Mutation Panel (mutations highlighted in green are recommended by the ACMG/ACOG)	Mutation frequencies among individuals with clinically diagnosed cystic fibrosis (%)				
	Caucasian	Hispanic American	African American	Asian American	Ashkenazi Jewish
ΔF508	72.42	54.38	44.07	38.95	31.41
ΔI507	0.88	0.68	1.87	0	0.22
G542X	2.28	5.1	1.45	0	7.55
G85E	0.29	0.23	0.12	0	0
R117H	0.7	0.11	0.06	0	0
621+1G>T	1.57	0.26	1.11	0	0
711+1G>T	0.43	0.23	0	0	0.1
R334W	0.14	1.78	0.49	0	0
R347P	0.45	0.16	0.06	0	0
A455E	0.34	0.05	0	0	0
1717-1G>A	0.48	0.27	0.37	0	0.67
R560T	0.38	0	0.17	0	0
R553X	0.87	2.81	2.32	0.76	0
G551D	2.25	0.56	1.21	3.15	0.22
1898+1G>A	0.16	0.05	0.06	0	0.1
2184delA	0.17	0.16	0.05	0	0.1
2789+5G>A	0.48	0.16	0	0	0.1
3120+1G>A	0.08	0.16	9.57	0	0.1
R1162X	0.23	0.58	0.66	0	0
3569delC	0.34	0.13	0.06	0	0
3849+10kbC>T	0.58	1.57	0.17	5.31	4.77
W1282X	1.5	0.63	0.24	0	45.92
N1303K	1.27	1.66	0.35	0.76	2.78
Mutation Detection Rate for ACMG/ACOG mutations[a]	**88.29[a]**	**71.72[a]**	**64.46[a]**	**48.93[a]**	**94.04[a]**
1078delT	0.02	0.09	0	0	0
394delTT[b]	0.2	NF	NF	NF	NF
Y122X[c]	NA	NA	NA	NA	NA
R347H[a]	0.06	NF	NF	1.6	NF
V520F[a]	0.09	0.04	NF	0.8	NF
A559T[a]	NF	NF	1.41	NF	NF
S549N[a]	0.05	0.66	0.8	3.2	NA
S549R(T>G)[b]	0.1	NF	NF	NF	NF
1898+5G>T[d]	NA	NA	NA	NA	NA
2183AA>G[a]	0.11	NF	NF	NF	NF
2307insA[a]	NF	0.07	0.67	NF	NF
Y1092X[a]	0.11	0.26	0.15	NF	NF
M1101K[b]	0.5	NF	NF	NF	NF
S1255X[b]	NF	NF	1	NF	NF
3876delA[a]	NF	0.48	NF	NF	NF
3905insT[a]	0.13	0.04	0.07	NF	NF

Mutation Detection Rate for xTAG CF39v2 mutations[a]	**89.66**	**73.36**	**68.56**	**54.53**	**94.04**
E60X[a]	0.13	0.11	NF	NF	NF
R75X[e]	NF	1.6	NF	NF	NF
405+3A>C[a]	NF	NF	0.29	NF	NF
406-1G>A[e]	NF	1.6	NF	NF	NF
444delA[b]	NF	NF	1.5	NF	NF
R117C[b]	0.2	NF	NF	NF	NF
G178R[b]	0.2	0.31[f]	NF	NA	NF
L206W[b]	0.1	0.58[g]	NF	NF	NF
935delA[e]	NF	0.8	NF	NF	NF
ΔF311[e]	NF	0.04	0.52	NF	NF
G330X[a]	NF	0.04	0.52	NF	NF
R352Q[b]	0.1	NA	NF	NF	NF
S364P[h]	NA	NA	NA	NA	NA
G480C[e]	NF	NF	1.65	NF	NF
Q493X[a]	0.2	0.04	0.07	NF	NF
1677delTA[b]	0.04	0.8	NF	NF	NF
1812-1G>A[a]	NF	0.04	0.44	NF	NF
G622D[i]	NA	NA	NA	NA	NA
2055del9>A[b]	NF	0.8	0.58[g]	NF	NF
2143delT[b]	0.14	NF	NF	NF	NF
K710X[b]	0.04	NF	NF	NF	NF
3791delC[b]	NF	NF	2	NF	NF
Q890X[a]	NF	0.18	NF	NF	NF
2869insG[j]	NA	NA	NA	NA	NA
3120G>A[b]	0.2	NF	NF	NF	NF
3199del6[b]	NF	0.8	NF	NF	NF
R1066C[a]	0.02	1.9[f]	0.07	NF	NF
W1089X[a]	NF	0.52	NF	NF	1.4[k]
D1152H[a]	0.03	0.1	NF	NF	0.50[l]
R1158X[a]	0.07	0.15	0.74	NF	NF
CFTRdele2,3[m]	NA	NA	NA	NA	NA
S1196X[n]	NA	NA	NA	NA	NA
Mutation Detection Rate (CF 60)	**90.63**	**83.69**	**72.54**	**54.53**	**95.94**
Mutation Detection Rate (CF 71)	**91.13**	**83.77**	**76.94**	**54.53**	**95.94**

CF 71 v2 mutations

a) Mutation frequencies based on the ACMG 2004 Policy Statement (Watson, Cutting et al. 2004).
b) Data from Heim et. al. (Heim, Sugarman et al. 2001).
c) Y122X accounts for about 48 percent of the CF mutations in the Reunion Islands, where Y122X and ΔF508 together account for 70 percent of the CF mutations (Bienvenu, Bousquet et al. 1993). Y122X has not been analyzed in any large North American study, or in any study of CF patients worldwide.
d) The 1898+5G>T mutation has been found in several Chinese and Taiwanese CF patients; however, it has not been analyzed in any large North American study, or in any study of CF patients worldwide (Zielenski, Markiewicz et al. 1995; Wu, Shu et al. 2000; Alper, Shu et al. 2003).
e) Data from Bobadilla et al. (Bobadilla, Macek et al. 2002).
f) Data from Sugarman et al. (Sugarman, Rohlfs et al. 2004).
g) Data from Schrijver et al. (Schrijver, Ramalingam et al. 2005).
h) The frequency of S364P has not been analyzed in any large study in North America, or elsewhere in the world.
i) The frequency of G622D among CF patients is not known; however, in a carrier screening study, the allele frequency of G622D was 0.18%. (Monaghan, Bluhm et al. 2004).
j) The 2869insG mutation has been studied in CF patients in Spain, and was found in 3.1% of the non-ΔF508 mutations in this population (Nunes, Bonizzato et al. 1992).
k) Data from Shoshani et al. (Shoshani, Augarten et al. 1994).
l) Data from Quint et al. (Quint, Lerer et al. 2005).
m) The frequency of the CFTRdele2,3 in the North American population or worldwide is unknown. This mutation is most commonly found in Central and Eastern Europeans, with frequencies ranging from 1.1 to 6.4% (Dork, Macek et al. 2000). The inclusion of this mutation will increase the mutation detection rate in Americans of central and eastern European origin (Bobadilla, Macek et al. 2002).
n) S1196X was first identified in a Russian patient with CF. In a study of 100 CF patients from Russia, S1196X was found in 2.2% of patients. It has not been analyzed in any large North American study, or in any study of CF patients worldwide (Petrova, Kapranov et al. 1997).

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