Table 7. Mutations in genes of CF airways isolates from 29 different patients

Mutated gene†	Nucleotide position mutated in gene	Type of mutation	Nucleotide(s) prior to mutation	Nucleotide(s) after mutation	Amino acid prior to mutation	Amino acid after mutation	Patient no.	Mutated isolate(s), named by patient age (yr) when isolate was collected	Isolates not containing the mutation, named by patient age (yr) when isolate was collected‡	Computational prediction of effect on protein function§
transcriptional regulator *mexZ* (PA2020)										
	26	nonsynonymous	C	T	S	F	10	15.4A, 15.4B	6.7	Not tolerated
	60	18bp deletion	CGAGCGGGTTTTCCTGGA	-	-	-	18	11.8, 18.4	4.2, 9.0	-
	74	nonsynonymous	T	C	L	P	19	22.5	7.2, 16.4	Not tolerated
	137	nonsynonymous	G	T	G	V	14	13.1B	4.3, 13.1A	Not tolerated
	139	nonsynonymous	G	C	A	P	8	13.3A	1.1	Not tolerated
	145	3bp deletion	TAC	-	-	-	9	19.7A, 19.7B	9.2	-
	231	14bp deletion	CCAGGGTGCCGGCGC	-	-	-	11	15.3	7.3, 13.7	-
	256–319	64bp deletion	-	-	-	-	24	22.5	8.5, 17.4	-
	308	1bp deletion	A	-	-	-	15	12.8A, 12.8B	5.7	-
	315	5bp deletion	G	-	-	-	25	14.5	1.2, 21.0	-
	337	13bp deletion	ACTTGAGGTAGAG	-	-	-	22	20.0	9.9	-

Mutated gene†	Nucleotide position mutated in gene	Type of mutation	Nucleotide(s) prior to mutation	Nucleotide(s) after mutation	Amino acid prior to mutation	Amino acid after mutation	Patient no.	Mutated isolate(s), named by patient age (yr) when isolate was collected	Isolates not containing the mutation, named by patient age (yr) when isolate was collected‡	Computational prediction of effect on protein function§
	352	13bp deletion	CGTCGCTGC GTTC	-	-	-	25	21.0	1.2, 14.5	-
	431	1bp deletion	C	-	-	-	14	13.1B	4.3, 13.1A	-
	452	nonsynonymous	C	T	P	L	16	17.6	7.1	Not tolerated
	461	nonsynonymous	T	C	L	P	20	17.3	9.9	Not tolerated
	490	nonsynonymous	C	T	Q	STOP	21	19.5	0.5, 10.7, 14.3, 15.2	-
	494	nonsynonymous	C	A	S	STOP	17	17.7	6.6, 22.2	-
	517	1bp deletion	A	-	-	-	28	14.8	8.2, 17.9	-
	527	nonsynonymous	G	A	W	STOP	21	10.7, 14.3, 15.2	0.5, 19.5	-
	532	nonsynonymous	G	T	E	STOP	17	22.2	6.6, 17.7	-
	555	nonsynonymous	G	A	W	STOP	23	14.0	6.2, 10.0	-
	605	nonsynonymous	C	T	S	F	13	19.6A, 19.6B	10.8	Not tolerated
	609	1bp deletion	C	-	-	-	14	13.1A	4.3, 13.1B	-
	631	nonsynonymous	T	G	STOP	G	13	19.6A, 19.6B	10.8	None
transcriptional regulator *lasR* (PA1430)										

