

**e-Table 1. Variants of *CFTR* identified in 103 Chinese children (100 families) with CF presented to Beijing Children's Hospital**

Case No.	Sex	Age at Dx, y	Sweat conductivity, mmol/L	Region	Nucleotide change (HGVS nomenclature)	Amino acid change	Type of mutation	Genotype	Familial targeted mutation study	
									Father	Mother
1 <sup>[4]</sup>	M	11.58	101	Ex 13	c.1699G>T	p.Asp567Tyr	Missense	Compound	+/-	-/-
				Ex 24	c.3909C>G	p.Asn1303Lys	Missense	heterozygous	-/-	+/-
2 <sup>[4]</sup>	F	10.58	103	Ex 3	c.263T>G	p.Leu88X	Nonsense	Compound heterozygous	-/-	+/-
				In 13	c.1766+5G>T	-	Splicing		+/-	-/-
				3'UTR	c.*110C>G	-	Sequence variation		-/-	+/-
3 <sup>[4]</sup>	M	13.25	101	Ex 22	c.3700A>G	p.Ile1234Val	Missense	Compound	+/-	-/-
				Ex 8	c.960dupA	p.Ser321IlefsX43	Frameshift	heterozygous	-/-	-/-
4 <sup>[4]</sup>	F	13.67	99	Ex 3	c.263T>G	p.Leu88X	Nonsense	Compound	-/-	+/-
				Ex 18	c.2909G>A	p.Gly970Asp	Missense	heterozygous	+/-	-/-
5 <sup>[4]</sup>	M	7.17	106	Ex 4	c.326A>G	p.Tyr109Cys	Missense	Compound	-/-	+/-
				Ex 8	c.1000C>T	p.Arg334Trp	Missense	heterozygous	+/-	-/-

				Ex 12	c.1666A>G	p.Ile556Val	Missense		+/+	-/-
6 <sup>[4]</sup>	F	10.67	127	Ex 6	c.595C>T	p.His199Tyr	Missense	Heterozygous	ND	ND
7 <sup>[4]</sup>	F	7.75	118	Ex 3	c.223C>T	p.Arg75X	Nonsense	Compound heterozygous	ND	ND
				Ex 4	c.326A>G	p.Tyr109Cys	Missense		ND	ND
8 <sup>[4]</sup>	F	7.33	118	Ex 8	c.1000C>T	p.Arg334Trp	Missense	Heterozygous	+/-	-/-
9 <sup>[4]</sup>	F	10.17	105	Ex 3	c.263T>G	p.Leu88X	Nonsense	Homozygous	+/-	+/-
10 <sup>[4]</sup>	F	11.08	96	Ex 12	c.1666A>G	p.Ile556Val	Missense	Heterozygous	ND	ND
11 <sup>[4]</sup>	M	8.25	115	Ex 4	c.293A>G	p.Gln98Arg	Missense	Compound heterozygous	-/-	+/-
				Ex 5	c.558C>G	p.Asn186Lys	Missense		+/-	-/-
12 <sup>[4]</sup>	F	4.17	101	Ex 4	c.326A>G	p.Tyr109Cys	Missense	Compound heterozygous	ND	ND
				Ex 14	c.2374C>T	p.Arg792X	Nonsense		ND	ND
13 <sup>[4]</sup>	M	3.67	99	Ex 12	c.1666A>G	p.Ile556Val	Missense	Homozygous	+/-	+/-
14 <sup>[4]</sup>	F	12.67	122	Ex 4	c.293A>G	p.Gln98Arg	Missense	Heterozygous	-/-	+/-
15 <sup>[4]</sup>	M	11.00	ND	Ex 6	c.648G>A	p.Trp216X	Nonsense	Homozygous	+/-	+/-
				In 14	c.2491-126T>C	-	Sequence variation		+/-	+/-
16 <sup>[4]</sup>	F	10.33	ND	Ex 20	c.3196C>T	p.Arg1066Cys	Missense	Homozygous	ND	ND

17 <sup>[4]</sup>	M	11.17	ND	Ex 4	c.413_415dupTA C	p.Leu138dup	In-frame insertion	Homozygous	ND	ND
18 <sup>[4]</sup>	F	3.42	ND	Ex 8	c.1075C>T	p.Gln359X	Nonsense	Compound heterozygous	ND	ND
				Ex 20	c.3307delA	p.Ile1103X	Nonsense		ND	ND
19 <sup>[4]</sup>	F	14.00	115	Ex 18	c.2909G>A	p.Gly970Asp	Missense	Homozygous	+/-	+/-
20 <sup>[6]</sup>	F	0.42	130	Ex 3	c.223C>T	p.Arg75X	Nonsense	Compound heterozygous	-/-	+/-
				Ex 18	c.2909G>A	p.Gly970Asp	Missense		+/-	-/-
21 <sup>[6]</sup>	F	2.16	134	Ex 11	c.1521_1523delC TT	p.Phe508del	In-frame deletion	Homozygous	+/-	+/-
22 <sup>[6]</sup>	F	8.25	122	Ex 8	c.1000C>T	p.Arg334Trp	Missense	Compound heterozygous	+/-	-/-
				Ex 13	c.1733T>C	p.Leu578Pro	Missense		-/-	+/-
23 <sup>[6]</sup>	M	0.75	134	Ex 3	c.264_268delAT ATT	p.Leu88PhefsX21	Frameshift	Compound heterozygous	-/-	+/-
				Ex 23	c.3860delG	p.Gly1287GlufsX 2	Frameshift		+/-	-/-
24 <sup>[6]</sup>	F	0.42	105	Ex 3	c.263T>G	p.Leu88X	Nonsense	Compound heterozygous	-/-	+/-
				Ex 18	c.2909G>A	p.Gly970Asp	Missense		+/-	-/-

25 <sup>[6]</sup>	M	0.67	93	In 8	c.1116+1G>A	–	Splicing	Compound heterozygous	+/-	-/-
				Ex 18	c.2909G>A	p.Gly970Asp	Missense		-/-	+/-
26 <sup>[6]</sup>	M	0.25	ND	Ex 18	c.2909G>A	p.Gly970Asp	Missense	Compound heterozygous	+/-	-/-
				Ex 23	c.3718-?_3873+? del	–	Large deletion		-/-	+/-
27 <sup>[6]</sup>	F	0.92	115	Ex 14	c.2236_2246delG AGGCGATACTi nsAAAAATC	p.Glu746LysfsX8	Frameshift	Compound heterozygous	-/-	+/-
				Ex 25	c.3635delT	p.Val1212AlafsX 16	Frameshift		+/-	-/-
28 <sup>[6]</sup>	M	6.67	125	In 9	c.1210-3C>G	–	Splicing	Compound heterozygous	+/-	-/-
				In 24	c.3964-7A>G	–	Splicing		-/-	+/-
29 <sup>[6]</sup>	M	0.75	125	Ex 19	c.3068T>G	p.Ile1023Arg	Missense	Compound heterozygous	+/-	-/-
				Ex 6	c.595C>T	p.His199Tyr	Missense		-/-	+/-
30-1	F	7.67	151	Ex 5	c.532G>A	p.Gly178Arg	Missense	Homozygous	+/-	+/-
30-2 <sup>[6]</sup>	F	0.50	ND	Ex 5	c.532G>A	p.Gly178Arg	Missense	Homozygous	+/-	+/-

