

Cystic Fibrosis (CF) – Prenatal Familial Variant Analysis

ORDER NAME

Cystic Fibrosis Prenatal Familial Variant Analysis (EPIC Order Code=LAB11184)

- If EPIC is not an option, a paper requisition can be found on the “Clinical Genomics Requisition and Consent” forms page of the website. Complete and submit paper requisition to Genomicslab@upmc.edu.

BACKGROUND

Cystic fibrosis (CF) is an autosomal recessive disorder affecting the respiratory tract, exocrine pancreas, intestine, male genital tract, hepatobiliary system, and exocrine sweat glands. Pulmonary disease is the major cause of morbidity and mortality, and pancreatic insufficiency with malabsorption occurs in the majority.

The cystic fibrosis prenatal familial variant analysis test determines if an individual or fetus is affected with cystic fibrosis (CF).

This test can be conducted postnatally or prenatally, utilizing amniocytes or chorionic samples. If the clinical indication is diagnosis of a pregnancy, and screening tests of the parents were performed outside of the UCGL laboratory, collection of maternal and paternal samples is required to confirm familial variants. Maternal Cell Contamination (MCC) studies are also required.

INDICATIONS FOR TESTING

- Abnormal fetal ultrasound suspicious of a diagnosis of CF
- Confirm a suspected diagnosis of CF

TEST METHODOLOGY

Targeted familial variant testing was performed via PCR amplification of the specific genomic region of interest, followed by bi directional Sanger sequencing. Only the region(s) containing the familial variant were analyzed; other parts of the gene were not assessed. Variant analysis was performed using the reference sequence *CFTR* NM_000492.4. Sequence variants were interpreted according to ACMG/AMP guidelines and reported using the standardized nomenclature recommended by the Human Genome Variation Society (HGVS).

TEST LIMITATIONS

This test detects only the specified *CFTR* variants and does not detect other sequence variants, large deletions or duplications, or rearrangements within the *CFTR* gene. While the test is highly accurate, rare errors may lead to false positive or false negative results. Contributing factors may include misrepresentation of familial relationships, rare single nucleotide polymorphisms (SNPs) affecting primer binding, sample contamination, or other causes.

SPECIMEN REQUIREMENTS

- Postnatal: Whole blood – EDTA tube required, 3-5 ml
- Prenatal: Amniocytes or chorionic samples
 - Maternal Cell Contamination studies: Whole blood – EDTA tube required, 3-5 ml



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TURNAROUND TIME

14 days

CPT CODE

81221