Cystic Fibrosis, CFTR, Sequencing

Test Code: 1222

Test Synonyms:
- CF
- CFTR-Related CBAVD
- CFTR-Related Disorders
- Cystic fibrosis transmembrane conductance regulator
- CF carrier screening

CPT Code(s):
- 83891
- 83892 x 28
- 83898 x 28
- 83904 x 28
- 83909 x 28

Background:
Cystic Fibrosis (CF) is the most common autosomal recessive genetic disease in the Caucasian population appearing in approximately one in every 2,500 newborns. In the Caucasian and Ashkenazi Jewish populations, about one in every 25 individuals carries one copy of a mutation in the CFTR gene at 7q31.2.

Reasons for Referral:
- Confirmation of clinical diagnosis
- Carrier identification in persons with a positive or negative family history
- Sperm & egg donors
- Abnormal fetal ultrasound
- Preconception and carrier screening

Methodology:
**Sequencing:** Genomic DNA is used for PCR amplification of all 27 exons contained in the coding region of CFTR. Primers for each exon flank the respective intron/exon junctions. The region analyzed for the final exon uses a primer set that flanks the intron/exon junction at the 5' end and the translation stop codon at the 3' end. All PCR products were sequenced in both the forward and reverse directions. The sensitivity is approximately 99% for the detection of nucleotide base changes, small deletions, and small insertions in the regions analyzed.

Test reporting follows the ACMG Standards & Guidelines for Clinical Genetics Laboratories, Ultra-Rare Disorders Guidelines, and Interpretation of Sequence Variants Guidelines.

Specimen Requirements:
- **Blood:** Lavender tube (EDTA) or yellow (ACD)
  - Adult: 6.0 mL
  - Child: 6.0 mL
  - Infant: 2.0 mL

A REQUISITION FORM MUST ACCOMPANY ALL SAMPLES. Please include detailed clinical information, including ethnicity, clinical history, and family history.
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Test Performed (Days):
Weekly

Turn Around Time:
14 – 21 Days

Shipment Sensitivity Requirements:
Keep specimen cool during transit, but do not ship on dry ice. Please use the cold pack provided in the KDL shipping kit. Ship the specimen overnight express, using the FedEx priority overnight label provided. The specimen must arrive at the lab no more than 24 hours after collection.

Additional Information:
Prior to any genetic testing we recommend genetic counseling. To receive forms and information about prenatal diagnostic testing, please contact Client Services at (855) 535-1522.

Please refer to the American College of Medical Genetics for more information on the Standard and Guidelines for CFTR testing.
http://www.acmg.net/AM/Template.cfm?Section=Laboratory_Standards_and_Guidelines&Template=/CM/ContentDisplay.cfm&ContentID=6328

References:
American College of Medical Genetics Standard and Guidelines for CFTR testing.
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