31-1	M	9.33	102	In 5	c.579+1_579+2in sACAT	–	Splicing	Compound heterozygous	+/-	-/-
				In 13	c.1766+5G>T	–	Splicing		-/-	+/-
31-2 <sup>[6]</sup>	F	8.83	141	In 5	c.579+1_579+2in sACAT	–	Splicing	Compound heterozygous	+/-	-/-
				In 13	c.1766+5G>T	–	Splicing		-/-	+/-
32	F	1.08	125	Ex 11	c.1521_1523delC TT	p.Phe508del	In-frame deletion	Compound heterozygous	+/-	-/-
				Ex 11	c.1429_1437delC CTTCAGAG	p.Pro477_Glu479 del	In-frame deletion		-/-	+/-
33	M	7.33	125	Ex 20	c.3209G>C	p.Arg1070Pro	Missense	Compound	+/-	-/-
				<b>Ex 14</b>	<b>c.2328dupA</b>	<b>p.Val777SerfsX2</b>	<b>Frameshift</b>	heterozygous	-/-	+/-
34	F	9.00	104	Ex 4	c.293A>G	p.Gln98Arg	Missense	Compound	+/-	-/-
				Ex 19	c.3068T>G	p.Ile1023Arg	Missense	heterozygous	-/-	+/-
35	F	5.67	109	Ex 11	c.1572C>A	p.Cys524X	Nonsense	Compound	+/-	-/-
				Ex 1	c.3G>A	p.Met1Ile	Missense	heterozygous	-/-	+/-
36	F	10.67	127	Ex 6	c.595C>T	p.His199Tyr	Missense		+/-	-/-

				<b>Ex 14</b>	<b>c.2058_2061delT</b>	<b>p.Phe687X</b>	<b>Nonsense</b>	Compound heterozygous	-/-	+/-
37	F	14.33	140	Ex 11	c.1409T>A	p.Val470Glu	Missense	Compound heterozygous	+/-	-/-
				In 9	c.1210-3C>G	-	Splicing		-/-	+/-
38	M	13.25	109	<b>Ex 10</b>	<b>c.1347_1350delA</b>	<b>p.Arg450AspfsX</b>	<b>Frameshift</b>	Compound heterozygous	ND	ND
				Ex 18	c.2909G>A	p.Gly970Asp	Missense		Compound ND	ND
39	M	10.50	83	In 23	c.3874-4522A>G	-	Splicing	Compound	+/-	-/-
				Ex 18	c.2936A>C	p.Asp979Ala	Missense	Heterozygous	-/-	+/-
40	M	9.83	141	Ex 8	c.1000C>T	p.Arg334Trp	Missense	Compound heterozygous	-/-	+/-
				Ex 3	c.264_268delATT	p.Leu88PhefsX21	Frameshift		+/-	-/-
41	F	5.58	67	Ex 6	c.650A>G	p.Glu217Gly	Missense	Compound heterozygous	-/-	+/-
				Ex 10	c.1231A>G	p.Lys411Glu	Missense		+/-	-/-
42	M	7.00	130	In 13	c.1766+5G>T	-	Splicing	Compound heterozygous	-/-	+/-
				Ex 22	c.3484C>T	p.Arg1162X	Nonsense		+/-	-/-

43	M	3.83	ND	Ex 6	c.595C>T	p.His199Tyr	Missense	Compound heterozygous	+/-	-/-
				Ex 14	c.2290C>T	p.Arg764X	Nonsense		-/-	+/-
44	M	14.42	64	In 22	c.3718-2477C>T	-	Sequence variation	Compound heterozygous	+/-	-/-
				Ex 17	c.2834C>T	p.Ser945Leu	Missense		-/-	+/-
				Ex 4	c.374T>C	p.Ile125Thr	Missense		-/-	+/-
45	M	8.50	163	Ex 11	c.1521_1523delC TT	p.Phe508del	In-frame deletion	Compound heterozygous	+/-	-/-
				Ex 4	c.293A>G	p.Gln98Arg	Missense		-/-	+/-
46	M	10.17	60	Ex 4	c.374T>C	p.Ile125Thr	Missense	Compound heterozygous	-/-	+/-
				<b>Ex 14</b>	<b>c.2042A&gt;T</b>	<b>p.Glu681Val</b>	<b>Missense</b>		+/-	-/-
47	M	13.08	99	Ex 17	c.2834C>T	p.Ser945Leu	Missense	Homozygous	+/-	+/-
48	F	11.25	118	Ex 14	c.2036G>A	p.Trp679X	Nonsense	Compound heterozygous	+/-	-/-
				In 2	c.164+2T >C	-	Splicing		-/-	+/-
49	F	4.67	85	Ex 4	c.325T>G	p.Tyr109Asp	Missense	Compound heterozygous	-/-	+/-
				Ex 20	c.3196C>T	p.Arg1066Cys	Missense		+/-	-/-
50	M	9.08	96	Ex 4	c.350G>A-7T	p.Arg117His-7T	Missense		-/-	+/-

				Ex 14	c.2036G>A	p.Trp679X	Nonsense	Compound heterozygous	+/-	-/-
51	F	11.00	96	Ex 18	c.2909G>A	p.Gly970Asp	Missense	Compound heterozygous	+/-	-/-
				Ex 8	c.1000C>T	p.Arg334Trp	Missense		-/-	+/-
52	M	14.50	114	Ex 3	c.223C>T	p.Arg75X	Nonsense	Compound heterozygous	+/-	-/-
				Ex 4	c.293A>G	p.Gln98Arg	Missense		-/-	+/-
53	F	3.92	122	Ex 21	c.3387delT	p.Gly1130ValfsX 4	Frameshift	Compound heterozygous	+/-	-/-
				Ex 11	c.1409T>A	p.Val470Glu	Missense		-/-	+/-
54	F	8.58	125	Ex 12	c.1657C>T	p.Arg553X	Nonsense	Compound heterozygous	+/-	-/-
				<b>Ex 14</b>	<b>c.1810A&gt;C</b>	<b>p.Thr604Pro</b>	<b>Missense</b>		-/-	+/-
55	M	10.67	108	Ex 18	c.2909G>A	p.Gly970Asp	Missense	Compound heterozygous	+/-	-/-
				Ex 20	c.3196C>A	p.Arg1066Ser	Missense		-/-	+/-
56	M	2.08	111	In 13	c.1766+2T>C	-	Splicing	Compound heterozygous	+/-	-/-
				<b>In 21</b>	<b>c.3469-2A&gt;T</b>	-	<b>Splicing</b>		-/-	+/-
57	F	9.42	108	In 23	c.3874-4522A>G	-	Sequence variation	Compound heterozygous	+/-	-/-



