Cystic Fibrosis Mutation Analysis, CFTR deletion/duplication

CPT Code(s): 81222

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: multiplex ligation-dependent probe amplification (MLPA)

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
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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 CAVD.

Incidence: Classic CF occurs in one in 2,500 Caucasians, on 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: Deletions/duplications in the promoter region and other non-coding regions of the CFTR gene, as well as partial deletions/duplications of the exons tested may not be detected by our assay. Although rare, false positive or false negative results may occur. All results should be interpreted in context of clinical findings, relevant history, and other laboratory data.

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