Mutated gene†	Nucleotide position mutated in gene	Type of mutation	Nucleotide(s) prior to mutation	Nucleotide(s) after mutation	Amino acid prior to mutation	Amino acid after mutation	Patient no.	Mutated isolate(s), named by patient age (yr) when isolate was collected	Isolates not containing the mutation, named by patient age (yr) when isolate was collected‡	Computational prediction of effect on protein function§
	97	11bp deletion	TCGAAGATCCT	-	-	-	20	17.5	-	-
	179	nonsynonymous	G	A	W	STOP	3	7.3	0.8, 6.8	-
	181	nonsynonymous	C	T	R	C	12	21.1B	8.6, 21.1A	Not tolerated
	194	nonsynonymous	A	G	D	G	17	17.7, 22.2	6.6	Not tolerated
	254	nonsynonymous	C	T	P	L	30	18.5	10.2, 16.8	Not tolerated
	292	nonsynonymous	C	T	Q	STOP	10	15.4A, 15.4B	6.7	-
	316	nonsynonymous	T	C	S	P	28	14.8, 17.9	8.2	Tolerated
	341	nonsynonymous	T	C	L	P	21	15.2	0.5, 10.7, 14.3, 19.5	Not tolerated
	344	nonsynonymous	C	T	T	I	8	13.3A	1.1, 13.3B	Tolerated
	350	nonsynonymous	C	T	P	L	23	14.0	6.2, 10.0	Not tolerated
	355	2bp insertion	CA	CACA	-	-	13	19.6A, 19.6B	10.8	-
	362	nonsynonymous	C	A	A	D	17	22.2	6.6, 17.7	Not tolerated
	455	1bp deletion	G	-	-	-	6	9.6B	1.0, 9.6A	-
	467	nonsynonymous	A	G	D	G	30	18.5	10.2, 16.8	Not tolerated
	532	nonsynonymous	A	G	T	A	21	19.5	0.5, 10.7, 14.3, 15.2	Not tolerated

Mutated gene†	Nucleotide position mutated in gene	Type of mutation	Nucleotide(s) prior to mutation	Nucleotide(s) after mutation	Amino acid prior to mutation	Amino acid after mutation	Patient no.	Mutated isolate(s), named by patient age (yr) when isolate was collected	Isolates not containing the mutation, named by patient age (yr) when isolate was collected‡	Computational prediction of effect on protein function§
	538	nonsynonymous	C	T	R	W	15	12.8A, 12.8B	5.7	Not tolerated
	561	nonsynonymous	G	A	W	STOP	27	6.4	9.1, 11.4	-
	578	nonsynonymous	C	T	T	I	9	19.7A, 19.7B	9.2	Not tolerated
	608	nonsynonymous	G	A	C	Y	24	22.5	8.5, 17.4	Not tolerated
	665	nonsynonymous	C	T	T	I	22	20.0	9.9	Tolerated
	678	IS insertion	-	-	-	-	20	9.9	-	-
	691	nonsynonymous	G	C	A	P	26	8.6	2.9, 13.3	Not tolerated
probable permease of ABC transporter (PA0313)										
	259	nonsynonymous	G	A	G	R	6	9.6B	1.0, 9.6A	Not tolerated
	353	nonsynonymous	G	A	R	H	29	13.5, 14.0	5.6	Not tolerated
	384	1bp deletion	A	-	Q	-	7	15.4A	7.8, 15.4B	-
	417	nonsynonymous	G	A	W	STOP	21	15.2	0.6, 10.7, 14.3, 19.5	-
	419	1bp insertion	GGGG	GGGGG	-	-	25	14.5	1.2, 21.0	-
	483	9bp insertion	CAGCCTGAT CAGCCTGAT	CAGCCTGAT CAGCCTGAT	-	-	8	13.3A, 13.3B	1.1	-

Mutated gene†	**Nucleotide position mutated in gene**	**Type of mutation**	**Nucleotide(s) prior to mutation**	**Nucleotide(s) after mutation**	**Amino acid prior to mutation**	**Amino acid after mutation**	**Patient no.**	**Mutated isolate(s), named by patient age (yr) when isolate was collected**	**Isolates not containing the mutation, named by patient age (yr) when isolate was collected[‡]**	**Computational prediction of effect on protein function[§]**
				CAGCCTGAT						
	483	9bp insertion	CAGCCTGAT CAGCCTGAT	CAGCCTGAT CAGCCTGAT CAGCCTGAT	-	-	9	19.7A, 19.7B	9.2	-
	483	9bp insertion	CAGCCTGAT CAGCCTGAT	CAGCCTGAT CAGCCTGAT CAGCCTGAT	-	-	11	13.7	7.3, 15.3	-
	483	9bp insertion	CAGCCTGAT CAGCCTGAT	CAGCCTGAT CAGCCTGAT CAGCCTGAT	-	-	13	19.6A, 19.6B	10.8	-
	483	9bp insertion	CAGCCTGAT CAGCCTGAT	CAGCCTGAT CAGCCTGAT CAGCCTGAT	-	-	15	12.8A, 12.8B	5.7	-
	483	9bp insertion	CAGCCTGAT CAGCCTGAT	CAGCCTGAT CAGCCTGAT CAGCCTGAT	-	-	25	21.0	1.2, 14.5	-
	483	9bp deletion	CAGCCTGAT CAGCCTGAT	CAGCCTGAT	-	-	21	14.3	0.6, 10.7, 15.2, 19.5	-
	483	9bp insertion	CAGCCTGAT CAGCCTGAT	CAGCCTGAT CAGCCTGAT CAGCCTGAT	-	-	30	16.8, 18.5	10.2	-