				<b>Ex 10</b>	<b>c.1368delT</b>	<b>p.Ala457LeufsX 12</b>	<b>Frameshift</b>		-/-	+/-
58	F	8.75	136	<b>Ex 11</b>	<b>c.1523_1534delT TGGTGTTTCC T</b>	<b>p.Phe508_Ser511 del</b>	<b>In-frame deletion</b>	Compound heterozygous	+/-	-/-
				<b>Ex 11</b>	<b>c.1393-?_1584+? del</b>	-	<b>Large deletion</b>		-/-	+/-
59	F	7.75	97	Ex 18	c.2977G>T	p.Asp993Tyr	Missense	Compound heterozygous	ND	ND
				<b>Ex 10</b>	<b>c.1265C&gt;T</b>	<b>p.Ser422Phe</b>	<b>Missense</b>		ND	ND
				In 19	c.3140-26A>G	-	Missense		ND	ND
60	M	1.75	144	Ex 24	c.3908delA	p.Asn1303ThrfsX 25	Frameshift	Compound heterozygous	+/-	-/-
				Ex 18	c.2909G>A	p.Gly970Asp	Missense		-/-	+/-
61	F	4.17	ND	Ex 15	c.2551C>T	p.Arg851X	Nonsense	Compound heterozygous	-/-	+/-
				<b>Ex 10</b>	<b>c.1219G&gt;T</b>	<b>p.Glu407X</b>	<b>Nonsense</b>		+/-	-/-
				Promoter	c.-152G>C	-	Sequence variation		-/-	+/-

62	M	16.83	ND	Ex 3	c.223C>T	p.Arg75X	Nonsense	Compound heterozygous	ND	ND
				Ex 18	c.2909G>A	p.Gly970Asp	Missense		ND	ND
63	F	13.75	ND	<b>Ex 14</b>	<b>c.1810A&gt;C</b>	<b>p.Thr604Pro</b>	<b>Missense</b>	Compound heterozygous	ND	ND
				<b>Ex 8</b>	<b>c.940G&gt;T</b>	<b>p.Gly314Trp</b>	<b>Missense</b>		ND	ND
				3'UTR	c.*133delT	–	Sequence variation		ND	ND
64	F	12.50	168	Ex 4	c.374T>C	p.Ile125Thr	Missense	Compound heterozygous	+/-	-/-
				Ex 11	c.1521_1523delC TT	p.Phe508del	In-frame deletion		-/-	+/-
65	F	1.67	89	Ex 13	c.1703T>A	p.Leu568X	Nonsense	Homozygous	+/-	+/-
66	M	4.00	ND	Ex 12	c.1657C>T	p.Arg553X	Nonsense	Homozygous	+/-	+/-
67	F	12.00	77	Ex 23	c.3841C>T	p.Gln1281X	Nonsense	Compound heterozygous	+/-	-/-
				<b>Ex 4</b>	<b>c.298C&gt;T</b>	<b>p.Leu100Phe</b>	<b>Missense</b>		-/-	+/-
68	M	11.00	161	In 14	c.2491-2A>G	–	Missense	Compound heterozygous	-/-	-/-
				Ex 20	c.3196C>T	p.Arg1066Cys	Missense		-/-	+/-
69	F	6.33	85	Ex 22	c.3472C>T	p.Arg1158X	Nonsense		+/-	-/-

				Ex 2-3	c.54-?_273+?del	–	Large deletion	Compound heterozygous	-/-	+/-
70	F	3.25	101	Ex 6	c.607A>T	p.Ile203Phe	Missense	Compound heterozygous	+/-	-/-
				Ex 18-20	c.2909-?_3367+?del	–	Large deletion		-/-	+/-
71	F	13.25	122	<b>Ex 20</b>	<b>c.3140-?_3367+?del</b>	–	<b>Large deletion</b>	Compound heterozygous	-/-	+/-
				Ex 3	c.233dupT	p.Trp79LeufsX32	Frameshift		+/-	-/-
72	F	7.33	118	Ex 8	c.1000C>T	p.Arg334Trp	Missense	Compound heterozygous	+/-	-/-
				<b>Ex 11</b>	<b>c.1393-?_1584+?del</b>	–	<b>Large deletion</b>		-/-	+/-
73	F	15.33	149	Ex 4	c.293A>G	p.Gln98Arg	Missense	Compound heterozygous	+/-	-/-
				Ex 11	c.1521_1523delC TT	p.Phe508del	In-frame deletion		-/-	+/-
74	M	5.08	130	Ex 12	c.1657C>T	p.Arg553X	Nonsense	Compound heterozygous	+/-	-/-
				Ex 19	c.3068T>G	p.Ile1023Arg	Missense		-/-	+/-

75	F	8.58	146	<b>Ex 3</b>	<b>c.222delG</b>	<b>p.Arg75AspfsX1 5</b>	<b>Frameshift</b>	Compound heterozygous	+/-	-/-
				Ex 3	c.263T>G	p.Leu88X	Nonsense		-/-	+/-
76-1	M	7.75	130	Ex 18-20	c.2909-?_3367+? del	-	Large deletion	Compound heterozygous	+/-	-/-
				Ex 20	c.3196C>T	p.Arg1066Cys	Missense		-/-	+/-
76-2	F	5.58	120	Ex 18-20	c.2909-?_3367+? del	-	Large deletion	Compound heterozygous	+/-	-/-
				Ex 20	c.3196C>T	p.Arg1066Cys	Missense		-/-	+/-
77	F	4.50	60	<b>Ex 4</b>	<b>c.298C&gt;T</b>	<b>p.Leu100Phe</b>	<b>Missense</b>	Compound heterozygous	-/-	+/-
				Ex 6	c.650A>G	p.Glu217Gly	Missense		+/-	-/-
78	F	12.92	119	Ex 22	c.3476C>T	p.Ser1159Phe	Missense	Compound heterozygous	+/-	-/-
				<b>In 21</b>	<b>c.3469-12T&gt;G</b>	-	<b>Splicing</b>		-/-	+/-
79	M	2.75	134	Ex 22	c.3484C>T	p.Arg1162X	Nonsense	Compound heterozygous	+/-	-/-
				Ex 10	c.1369G>C	p.Ala457Pro	Missense		-/-	+/-
80	F	10.67	126	<b>Ex 18-21</b>	<b>c.2909-?_3468+? del</b>	-	<b>Large deletion</b>	Compound heterozygous	+/-	-/-

				Ex 18	c.2909G>A	p.Gly970Asp	Missense		-/-	+/-
81	F	10.58	127	Ex 3	c.223C>T	p.Arg75X	Nonsense	Compound	+/-	-/-
				Ex 18	c.2909G>A	p.Gly970Asp	Missense	heterozygous	-/-	+/-
82	M	6.00	144	In 13	c.1766+5G>T	-	Splicing	Homozygous	+/-	+/-
83	F	3.17	87	Ex 3	c.264_268delATA TT	p.Leu88PhefsX21	Frameshift	Compound	+/-	-/-
				In 22	c.3718-2477C>T	-	Sequence variation	heterozygous	-/-	+/-
84	F	0.50	ND	Ex 2-3	c.54-?_273+?del	-	Large deletion	Compound	+/-	-/-
				Ex 18	c.2909G>A	p.Gly970Asp	Missense	heterozygous	-/-	+/-
85	F	11.00	130	Ex 3	c.233dupT	p.Trp79LeufsX32	Frameshift	Compound	+/-	-/-
				Ex 14	<b>c.2489dupA</b>	<b>p.Glu831GlyfsX 5</b>	<b>Frameshift</b>	heterozygous	-/-	+/-
86	F	11.33	115	Ex 18	c.2909G>A	p.Gly970Asp	Missense	Compound	+/-	-/-
				Ex 18-20	c.2909-?_3367+? del	-	Large deletion	heterozygous	-/-	+/-

