

REGIONAL PATHOLOGY SERVICES

DEPARTMENT OF PATHOLOGY AND MICROBIOLOGY

Cystic Fibrosis Mutation Detection

Synonym: CF Mutation

Test Overview: This test simultaneously screens for 40 mutations and 4 polymorphisms in the cystic fibrosis transmembrane conductance regulator (CFTR) gene. The 25 CFTR gene mutations and 4 polymorphisms recommended by the American College of Medical Genetics and American College of Obstetricians and Gynecologists in addition to 15 of the world's most common and North American-prevalent mutations. (See Table 1).

ΔF508*	A455E*	3849+10kbC>T*	2183AA>G
Δ1507*	1717-1G>A*	W1282X*	2307insA
G542X*	R560T*	N1303K*	Y1092X
G85E*	R553X*	394delITT	M1101K
R117H*	G551D*	Y122X	S1255X
I148T	1898+1G>A*	R347H	3876delA
621+1G>T*	2184delA*	V520F	3905insT
711+1G>T*	2789+5G>A*	A559T	5/7/9T
1078delT	3120+1G>A*	S549N	F508C
R334W*	R1162X*	S549R (T>G)	1507V
R347P*	3659delC*	1898+5G>T	1506

Clinical Significance: This test is intended for individuals with a negative personal family history for cystic fibrosis.

Method: Multiplex PCR, allele specific primer extension and bead hybridization.

Availability: Once/week; Results 1 week.

Specimen: Blood

Collect: One 5.0 mL EDTA (Lavender) tube.

Volume: 1.0 mL whole blood.

Transport: Refrigerated.

Reference Range: With Report

CPT Code: 83900; 83912; 88384; 83914; 83890

Additional Information: A patient/family history form must accompany each specimen. Call Regional Pathology for the form or refer to the test information on the Test Directory web site. www.reglab.org

Test performed by the Nebraska Medical Center Clinical Laboratory.