Mutated gene†	Nucleotide position mutated in gene	Type of mutation	Nucleotide(s) prior to mutation	Nucleotide(s) after mutation	Amino acid prior to mutation	Amino acid after mutation	Patient no.	Mutated isolate(s), named by patient age (yr) when isolate was collected	Isolates not containing the mutation, named by patient age (yr) when isolate was collected[‡]	Computational prediction of effect on protein function[§]
	538	nonsynonymous	G	A	E	K	18	11.8	4.2, 9.0, 18.4	Not tolerated
RND multidrug efflux membrane fusion protein *mexA* (PA0425)										
	352	nonsynonymous	C	T	Q	STOP	21	15.2	0.6, 10.7, 14.3	-
	379	nonsynonymous	C	T	Q	STOP	20	9.9	17.3	-
	479	nonsynonymous	T	G	V	G	9	19.7A, 19.7B	9.2	Not tolerated
	520	nonsynonymous	A	G	T	A	8	13.3A	1.1, 13.3B	Not tolerated
	658	nonsynonymous	G	C	E	Q	14	13.1A, 13.1B	4.3	Tolerated
	658	nonsynonymous	G	T	E	STOP	14	4.3	13.1A, 13.1B	-
	658	nonsynonymous	G	T	E	STOP	12	8.6, 21.1A, 21.1B	-	-
	768	1bp deletion	C	-	-	-	19	16.4, 22.5	7.2	-
	800	nonsynonymous	A	G	N	S	23	6.2	10.0, 14.0	Not tolerated
	812	nonsynonymous	A	G	E	G	23	6.2	10.0, 14.0	Tolerated
	824	nonsynonymous	G	C	G	A	15	12.8A, 12.8B	5.7	Not tolerated
	838	nonsynonymous	G	C	A	P	20	17.3	9.9	Not tolerated
	878	nonsynonymous	C	A	A	D	17	22.2	6.6, 17.7	Not tolerated

Mutated gene†	Nucleotide position mutated in gene	Type of mutation	Nucleotide(s) prior to mutation	Nucleotide(s) after mutation	Amino acid prior to mutation	Amino acid after mutation	Patient no.	Mutated isolate(s), named by patient age (yr) when isolate was collected	Isolates not containing the mutation, named by patient age (yr) when isolate was collected[‡]	Computational prediction of effect on protein function[§]
	1046	nonsynonymous	T	C	L	P	30	18.5	10.2, 16.8	Not tolerated
biotin carboxylase *accC* (PA4848)										
	2	nonsynonymous	T	A	M	K	28	14.8, 17.9	8.2	Not tolerated
	3	nonsynonymous	G	A	M	I	27	9.1, 11.4	6.4	Not tolerated
	395	nonsynonymous	C	T	P	L	6	9.6A	1.0, 9.6B	Not tolerated
	494	3bp insertion	CGGCGGCGGCGG	CGGCGGCGGCGGCGG	-	-	13	19.6A, 19.6B	10.8	-
	818	nonsynonymous	G	A	G	D	18	9.0	4.2, 11.8, 18.4	Not tolerated
	845	nonsynonymous	G	A	G	D	23	6.2	10.0, 14.0	Tolerated
	1159	nonsynonymous	A	G	K	E	25	14.5, 21.0	1.2	Not tolerated
	1331	IS insertion	-	-	-	-	20	9.9, 17.3	-	-
	1331	IS insertion	-	-	-	-	30	16.8	10.2, 18.5	-
*transcriptional regulator *vfr* (PA0652)										