87	M	12.50	124	Ex 3	c.263T>G	p.Leu88X	Nonsense	Compound heterozygous	+/-	-/-
				<b>Ex 11</b>	<b>c.1514delA</b>	<b>p.Asn505IlefsX2 2</b>	<b>Frameshift</b>		-/-	+/-
88	F	5.67	95	Ex 4	c.374T>C	p.Ile125Thr	Missense	Compound heterozygous	+/-	-/-
				<b>Ex 22</b>	<b>c.3659C&gt;T</b>	<b>p.Thr1220Ile</b>	<b>Missense</b>		-/-	+/-
89	M	4.83	152	Ex 10	c.1388G>A	p.Gly463Asp	Missense	Compound heterozygous	+/-	-/-
				In 16	c.2657+5G>A	-	Splicing		-/-	+/-
90	F	12.00	ND	Ex 18	c.2909G>A	p.Gly970Asp	Missense	Compound heterozygous	+/-	-/-
				Ex 20	c.233dupT	p.Trp79LeufsX32	Frameshift		-/-	+/-
91	F	5.42	ND	Ex 6	c.595C>T	p.His199Tyr	Missense	Compound heterozygous	+/-	-/-
				Ex 20	c.3176T>G	p.Leu1059X	Nonsense		-/-	+/-
92	F	3.17	ND	Ex 4	c.319_326delGC TTCCTA	p.Ala107X	Nonsense	Compound heterozygous	-/-	+/-
				Ex 11	c.1456G>T	p.Gly486X	Nonsense		+/-	-/-
93	F	7.58	112	Ex 14	c.2374C>T	p.Arg792X	Nonsense	Compound heterozygous	+/-	-/-
				Ex 4-11	c.274-?_1584+?d el	-	Large deletion		-/-	+/-

94	M	7.17	81	<b>Ex 14</b>	<b>c.1772T&gt;C</b>	<b>p.Val591Ala</b>	<b>Missense</b>	Compound heterozygous	+/-	-/-
				Ex 4-11	c.274-?_1584+?del	-	Large deletion		-/-	+/-
95	M	0.50	ND	Ex 17	c.2812G>T	p.Val938Leu	Missense	Compound heterozygous	+/-	-/-
				Ex 18	c.2909G>A	p.Gly970Asp	Missense		-/-	+/-
96	F	3.75	ND	Ex 18	c.2909G>A	p.Gly970Asp	Missense	Compound heterozygous	+/-	-/-
				<b>Ex 4</b>	<b>c.464C&gt;G</b>	<b>p.Ala155Gly</b>	<b>Missense</b>		-/-	+/-
97	M	0.58	ND	In 13	c.1766+2T>C	-	Splicing	Homozygous	+/-	+/-
98	F	6.00	ND	Ex 3	c.263T>G	p.Leu88X	Nonsense	Compound heterozygous	+/-	-/-
				In 13	c.1766+2T>C	-	Splicing		-/-	+/-
99	F	13.50	68	Ex 18	c.2909G>A	p.Gly970Asp	Missense	Compound heterozygous	+/-	-/-
				In 15	c.2619+2T>A	-	Splicing		-/-	+/-
100	M	0.42	103	Ex 8	c.1064C>G			Compound heterozygous	+/-	-/-
				In 9	c.1210-3C>G	-	Splicing		-/-	+/-

Novel variants are formatted in bold.

**e-Table 2. Spectrum of *CFTR* variants detected in 202 Chinese patients with CF**

No.	Region	Mutations	Amino acid change	Pathogenic significance	Reported/Novel	No. of alleles	Frequency, %
		(HGVS nomenclature)					
1 <sup>[4, 6, 10, 12, 16, 20, 24, 25, 27, 29, 36, 51]</sup>	Ex 18	c.2909G>A	p.Gly970Asp	CF-causing	Reported	47	12.11
2 <sup>[4, 6, 10, 32, 34, 40, 42, 43, 46-48, 50]</sup>	In 13	c.1766+5G>T	–	CF-causing	Reported (Chinese/Thai)	21	5.41



3 <sup>[9, 10, 12, 13, 34, 42, 45, 48]</sup>	Ex 12	c.1657C>T	p.Arg553X	CF-causing	Reported	14	3.61
4 <sup>[4, 10, 12]</sup>	Ex 4	c.293A>G	p.Gln98Arg	CF-causing	Reported	12	3.09
5 <sup>[6, 10, 42, 47, 48]</sup>	Ex 19	c.3068T>G	p.Ile1023Arg	Unknown	Reported (Chinese only)	11	2.84
6 <sup>[4, 6, 30, 37]</sup>	Ex 8	c.1000C>T	p.Arg334Trp	CF-causing	Reported	10	2.58
7 <sup>[4, 6, 20, 21]</sup>	Ex 3	c.263T>G	p.Leu88X	CF-causing	Reported	10	2.58
8 <sup>[4, 6, 10, 19, 27]</sup>	Ex 6	c.595C>T	p.His199Tyr	CF-causing	Reported	9	2.32

9 <sup>[4, 10, 26, 38]</sup>	Ex 20	c.3196C>T	p.Arg1066Cys	CF-causing	Reported	8	2.06
10 <sup>[6, 20]</sup>	Ex 11	c.1521_1523delCTT	p.Phe508del	CF-causing	Reported	7	1.80
11 <sup>[18, 31, 38]</sup>	Ex 6	c.650A>G	p.Glu217Gly	Unknown	Reported	6	1.55
12 <sup>[4, 12, 28]</sup>	3Ex 12	c.1666A>G	p.Ile556Val	Unknown	Reported	6	1.55
13 <sup>[4, 6]</sup>	Ex 3	c.223C>T	p.Arg75X	CF-causing	Reported	5	1.29
14 <sup>[33]</sup>	Ex 4	c.374T>C	p.Ile125Thr	Unknown	Reported	5	1.29
15 <sup>[6, 16]</sup>	In 9	c.1210-3C>G	–	Unknown	Reported (Chinese only)	4	1.03
16 <sup>[37]</sup>	Ex 11	c.1409T>A	p.Val470Glu	Unknown	Reported (Chinese only)	4	1.03
17 <sup>[17, 39]</sup>	Ex 14	c.2036G>A	p.Trp679X	Suspected	Reported	4	1.03
18 <sup>[38]</sup>	Ex 17	c.2834C>T	p.Ser945Leu	CF-causing	Reported	4	1.03

