



INTERPRETATION GUIDELINES

Cystic Fibrosis Screening

Indication: Cystic Fibrosis carrier screening

Methods: The CF transmembrane conductance regulator (CFTR) gene was tested for the presence of these specific mutations (and benign variants when indicated) by polymerase chain reactions and allele-specific oligonucleotide hybridization.

38 Mutations Detected by MDL Panel				
3120+1G>A	A455E	G85E	R334W	1717-1G>A
3659ΔC	ΔF508	R347P	1898+1G>A	3849+10kbC>T
ΔI507	N1303K	R553X	2184ΔA	621+1G>T
G542X	R1162X	R560T	2789+5G>A	711+1G>T
G551D	R117H	W1282X	W1282C	R553G
R117L	R117P	K684K	1898+1G>T	R560K
G85V	R347L			
6 CFTR variants included as a reflex test				
I506V	I507V	F508C	5T/7T/9T	I506L
F508S				

Reflex test is performed only when mutations ΔF508 and / or R117H are positive.

Results: One or more mutations detected.

Interpretation: One or more of the assayed mutations was identified indicating that this individual is a carrier for CF. This interpretation is based on the assumption that this individual is not clinically affected with CF. It is recommended that carrier testing by mutation analysis be offered to relatives and reproductive partners of known CF carriers along with appropriate genetic counseling.

This test was developed and its performance characteristics determined by this laboratory. It has not been cleared or approved by the Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary. This test is used for clinical purposes. It should not be regarded as investigational or for research. The laboratory is regulated by the Clinical Laboratory Improvement Act of 1988.

General Disclaimer: DNA studies do not constitute a definitive carrier test for cystic fibrosis in all individuals. Thus, interpretation is given as a probability. It should be realized that there are many possible sources of diagnostic error. Genotyping errors can result from trace contamination of polymerase chain reactions and from rare genetic variants that interfere with analysis. Accurate risk calculation requires accurate family history information. Inaccurate reporting of family history of CF will lead to errors in residual risk assessment.