Cystic Fibrosis (CF) – Diagnostic testing

ORDER NAME
Cystic Fibrosis, DNA Analysis, Fetus (EPIC Order Code=LAB13433)

- If EPIC is not an option, complete and submit a paper requisition to Genomicslab@upmc.edu
  Paper requisition can be found on the INFONET. Search for “genetics requisition form.”

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 diagnostic test determines if an individual or fetus is affected with cystic fibrosis (CF).

This diagnostic 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

RESULT
Affected individuals have two disease-causing variants, one from each parent.

METHOD
Polymerase Chain Reaction (PCR) followed by Sanger Sequencing or mass spectrometry.

LIMITATIONS
This test detects only the CFTR variant (s) specified by the family. It will not detect other sequence variants outside of the exon(s) tested, large deletions or duplications, or rearrangements in the CFTR gene.

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

TURNAROUND TIME
14 days

CPT CODE
81221