19 <sup>[12]</sup>	Ex 18-20	c.2909-?_3367+?del	–	Suspected	Reported	4	1.03
20 <sup>[27, 29, 38]</sup>	Ex 25	c.4056G>C	p.Gln1352His	Unknown	Reported	4	1.03
21 <sup>[10]</sup>	Ex 2-3	c.54-?_273+?del	–	Suspected	Reported	3	0.77
22	Ex 3	c.233dupT	p.Trp79LeufsX32	CF-causing	Reported	3	0.77
23 <sup>[6]</sup>	Ex 3	c.264_268delATATT	p.Leu88PhefsX21	Suspected	Reported	3	0.77
24 <sup>[4]</sup>	Ex 4	c.326A>G	p.Tyr109Cys	Unknown	Reported	3	0.77
25 <sup>[6, 24, 29]</sup>	In 8	c.1116+1G>A	–	CF-causing	Reported	3	0.77
26 <sup>[10, 20]</sup>	Ex 14	c.2125C>T	p.Arg709X	CF-causing	Reported	3	0.77
27 <sup>[4, 20]</sup>	Ex 14	c.2374C>T	p.Arg792X	CF-causing	Reported	3	0.77
28 <sup>[6, 10, 20]</sup>	Ex 22	c.3635delT	p.Val1212AlafsX1 6	Suspected	Reported (Chinese only)	3	0.77
29 <sup>[40]</sup>	Promoter	c.-152G>C	–	Unknown	Reported	2	0.52
30 <sup>[26]</sup>	Ex 1	c.3G>A	p.Met1Ile	Unknown	Reported	2	0.52

31 <sup>[12, 33]</sup>	Ex 2	c.95T>C	p.Leu32Pro	Unknown	Reported	2	0.52
32	Ex 4-11	c.274-?_1584+?del	–	Suspected	Reported	2	0.52
<b>33</b>	<b>Ex 4</b>	<b>c.298C&gt;T</b>	<b>p.Leu100Phe</b>	<b>Unknown</b>	<b>Novel</b>	<b>2</b>	<b>0.52</b>
34 <sup>[51]</sup>	Ex 4	c.319_326delGCTTCCTA	p.Ala107X	Suspected	Reported	2	0.52
35 <sup>[10]</sup>	Ex 4	c.325T>G	p.Tyr109Asp	Unknown	Reported (Chinese only)	2	0.52
36 <sup>[36]</sup>	Ex 4	c.380T>G	p.Leu127X	Suspected	Reported	2	0.52
37 <sup>[4]</sup>	Ex 4	c.413_415dupTAC	p.Leu138dup	CF-causing	Reported	2	0.52
38 <sup>[6]</sup>	Ex 5	c.532G>A	p.Gly178Arg	CF-causing	Reported	2	0.52
39 <sup>[4, 12]</sup>	Ex 5	c.558C>G	p.Asn186Lys	Unknown	Reported (Chinese only)	2	0.52
40 <sup>[10]</sup>	Ex 6	c.607A>T	p.Ile203Phe	Unknown	Reported (Chinese only)	2	0.52
41 <sup>[4]</sup>	Ex 6	c.648G>A	p.Trp216X	Suspected	Reported (Chinese only)	2	0.52

42 <sup>[4, 20]</sup>	Ex 8	c.960dupA	p.Ser321IlefsX43	Suspected	Reported (Chinese only)	2	0.52
43 <sup>[35]</sup>	Ex 10	c.1369G>C	p.Ala457Pro	Unknown	Reported	2	0.52
44 <sup>[10]</sup>	Ex 10	c.1388G>A	p.Gly463Asp	Unknown	Reported	2	0.52
<b>45</b>	<b>Ex 11</b>	<b>c.1393-?_1584+?del</b>	–	<b>Suspected</b>	<b>Novel</b>	<b>2</b>	<b>0.52</b>
46 <sup>[26]</sup>	Ex 11	c.1572C>A	p.Cys524X	CF-causing	Reported	2	0.52
47 <sup>[10, 12]</sup>	In 12	c.1679+2T>C	–	Unknown	Reported	2	0.52
48	Ex 13	c.1703T>A	p.Leu568X	Suspected	Reported	2	0.52
<b>49</b>	<b>Ex 14</b>	<b>c.1810A&gt;C</b>	<b>p.Thr604Pro</b>	<b>Unknown</b>	<b>Novel</b>	<b>2</b>	<b>0.52</b>
50 <sup>[46, 43]</sup>	Ex 14	c.2083dupG	p.Glu695GlyfsX35	Suspected	Reported (Chinese only)	2	0.52
51 <sup>[19, 25]</sup>	Ex 14	c.2290C>T	p.Arg764X	CF-causing	Reported	2	0.52
52 <sup>[4]</sup>	In 14	c.2491-126T>C	–	Unknown	Reported (Chinese only)	2	0.52

53 <sup>[10, 38]</sup>	Ex 15	c.2547C>A	p.Tyr849X	CF-causing	Reported	2	0.52
54 <sup>[38]</sup>	Ex 15	c.2551C>T	p.Arg851X	CF-causing	Reported	2	0.52
55 <sup>[10, 12]</sup>	In 16	c.2658-1G>C	–	CF-causing	Reported	2	0.52
56 <sup>[46, 43]</sup>	Ex 17	c.2684G>A	p.Ser895Asn	Unknown	Reported (Chinese only)	2	0.52
57 <sup>[29, 38]</sup>	Ex 19	c.3062C>T	p.Pro1021Leu	Unknown	Reported (Chinese only)	2	0.52
58 <sup>[48]</sup>	In 19	c.3140-26A>G	–	CF-causing	Reported	2	0.52
59 <sup>[23]</sup>	Ex 20	c.3140-454_c.3367+249del193ins 13	–	Suspected	Reported (Chinese only)	2	0.52
60 <sup>[10]</sup>	Ex 20	c.3196C>A	p.Arg1066Ser	Unknown	Reported	2	0.52
61	Ex 22	c.3484C>T	p.Arg1162X	CF-causing	Reported	2	0.52
62 <sup>[4, 20]</sup>	Ex 22	c.3700A>G	p.Ile1234Val	CF-causing	Reported	2	0.52

63	In 22	c.3717+10kbC>T	–	Unknown	Reported	2	0.52
64	In 22	c.3718-2477C>T	–	CF-causing	Reported	2	0.52
65	In 23	c.3874-4522A>G	–	Unknown	Reported	2	0.52
66 <sup>[36]</sup>	3'UTR	c.*133delT	–	Unknown	Reported	2	0.52
67 <sup>[44]</sup>	Ex 1	c.19G>T	p.Glu7X	Suspected	Reported (Chinese only)	1	0.26
68 <sup>[14]</sup>	Ex 2	c.54_164del30bp	–	Suspected	Reported (Chinese only)	1	0.26
69	In 2	c.164+2T>C	–	CF-causing	Reported	1	0.26
70 <sup>[18]</sup>	Ex 3	c.214G>A	p.Ala72Thr	Unknown	Reported	1	0.26
<b>71</b>	<b>Ex 3</b>	<b>c.222delG</b>	<b>p.Arg75AspfsX16</b>	<b>Suspected</b>	<b>Novel</b>	<b>1</b>	<b>0.26</b>
72 <sup>[22]</sup>	Ex 3	c.271G>A	p.Gly91Arg	CF-causing	Reported	1	0.26
73 <sup>[21]</sup>	Ex 4-6	c.274-?_743+?del	–	Suspected	Reported	1	0.26
74 <sup>[35]</sup>	Ex 4	c.320C>A	p.Ala107Asp	Unknown	Reported (Chinese only)	1	0.26
75	Ex 4	c.350G>A	p.Arg117His	CF-causing	Reported	1	0.26
76 <sup>[10]</sup>	Ex 4	c.405_406dupAC	p.Leu136HisfsX18	Suspected	Reported	1	0.26

