe];[p.Leu997Phe]	2	CFTR-RD (1 case); healthy (1 case)
p.[Leu997Phe];[class I-II mutation]	16	CFTR-RD (8 cases); healthy (8 cases)
p.[Leu997Phe];[class IV-V mutation]	16	CFTR-RD (6 cases); healthy (10 cases)
c.[1210-34TG[12];1210-12T[5];2930C>T];[class I-II mutation]	3	CF (1 case); CFTR-RD (2 cases)