Mutated gene†	Nucleotide position mutated in gene	Type of mutation	Nucleotide(s) prior to mutation	Nucleotide(s) after mutation	Amino acid prior to mutation	Amino acid after mutation	Patient no.	Mutated isolate(s), named by patient age (yr) when isolate was collected	Isolates not containing the mutation, named by patient age (yr) when isolate was collected‡	Computational prediction of effect on protein function§
	224	nonsynonymous	T	A	L	Q	23	10.0, 14.0	6.2	Not tolerated
	310	nonsynonymous	T	C	Y	H	30	18.5	10.2, 16.8	Tolerated
	342-343	222-bp insertion	-	Duplicates nucleotides 343 to 565 inside gene	-	-	28	14.8, 17.9	8.2	-
	442	1-bp deletion	G	-	-	-	22	9.9, 20.0	none	-
	461	nonsynonymous	A	G	D	G	20	17.3	9.9	Not tolerated
	490	1-bp deletion	C	-	-	-	4	9.1A	3.0, 9.1B	-
	575	nonsynonymous	T	C	L	P	20	17.3	9.9	Not tolerated
probable oxidoreductase *mexS* (PA2491)										

Mutated gene†	**Nucleotide position mutated in gene**	**Type of mutation**	**Nucleotide(s) prior to mutation**	**Nucleotide(s) after mutation**	**Amino acid prior to mutation**	**Amino acid after mutation**	**Patient no.**	**Mutated isolate(s), named by patient age (yr) when isolate was collected**	**Isolates not containing the mutation, named by patient age (yr) when isolate was collected‡**	**Computational prediction of effect on protein function§**
	142	nonsynonymous	C	T	R	C	29	13.5, 14.0	5.6	Not tolerated
	155	nonsynonymous	C	A	A	D	17	22.2	6.6, 17.7	Tolerated
	280	nonsynonymous	C	T	P	S	23	6.2	10.0, 14.0	Tolerated
	304	nonsynonymous	G	T	D	Y	9	19.7A, 19.7B	9.2	Not tolerated
	529	nonsynonymous	A	G	T	A	20	17.3	9.9	Not tolerated
	549	6bp deletion	GAGTTC	-	-	-	7	15.4A	7.8, 15.4B	-
	730	nonsynonymous	G	A	G	S	7	15.4B	7.8, 15.4A	Not tolerated
	935	nonsynonymous	T	G	F	C	23	10.0, 14.0	6.2	Not tolerated
transcriptional regulator *exsA* (PA1713)										
	128	IS insertion	-	-	-	-	20	17.3	9.9	-
	427	nonsynonymous	G	A	E	K	2	12.8, 13.0	6.3	Not tolerated
	463	3bp insertion	GGGG	GGGGGGG	-	-	22	20.0	9.9	-
	467	nonsynonymous	C	T	P	L	2	13.0	12.8, 6.3	Tolerated
	596	nonsynonymous	C	T	T	I	23	14.0	6.2, 10.0	Tolerated
	676	nonsynonymous	C	T	H	Y	23	10.0	6.2, 14.0	Not tolerated
	727	nonsynonymous	G	C	A	P	27	11.4	6.4, 9.1	Not tolerated

Mutated gene†	**Nucleotide position mutated in gene**	**Type of mutation**	**Nucleotide(s) prior to mutation**	**Nucleotide(s) after mutation**	**Amino acid prior to mutation**	**Amino acid after mutation**	**Patient no.**	**Mutated isolate(s), named by patient age (yr) when isolate was collected**	**Isolates not containing the mutation, named by patient age (yr) when isolate was collected[‡]**	**Computational prediction of effect on protein function[§]**
	730	nonsynonymous	G	A	G	S	14	13.1A, 13.1B	4.3	Not tolerated
probable acyl-CoA dehydrogenase (PA0506)										
	578	nonsynonymous	G	A	G	D	22	20.0	9.9	Not tolerated
	783	2bp deletion	AC	-	-	-	20	17.3	9.9	-
	1091	nonsynonymous	T	A	L	Q	17	17.7, 22.2	6.6	Tolerated
	1285	nonsynonymous	C	G	Q	E	27	9.1	6.4, 11.4	Not tolerated
	1525	nonsynonymous	G	C	A	P	7	15.4B	7.8, 15.4A	Not tolerated
	1651	nonsynonymous	A	G	T	A	21	19.5	0.5, 10.7, 14.3, 15.2	Tolerated
*probable methylesterase involved in chemosensory signal transduction *wspF* (PA3703)										
	37-63	27-bp deletion	GTCGAGGC GCTGCGCC	-	-	-	20	17.3	9.9	-