77	Ex 4	c.464C>G	p.Ala155Gly	Unknown	Novel	1	0.26
78 <sup>[15]</sup>	Ex 5	c.567C>A	p.Asn189Lys	Unknown	Reported (Chinese only)	1	0.26
79 <sup>[6]</sup>	In 5	c.579+1_579+2insACAT	–	Unknown	Reported (Chinese only)	1	0.26
80 <sup>[30]</sup>	In 5	c.580-1G>T	–	CF-causing	Reported	1	0.26
81 <sup>[12]</sup>	Ex 7-11	c.744-?_1584+ ?del	–	Suspected	Reported (Chinese only)	1	0.26
82 <sup>[41]</sup>	Ex 7	c.753_754delAG	p.Arg251SerfsX6	Suspected	Reported (Chinese only)	1	0.26
83 <sup>[44]</sup>	Ex 7	c.860dupA	p.Asn287LysfsX21	Suspected	Reported (Chinese only)	1	0.26
84 <sup>[38]</sup>	Ex 7	c.861C>G	p.Asn287Lys	Unknown	Reported	1	0.26
85 <sup>[11]</sup>	Ex 7	c.865A>T	p.Arg289X	Suspected	Reported (Chinese only)	1	0.26
86 <sup>[48]</sup>	Ex 7	c.868C>T	p.Gln290X	CF-causing	Reported	1	0.26
87 <sup>[26]</sup>	In 7	c.870-1G>C	–	Unknown	Reported	1	0.26



88 <sup>[38]</sup>	Ex 8	c.884delT	p.Leu295ArgfsX8	Suspected	Reported (Chinese only)	1	0.26
<b>89</b>	<b>Ex 8</b>	<b>c.940G&gt;T</b>	<b>p.Gly314Trp</b>	<b>Unknown</b>	<b>Novel</b>	<b>1</b>	<b>0.26</b>
90 <sup>[29]</sup>	Ex 8	c.1040G>A	p.Arg347His	CF-causing	Reported	1	0.26
<b>91</b>	<b>Ex 8</b>	<b>c.1064C&gt;G</b>	<b>p.Pro355Arg</b>	<b>Unknown</b>	<b>Novel</b>	<b>1</b>	<b>0.26</b>
92 <sup>[4]</sup>	Ex 8	c.1075C>T	p.Gln359X	Suspected	Reported (Chinese only)	1	0.26
93 <sup>[27]</sup>	In 8	c.1117-1G>C	–	Unknown	Reported (Chinese only)	1	0.26
<b>94</b>	<b>Ex 10</b>	<b>c.1219G&gt;T</b>	<b>p.Glu407X</b>	<b>Suspected</b>	<b>Novel</b>	<b>1</b>	<b>0.26</b>
95	Ex 10	c.1231A>G	p.Lys411Glu	Unknown	Reported	1	0.26
<b>96<sup>[41]</sup></b>	Ex 10	c.1240C>T	p.Gln414X	CF-causing	Reported	<b>1</b>	0.26
<b>97</b>	<b>Ex 10</b>	<b>c.1265C&gt;T</b>	<b>p.Ser422Phe</b>	<b>Unknown</b>	<b>Novel</b>	<b>1</b>	<b>0.26</b>
<b>98</b>	<b>Ex 10</b>	<b>c.1347_1350delAGAA</b>	<b>p.Arg450AspfsX1 8</b>	<b>Suspected</b>	<b>Novel</b>	<b>1</b>	<b>0.26</b>
99 <sup>[38]</sup>	Ex 10	c.1352G>T	p.Gly451Val	Unknown	Reported	1	0.26
<b>100</b>	<b>Ex 10</b>	<b>c.1368delT</b>	<b>p.Ala457LeufsX12</b>	<b>Suspected</b>	<b>Novel</b>	<b>1</b>	<b>0.26</b>

101 <sup>[22]</sup>	Ex 10	c.1373G>A	p.Gly458Glu	Unknown	Reported (Chinese only)	1	0.26
102 <sup>[38]</sup>	In 10	c.1393-4C>A	–	Unknown	Reported (Chinese only)	1	0.26
103 <sup>[13]</sup>	Ex 11	c. 1423delC	p.Leu475TrpfsX52	Suspected	Reported (Chinese only)	1	0.26
104 <sup>[6]</sup>	Ex 11	c.1429_1437delCCTTCA GAG	p.Pro477_Glu479d el	Unknown	Reported (Chinese only)	1	0.26
105	Ex 11	c.1456G>T	p.Gly486X	Suspected	Reported	1	0.26
<b>106</b>	<b>Ex 11</b>	<b>c.1514delA</b>	<b>p.Asn505IlefsX22</b>	<b>Suspected</b>	<b>Novel</b>	<b>1</b>	<b>0.26</b>
<b>107</b>	<b>Ex 11</b>	<b>c.1523_1534delTTGGTG TTTCCT</b>	<b>p.Phe508_Ser511d el</b>	<b>Suspected</b>	<b>Novel</b>	<b>1</b>	<b>0.26</b>
108 <sup>[29]</sup>	Ex 11	c.1526G>C	p.Gly509Ala	Unknown	Reported (Chinese only)	1	0.26
109 <sup>[4]</sup>	Ex 13	c.1699G>T	p.Asp567Tyr	Unknown	Reported (Chinese only)	1	0.26
110 <sup>[10]</sup>	Ex 13	c.1716C>A	p.Asp572Glu	Unknown	Reported	1	0.26

					(Chinese only)		
111 <sup>[6]</sup>	Ex 13	c.1733T>C	p.Leu578Pro	Unknown	Reported (Chinese only)	1	0.26
112 <sup>[49]</sup>	In 13	c.1766+1G>T	–	CF-causing	Reported	1	0.26
113	In 13	c.1766+2T>C	–	Unknown	Reported	1	0.26
<b>114</b>	<b>Ex 14</b>	<b>c.1772T&gt;C</b>	<b>p.Val591Ala</b>	<b>Unknown</b>	<b>Novel</b>	<b>1</b>	<b>0.26</b>
115 <sup>[20]</sup>	Ex 14	c.1997T>G	p.Leu666X	Suspected	Reported (Chinese only)	1	0.26
<b>116</b>	<b>Ex 14</b>	<b>c.2042A&gt;T</b>	<b>p.Glu681Val</b>	<b>Unknown</b>	<b>Novel</b>	<b>1</b>	<b>0.26</b>
117 <sup>[38]</sup>	Ex 14	c.2052delA	p.Lys684AsnfsX38	CF-causing	Reported	1	0.26
118 <sup>[12]</sup>	Ex 14	c.2052dupA	p.Gln685ThrfsX4	CF-causing	Reported	1	0.26
<b>119</b>	<b>Ex 14</b>	<b>c.2058_2061delTT</b>	<b>p.Phe687X</b>	<b>Suspected</b>	<b>Novel</b>	<b>1</b>	<b>0.26</b>
120 <sup>[6]</sup>	Ex 14	c.2236_2246delGAGGCG ATACTinsAAAAATC	p.Glu746LysfsX8	Suspected	Reported (Chinese only)	1	0.26
<b>121</b>	<b>Ex 14</b>	<b>c.2328dupA</b>	<b>p.Val777SerfsX2</b>	<b>Suspected</b>	<b>Novel</b>	<b>1</b>	<b>0.26</b>
122 <sup>[10]</sup>	Ex 14	c.2353C>T	p.Arg785X	CF-causing	Reported	1	0.26
123	Ex 14	c.2475_2478dupCGAA	p.Glu827ArgfsX10	Suspected	Reported	1	0.26

