2013661 Cystic Fibrosis (*CFTR*) 165 Pathogenic Variants CF VAR

Interpretive Data:
Background information for Cystic Fibrosis (*CFTR*), 165 Pathogenic Variants:
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 pathogenic variants, variable for moderate and 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: See the "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.

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

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