



CFTR Full Sequence Analysis

Patient Name: Sample Patient
Specimen #: 1000000-01
Patient ID: 2000000-01
Case #:
Client # 12345

DOB: 5/2/1972 Date Collected: 10/31/06 Referring Physician: John Doe, M.D.
SSN: Date Received: 11/1/06 Genetic Counselor:
Sex: Lab ID #: Additional Report To:
Specimen: Blood Hospital ID #:

Indication: Carrier testing – family history Ethnicity: Hispanic

RESULT: Sequence change detected – see below
PolyT status: 7T, 9T

Sequence change	Classification	
W216X	Mutation	See Interpretation

INTERPRETATION: This analysis identified one CF mutation. This individual is therefore a carrier of cystic fibrosis.

COMMENTS:

The above interpretation is based on the clinical information provided and the current understanding of the molecular genetics of cystic fibrosis. The CFTR full sequencing assay detects ~98% of CFTR mutations. This analysis cannot rule out the presence of large deletion or duplication mutations, complex rearrangements or mutations in regions not analyzed.

Consultation with a physician or genetic counselor is recommended to discuss the potential clinical and/or reproductive implications of this result, as well as recommendations for testing other family members.

METHOD:

DNA is isolated and amplified by the polymerase chain reaction (PCR). The 27 exons of the CFTR gene and their flanking intronic sequences (-6 and +15), as well as regions of CFTR introns 1, 3, and 19, are analyzed by bi-directional direct DNA sequencing using capillary gel electrophoresis and fluorescence detection. Sequence changes are classified into one of 5 categories (mutation, likely mutation, likely benign variant, benign variant, unknown) after analysis of information from sources such as PubMed, PolyPhen, SIFT and the Cystic Fibrosis Mutation Database.

REFERENCES:

PubMed: www.pubmed.gov
PolyPhen: <http://genetics.bwh.harvard.edu/pph/>
SIFT: <http://blocks.fhcrc.org/sift/SIFT.html>
Cystic fibrosis mutation database: <http://www.genet.sickkids.on.ca/cftr/app>

False positive or negative results may occur for reasons that include genetic variants, blood transfusions, bone marrow transplantation, erroneous representation of family relationships or contamination of a fetal sample with maternal cells. This test was developed and its performance characteristics determined by Genzyme Genetics. It has not been cleared or approved by the U.S. Food and Drug Administration. 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 under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high complexity clinical testing.