					(Chinese only)		
<b>124</b>	<b>Ex 14</b>	<b>c.2489dupA</b>	<b>p.Glu831GlyfsX5</b>	<b>Suspected</b>	<b>Novel</b>	<b>1</b>	<b>0.26</b>
125	In 14	c.2491-2A>G	–	Unknown	Reported	1	0.26
126	In 15	c.2619+2T>A	–	Unknown	Reported	1	0.26
127	In 16	c.2657+5G>A	–	CF-causing	Reported	1	0.26
128 <sup>[32]</sup>	Ex 17	c.2805delA	p.Pro936HisfsX6	Suspected	Reported (Chinese only)	1	0.26
129	Ex 17	c.2812G>T	p.Val938Leu	Unknown	Reported	1	0.26
130 <sup>[20]</sup>	Ex 17	c.2907A>C	p.A969A	Unknown	Reported (Chinese only)	1	0.26
<b>131</b>	<b>Ex 18-21</b>	<b>c.2909-?_3468+?del</b>	–	<b>Suspected</b>	<b>Novel</b>	<b>1</b>	<b>0.26</b>
132	Ex 18	c.2936A>C	p.Asp979Ala	Unknown	Reported	1	0.26
133	Ex 18	c.2977G>T	p.Asp993Tyr	Unknown	Reported	1	0.26
134 <sup>[17]</sup>	In 18	c.2988+2T>C	–	Unknown	Reported (Chinese only)	1	0.26
135 <sup>[10]</sup>	Ex 19	c.2997_3000delAATT	p.Ile1000X	Suspected	Reported	1	0.26
136 <sup>[38]</sup>	Ex 19	c.3123dupA	p.Gln1042ThrfsX5	Suspected	Reported	1	0.26

<b>137</b>	<b>Ex 20</b>	<b>c.3140-?_3367+?del</b>	–	<b>Suspected</b>	<b>Novel</b>	<b>1</b>	<b>0.26</b>
138	Ex 20	c.3176T>G	p.Leu1059X	Suspected	Reported	1	0.26
139 <sup>[40]</sup>	Ex 20	c.3197G>A	p.Arg1066His	CF-causing	Reported	1	0.26
140	Ex 20	c.3209G>C	p.Arg1070Pro	Unknown	Reported	1	0.26
141 <sup>[4]</sup>	Ex 20	c.3307delA	p.Ile1103X	Suspected	Reported (Chinese only)	1	0.26
142	Ex 21	c.3387delT	p.Gly1130ValfsX4	Suspected	Reported	1	0.26
143 <sup>[18]</sup>	Ex 21	c.3406G>A	p.Ala1136Thr	Unknown	Reported	1	0.26
<b>144</b>	<b>In 21</b>	<b>c.3469-12T&gt;G</b>	–	<b>Unknown</b>	<b>Novel</b>	<b>1</b>	<b>0.26</b>
<b>145</b>	<b>In 21</b>	<b>c.3469-2A&gt;T</b>	–	<b>Unknown</b>	<b>Novel</b>	<b>1</b>	<b>0.26</b>
146	Ex 22	c.3472C>T	p.Arg1158X	CF-causing	Reported	1	0.26
147	Ex 22	c.3476C>T	p.Ser1159Phe	CF-causing	Reported	1	0.26
148 <sup>[11]</sup>	Ex 22	c.3653_3656dupAATA	p.Tyr1219X	Suspected	Reported (Chinese only)	1	0.26
<b>149</b>	<b>Ex 22</b>	<b>c.3659C&gt;T</b>	<b>p.Thr1220Ile</b>	<b>Unknown</b>	<b>Novel</b>	<b>1</b>	<b>0.26</b>
150 <sup>[15]</sup>	Ex 22	c.3691delT	p.Ser1231ProfsX4	CF-causing	Reported	1	0.26
151 <sup>[6]</sup>	Ex 23	c.3718-?_3873+?del	–	Suspected	Reported	1	0.26

					(Chinese only)		
152	Ex 23	c.3841C>T	p.Gln1281X	Suspected	Reported	1	0.26
153 <sup>[6]</sup>	Ex 23	c.3860delG	p.Gly1287GlufsX2	Suspected	Reported (Chinese only)	1	0.26
154 <sup>[10]</sup>	Ex 24	c.3883_3886delATTT	p.Ile1295PhefsX32	CF-causing	Reported	1	0.26
155	Ex 24	c.3908delA	p.Asn1303ThrfsX2 5	CF-causing	Reported	1	0.26
156 <sup>[4]</sup>	Ex 24	c.3909C>G	p.Asn1303Lys	CF-causing	Reported	1	0.26
157 <sup>[6]</sup>	In 24	c.3964-7A>G	–	Unknown	Reported (Chinese only)	1	0.26
158 <sup>[4]</sup>	3'UTR	c.*110C>G	–	Unknown	Reported (Chinese only)	1	0.26

HGVS, Human Genome Variation Society.

Novel variants are formatted in bold.

e-Table 3. Demographic and clinical features of 24 Chinese CF patients with novel variants

Case No.	Sex	Age at Dx, y	Age of symptom onset, y	Weight at Dx, kg (centile)	Clinical presentation	Sputum pathogens	Fecal Sudan III stain	Sweat conductivity, mmol/L	Genotype	Distribution (Province)
33	M	7.33	7.00	18.0 (<3rd)	Bronchiectasis/Recurrent pneumonia/ABPA/Sinusitis/Steatorrhea/Liver disease/FTT/Finger clubbing	<i>P. aeruginosa/S. aureus</i>	+	125	<b>c.2328dupA/c.3209G&gt;C</b>	Hebei
36	F	10.67	5.00	37.5 (50-75th)	Bronchiectasis/Recurrent pneumonia/Sinusitis/Liver disease/Diabetes/Finger clubbing	<i>H. influenzae/P. aeruginosa/S. aureus</i>	–	127	<b>c.2058_2061delT T/c.595C&gt;T</b>	Shanxi

