Quarterly HOTLINE: Effective February 20, 2018

2013662 Cystic Fibrosis (*CFTR*) 165 Pathogenic Variants, Fetal CF VAR FE

Specimen Required: Collect: **Fetal Specimen:** Two T-25 flasks of cultured amniocytes at 80 percent confluency. ***If the client is unable to culture amniocytes, this can be arranged by contacting ARUP Client Services at (800) 522-2787.**
Maternal Specimen: Lavender (EDTA), Pink (K_2EDTA), or Yellow (ACD Solution).
Specimen Preparation: **Cultured Amniocytes**: Fill flasks with culture media. Backup cultures must be retained at the client's institution until testing is complete.
Maternal Specimen: Transport 3 mL whole blood. (Min. 1 mL)
Storage/Transport Temperature: **Cultured Amniocytes**: **CRITICAL ROOM TEMPERATURE.** Must be received within 48 hours of shipment due to lability of cells.
Maternal Specimen: Refrigerated.
Remarks: **Maternal sample is recommended for proper test interpretation; order Maternal Cell Contamination, Maternal Specimen.** Patient History Form is available on the ARUP Web site or by contacting ARUP Client Services.
Unacceptable Conditions: **Maternal Specimen**: Plasma or serum. Specimens collected in sodium heparin or lithium heparin tubes.
Stability (collection to initiation of testing): **Fetal Specimen:** Ambient: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable
Maternal Specimen: Ambient: 72 hours; Refrigerated: 2 weeks; Frozen: 1 month

Interpretive Data:

Background information for Cystic Fibrosis (*CFTR*), 165 Pathogenic Variants, Fetal
Characteristics of Classic Cystic Fibrosis (CF): Chronic sino-pulmonary disease, gastrointestinal malabsorption/pancreatic insufficiency, and obstructive azoospermia. Symptoms of a *CFTR*- related disorder are often limited to a single organ system such as isolated pancreatitis, bilateral absence of the vas deferens, nasal polyposis, or bronchiectasis.
Incidence: 1 in 2,300 Ashkenazi Jewish, 1 in 2,500 Caucasians, 1 in 13,500 Hispanics, 1 in 15,100 African Americans, 1 in 35,100 Asians.
Inheritance: Autosomal recessive.
Penetrance: High for severe and moderately severe pathogenic variants, variable for mild pathogenic variants.
Cause of Classic CF: Two severe, or one severe and one moderate, pathogenic *CFTR* variants on opposite chromosomes.
Cause of *CFTR*-Related Disorders: Two pathogenic *CFTR* variants on opposite chromosomes in any of the following combinations; two mild, one mild and one severe or one mild and one moderate.
Pathogenic Variants Tested: Refer to "Additional Technical Information" document.
Clinical Sensitivity: Ashkenazi Jewish 96 percent; Caucasian 92 percent; Hispanic 80 percent; African American 78 percent; Asian American 55 percent.
Methodology: Polymerase chain reaction (PCR) and fluorescence monitoring.
Analytical Sensitivity & Specificity: 99 percent.
Limitations: Diagnostic errors can occur due to rare sequence variations. Only the 165 pathogenic *CFTR* variants and 5T variant will be interrogated.

For quality assurance purposes, ARUP Laboratories will confirm the above result at no charge following delivery. Order Confirmation of Fetal Testing and include a copy of the original fetal report (or the mother's name and date of birth) with the test submission. Please contact an ARUP genetic counselor at (800) 242-2787 extension 2141 prior to specimen submission.

See Compliance Statement C: www.aruplab.com/CS

Note: The CF 165-Variants assay includes the 23 pathogenic CF variants recommended by the American College of Medical Genetics for population carrier screening.