Cystic Fibrosis Mutation Analysis, CFTR sequencing

CPT Code(s): 81223

Service Code (IU Health): 

Ordering Recommendation: CFTR mutation analysis is recommended for individuals with clinical symptoms of cystic fibrosis; individuals with a positive family history of cystic fibrosis; and individuals planning or having a pregnancy.

Synonyms: CFTR, cystic fibrosis, chronic sinopulmonary disease, congenital bilateral absence of the vas deferens (CBAVD), azoospermia

Methodology: Sanger sequence analysis. The CFTR cDNA reference sequence used is NM_000492.

Performed: Mon-Fri

Reported: 14 days

Specimen Requirements

Collect: Preferred: whole blood in a lavender top (EDTA) tubes, cultured and uncultured cells

Specimen Volume: Blood: 3 mL whole blood (minimum 1 mL)

Storage/Transport: Refrigerated/Room temperature

Unacceptable Conditions: Grossly hemolyzed or clotted

Remarks:

Stability: 1 month refrigerated; 1 month frozen

Reference Interval: by report

Interpretive Data
Characteristics: CFTR-related disorders include cystic fibrosis (CF) and congenital bilateral absence of the vas deferens (CBAVD). CF is characterized by chronic sinopulmonary disease, gastrointestinal/nutritional abnormalities, obstructive azoospermia and salt loss syndromes. Males can present CBAVD without presenting the traditional characteristics of CF. Affected males develop azoospermia and are infertile.

Inheritance: autosomal recessive

Cause: CFTR is the only gene known to be associated with the CFTR-related disorders, CF and CBAVD.

Incidence: Classic CF occurs in one in 2,500 Caucasians, one in 13,500 Hispanics, one in 15,000 African Americans and one in 31,000 Asian Americans

Penetrance: Cystic fibrosis is 100% penetrant.

Analytical sensitivity and specificity: 99%

Limitations: It should be noted that only the coding and immediate flanking regions of the CFTR gene are analyzed by DNA sequencing. Changes in the promoter region and other non-coding regions will therefore not be detected by our assay. In addition, the presence of a large intragenic deletion of the CFTR gene (such as the deletion of an exon) will not be detected by sequence analysis. All results should be interpreted in context of clinical findings, relevant history, and other laboratory data.

References: Gene Reviews, Gene Cards, OMIM, CFTR2 database