38	M	13.25	7.00	30.0 7 (<3rd)	Bronchiectasis/Recurrent pneumonia/ABPA/Asthma/Hemoptysis/Sinusitis/Nasal polyps/FTT/Pancreatitis/Finger clubbing	<i>P. aeruginosa/S. aureus/A. fumigatus/A. niger</i>	+	109	<b>c.1347_1350delA GAA/c.2909G&gt;A</b>	Zhejiang
46	M	10.17	7.00	27.0 (10th)	Bronchiectasis/ Asthma/Sinusitis	–	–	60	<b>c.2042A&gt;T/c.374 T&gt;C</b>	Yunnan
54	F	8.58	5.00	27.0 (25- 50th)	Bronchiectasis/Sinusitis/Nasal polyps	<i>P. aeruginosa</i>	–	125	<b>c.1810A&gt;C/c.165 7C&gt;T</b>	Shanghai
56	M	2.08	1.00	9.5 (<3rd)	Bronchiectasis/Recurrent pneumonia/Sinusitis/Steatorrhea/FTT/Finger clubbing	<i>P. aeruginosa</i>	+	111	<b>c.3469-2A&gt;T/c.1766+2T &gt;C</b>	Henan
57	F	9.42	5.00	26.0 (10- 25th)	Bronchiectasis/Recurrent pneumonia/Sinusitis/Pancreatitis/Finger clubbing	<i>P. aeruginosa/A. fumigatus/A. flavus</i>	–	108	<b>c.1368delT/c.387 4-4522A&gt;G</b>	Yunnan
58	F	8.75	2.00	19.0	Bronchiectasis/Recurrent	<i>P. aeruginosa/C.</i>	+	136	<b>c.1393-?_1584+?</b>	Henan



				(<3rd)	pneumonia/Sinusitis/Steatorrhea/Rectal prolapse/Liver disease/FTT/Finger clubbing	<i>Albicans</i>			<b>del/c.1523_1534delTTGGTGTTT CCT</b>	
59	F	7.75	0.75	21.0 (10-25th)	Bronchiectasis/Recurrent pneumonia/ABPA/Sinusitis/Finger clubbing	<i>P. aeruginosa/S. maltophilia</i>	-	97	<b>c.1265C&gt;T/c.2977G&gt;T/c.3140-26A&gt;G</b>	Shandong
61	F	4.17	1.00	12.0 (<3rd)	Bronchiectasis/Recurrent pneumonia/Sinusitis/Meconium ileus/Steatorrhea/Rectal prolapse/Liver disease/FTT/Finger clubbing	<i>P. aeruginosa/S. aureus/C. lusitaniae</i>	+	ND	<b>c.1219G&gt;T/c.2551C&gt;T/c.-152G&gt;C</b>	Zhejiang
63	F	13.75	5.00	29.0 (<3rd)	Bronchiectasis/Recurrent pneumonia/Sinusitis/Pancreatitis/Steatorrhea/FTT/Finger clubbing	<i>P. aeruginosa/A. flavus</i>	+	ND	<b>c.1810A&gt;C/c.940G&gt;T/c.*133delT</b>	Zhejiang

67	F	12.00	9.00	47.6 (75-90th)	Bronchiectasis/Sinusitis/ABPA/Wheeze	<i>P. aeruginosa/A. terreus</i>	–	77	<b>c.298C&gt;T/c.3841C&gt;T</b>	Anhui
71	F	13.25	8.00	35.0 (3rd-10th)	Bronchiectasis/Sinusitis/FTT/Finger clubbing	<i>P. aeruginosa/C. Albicans</i>	–	122	<b>c.3140-?_3367+?del/c.233dupT</b>	Shandong
72	F	7.33	1.00	19.0 (3rd-10th)	Bronchiectasis/Sinusitis/FTT	<i>K. pneumoniae/S. aureus</i>	–	118	<b>c.1393-?_1584+?del/c.1000C&gt;T</b>	Shanxi
75	F	8.58	0.58	19.0 (<3rd)	Bronchiectasis/Recurrent pneumonia/ABPA/Sinusitis/Steatorrhea/Diabetes/FTT/Finger clubbing	<i>P. aeruginosa/A. fumigatus/C. Albicans</i>	+	146	<b>c.222delG/c.263T&gt;G</b>	Jiangxi
77	F	4.50	4.00	19.0 (75th)	Atelectasis/Asthma/Sinusitis	–	–	60	<b>c.298C&gt;T/c.650A&gt;G</b>	Jiangsu
78	F	12.92	10.00	44.0 (50th)	Bronchiectasis/Hemoptysis/Sinusitis/Nasal polyps/Finger clubbing	<i>P. aeruginosa/M. abscessus/A. terreus/C. Albicans</i>	–	119	<b>c.3469-12T&gt;G/c.3476C&gt;T</b>	Liaoning

80	F	10.67	6.00	33.0 (25-50th)	Bronchiectasis/ABPA/Asthma/Sinusitis/Pancreatitis	<i>P. aeruginosa/S. aureus</i>	-	126	<b>c.2909-?_3468+?del/c.2909G&gt;A</b>	Henan
85	F	11.00	10.00	30.5 (10-25th)	Bronchiectasis/Sinusitis/Appendicitis	<i>P. aeruginosa</i>	-	130	<b>c.2489dupA/c.233dupT</b>	Anhui
87	M	12.50	0.17	31.0 (3rd)	Bronchiectasis/Recurrent pneumonia/Sinusitis/Steatorrhea/Liver disease/FTT/Finger clubbing	<i>S. aureus/C. Albicans</i>	+	124	<b>c.1514delA/c.263T&gt;G</b>	Sichuan
88	F	5.67	0.25	23.0 (90th)	Bronchiectasis/Asthma/Sinusitis	<i>S. pneumoniae/C. Albicans</i>	-	95	<b>c.3659C&gt;T/c.374T&gt;C</b>	Henan
94	M	7.17	6.00	19.0 (3rd)	Bronchiectasis/Sinusitis/FTT/Finger clubbing	<i>P. aeruginosa/C. Albicans</i>	-	ND	<b>c.1772T&gt;C/c.274-?_1584+?del)</b>	Henan
96	F	3.75	3.00	13.0 (3rd-10th)	Bronchiectasis/ABPA/Sinusitis/FTT	-	-	ND	<b>c.464C&gt;G/c.2909G&gt;A</b>	Henan
102	M	0.42	0.08	8.0 (50th)	Pseudo-Bartter syndrome	-	-	103	<b>c.1064C&gt;G/c.1210-3C&gt;G</b>	Henan

Abbreviations: *A. fumigatus* = *Aspergillus fumigatus*; *A. flavus* = *Aspergillus flavus*; *A. niger* = *Aspergillus niger*; *A. terreus* = *Aspergillus terreus*; *C. Albicans* = *Candida Albicans*; *C. lusitaniae* = *Candida lusitaniae*; *H. influenzae* = *Haemophilus influenzae*; *K. pneumonia* = *Klebsiella pneumoniae*; *M. abscessus* = *Mycobacterium abscessus*; *MRSA* = *Methicillin-resistant Staphylococcus aureus*; *P. aeruginosa* = *Pseudomonas aeruginosa*; *S. aureus* = *Staphylococcus aureus*; *S. maltophilia* = *Stenotrophomonas maltophilia*; *S. pneumoniae* = *Streptococcus pneumoniae*; Dx = Diagnosis; FTT = Failure to thrive; ND = Not done; Y = Years.

Novel variants are formatted in bold.