Mutated gene†	**Nucleotide position mutated in gene**	**Type of mutation**	**Nucleotide(s) prior to mutation**	**Nucleotide(s) after mutation**	**Amino acid prior to mutation**	**Amino acid after mutation**	**Patient no.**	**Mutated isolate(s), named by patient age (yr) when isolate was collected**	**Isolates not containing the mutation, named by patient age (yr) when isolate was collected[‡]**	**Computational prediction of effect on protein function[§]**
			GCGCGCTG GCC							
	494	nonsynonymous	C	G	S	W	16	7.0	23.4	Not tolerated
	761	nonsynonymous	A	G	D	G	10	6.7	15.4A, 15.4B	Not tolerated
	800-801	16-bp insertion	-	TCGCCACGC CAATAAT	-	-	17	17.7, 22.2	6.6	-
	895	nonsynonymous	C	T	Q	STOP	23	10.0, 14.0	6.2	-
	955	nonsynonymous	C	T	Q	STOP	25	14.5	1.2, 21.0	-
*RNA polymerase sigma-54 factor *rpoN* (PA4462)										
	99	1-bp deletion	CC	C	-	-	13	19.6A, 19.6B	10.8	-
	324	nonsynonymous	G	A	W	STOP	9	19.7A, 19.7B	9.2	-
	1019	3-bp insertion	-	AGC	-	S	17	17.7	6.6, 22.2	-

Mutated gene†	**Nucleotide position mutated in gene**	**Type of mutation**	**Nucleotide(s) prior to mutation**	**Nucleotide(s) after mutation**	**Amino acid prior to mutation**	**Amino acid after mutation**	**Patient no.**	**Mutated isolate(s), named by patient age (yr) when isolate was collected**	**Isolates not containing the mutation, named by patient age (yr) when isolate was collected[‡]**	**Computational prediction of effect on protein function[§]**
	1279-1280	2-bp deletion	GT	-	-	-	30	16.8, 18.5	10.2	-
	1346	nonsynonymous	C	T	A	V	2	12.8, 13.0	6.3	Not tolerated
*transcriptional regulator of flagella synthesis *fleQ* (PA1097)										
	230	nonsynonymous	A	G	D	G	13	19.6A	10.8, 19.6B	Tolerated
	235	nonsynonymous	T	C	Y	H	30	18.5	10.2, 16.8	Tolerated
	364	nonsynonymous	C	T	Q	STOP	11	15.3	7.3, 13.7	-
	433	nonsynonymous	A	G	S	G	21	19.5	0.5, 10.7, 14.3, 15.2	Not tolerated
	1135	nonsynonymous	G	A	G	R	12	21.1A	-	Not tolerated
transcriptional regulator *mexT* (PA2492)										
	468	nonsynonymous	C	A	P	Q	17	22.2	6.6, 17.7	Not tolerated

Mutated gene†	**Nucleotide position mutated in gene**	**Type of mutation**	**Nucleotide(s) prior to mutation**	**Nucleotide(s) after mutation**	**Amino acid prior to mutation**	**Amino acid after mutation**	**Patient no.**	**Mutated isolate(s), named by patient age (yr) when isolate was collected**	**Isolates not containing the mutation, named by patient age (yr) when isolate was collected[‡]**	**Computational prediction of effect on protein function[§]**
	659	nonsynonymous	G	A	D	N	13	19.6A, 19.6B	10.8	Not tolerated
	893	nonsynonymous	G	C	G	R	27	9.1, 11.4	6.4	Tolerated
	977	nonsynonymous	G	A	A	T	7	15.4A	7.8	Not tolerated
*probable transcriptional regulator *nalD* (PA3574)										
	26-31	1-bp deletion	AAAAAA	AAAAA	-	-	13	19.6A, 19.6B	10.8	-
	123	synonymous	G	A	G	G	23	6.2	10.0, 14.0	-
	160	nonsynonymous	A	C	K	Q	26	8.6, 13.3	2.9	None
*β-lactamase expression regulator *ampD* (PA4522)										
	78-88	11-bp deletion	AGGGGACG CGG	-	-	-	10	15.4A, 15.4B	6.7	-
	83	nonsynonymous	A	G	D	G	22	20.0	9.9	Tolerated

