



PATHOLOGY ASSOCIATES
MEDICAL LABORATORIES

Table 2 – Mutations Detected in PAML CF Carrier Screen

American College of Medical Genetics Recommended 25 Mutation Panel with Reflex Testing			Additional mutations tested
R553X	W1282X	2789+5G→A	S549N
G551D	R334W	1898+1G→A	S549R
ΔI507	1078delT	621+1G→T	V520F
ΔF508	3849+10kbC→T	711+1G→T	3876delA
1717-1G→A	R1162X	G85E	R347H
G542X	N1303K	Reflex	3905insT
R560T	3659delC	I506V	394delTT
3120+1G→A	A455E	I507V	F508C
R347P	R117H	IVS-8 5T	
I148T	2184delA		

Quick Facts

- Cystic Fibrosis is one of the most common autosomal recessive syndromes in Caucasian populations.
- The carrier frequency is as high as 1 in 25 in Caucasian groups.
- The risk of having an affected child is 1 in 4 if both individuals of a couple are CF carriers.
- ACMG and ACOG jointly recommend that CF carrier screening be offered to **all** couples anticipating a pregnancy or seeking prenatal care.