Mutated gene†	Nucleotide position mutated in gene	Type of mutation	Nucleotide(s) prior to mutation	Nucleotide(s) after mutation	Amino acid prior to mutation	Amino acid after mutation	Patient no.	Mutated isolate(s), named by patient age (yr) when isolate was collected	Isolates not containing the mutation, named by patient age (yr) when isolate was collected[‡]	Computational prediction of effect on protein function[§]
	122	nonsynonymous	C	T	P	L	8	13.3A	1.1, 13.3B	Not tolerated
	339	nonsynonymous	C	A	F	L	17	22.2	6.6, 17.7	Not tolerated
probable aldehyde dehydrogenase (PA0366)										
	413	nonsynonymous	G	A	G	D	30	18.5	10.2, 16.8	Tolerated
	1380	1bp deletion	CCCCCCC	CCCCCC	-	-	22	20.0	9.9	-
	1380	1bp insertion	CCCCCCC	CCCCCCCC	-	-	23	6.2	10.0, 14.0	-
*adenylate cyclase *cyaB* (PA3217)										
	316	nonsynonymous	T	C	F	L	21	19.5	0.5, 10.7, 14.3, 15.2	Tolerated
	411	synonymous	C	T	V	V	23	6.2	10.0, 14.0	-
	1324	nonsynonymous	G	A	D	N	20	17.3	9.9	Not tolerated
hypothetical protein P1-001										

Mutated gene†	Nucleotide position mutated in gene	Type of mutation	Nucleotide(s) prior to mutation	Nucleotide(s) after mutation	Amino acid prior to mutation	Amino acid after mutation	Patient no.	Mutated isolate(s), named by patient age (yr) when isolate was collected	Isolates not containing the mutation, named by patient age (yr) when isolate was collected[‡]	Computational prediction of effect on protein function[§]
	50	nonsynonymous	C	T	P	L	13	19.6A	10.8, 19.6B	None
	262	1bp deletion	CCCCCCCC	CCCCCCC	-	-	22	20.0	9.9	-
	262	2bp insertion	CCCCCCCC	CCCCCCCCCC	-	-	13	19.6A	10.8, 19.6B	-
probable methyltransferase (PA3817)										
	45	synonymous	C	A	P	P	17	22.2	6.6, 17.7	-
	529	nonsynonymous	A	G	T	A	21	14.3	0.6, 10.7, 15.2, 19.5	Tolerated
	593	nonsynonymous	A	G	E	G	23	6.2	10.0, 14.0	Not tolerated
type 4 fimbrial precursor *pilB* (PA4526)										
	936	synonymous	G	A	L	L	11	7.3	15.3	-
	618	nonsynonymous	G	C	E	D	26	8.6	2.9, 13.3	Not tolerated
probable transcriptional regulator (PA3565)										

Mutated gene†	Nucleotide position mutated in gene	Type of mutation	Nucleotide(s) prior to mutation	Nucleotide(s) after mutation	Amino acid prior to mutation	Amino acid after mutation	Patient no.	Mutated isolate(s), named by patient age (yr) when isolate was collected	Isolates not containing the mutation, named by patient age (yr) when isolate was collected‡	Computational prediction of effect on protein function§
	913	nonsynonymous	A	G	R	G	8	13.3A	1.1, 13.3B	None
probable transcriptional regulator (PA2312)										
	306	synonymous	G	A	A	A	7	15.4A	7.8, 15.4B	-

IS, insertion element.

†Genes with names preceded by an asterisk were chosen for sequencing on the basis of other studies. The remaining 24 genes were mutated in the 96-month isolate of patient 1.

‡For each mutation, all isolates listed are clonally related and are from the same patient.

§Nonsynonymous mutations that are predicted to cause partial or complete loss of function are labeled “Not tolerated.” No prediction was made when fewer than four homologs were available.