GENETIC ASSAYS
A Molecular Diagnostics Laboratory

CF Cystic Fibrosis Mutation Detection

GA Test Code	**6262**
Method	***FDA-approved*** xTAG™ (Luminex) - qualitative genotyping *Note: It is the practitioner's responsibility to obtain Informed Consent from the patient prior to collecting/submitting a sample for CF testing.*
Specimens	**EDTA or ACD Whole Blood**: 5.0 mL (2.0 mL), refrigerated. Store up to 7 days refrigerated (4 days if ambient). **ThinPrep**: 2.0 mL (1.0 mL), store and ship ambient (up to 3 months). **SurePath**: 1.0 mL (0.5 mL), store and ship ambient (14 days). **Buccal Swabs**: 2 swabs, place in original wrapper, transport in speci-guard bag. Store and ship ambient (21 days). GA provides buccal swab collection kits. **G Swab®:** G Swab kits are provided by GA. Collect vaginal specimen with swab and place in tube with liquid media. Break-off swab (pre-scored) and seal tube for transport. Sample is stable for 30 days at room temperature (15-30°C). **Others**: Please contact GA with questions regarding other specimen types.
Causes for Rejection	Time and/or temperature instructions not followed as specified; buccal swabs if not in original wrapper; whole blood if in heparin or if hemolysis occurred.
Reference Range	Interpretive Report
Turnaround Time	Once per week (usually Wednesday)
CPT Code	81220

Description

The xTAG™ Cystic Fibrosis Kit is an FDA-approved, in vitro diagnostic, qualitative genotyping test that provides information intended to be used for carrier testing in adults of reproductive age, as an aid in newborn screening, and in confirmatory diagnostic testing in newborns and children. It is not intended for use in fetal diagnostic or pre-implementation testing and is not indicated for stand-alone diagnostic purposes.

Clinical Utility

This assay is used to determine affected or carrier status of the 39 most common Cystic Fibrosis (CF) mutations and 4 associated polymorphisms. This panel provides approximate mutation detection rates for the following populations: 90.5 for North American Caucasians, 73.8 for Hispanic Americans, 67.5 for African Americans, 48.9 for Asian Americans, and 94.0 for Ashkenazi Jewish population. The American Congress of Obstetricians and Gynecologists (ACOG) now recommends that Ob-Gyns make DNA screening for CF available to all couples seeking preconception or prenatal care - not just those with a personal family history of carrying the CF gene, as previously recommended. ACOG recommends a screening panel of 23 common CF mutations and 4 associated polymorphisms. CF is one of the most common inherited diseases in the United States, affecting 1 in 3,200 live births. If both parents are carriers, then their child has a 25% (1 in 4) chance of being born with CF; a 50% chance that the child will not have CF but will be a carrier; and a 25% chance that the child will not be a carrier. Those affected have high levels of sodium chloride (salt) in their sweat. More noticeably, a thick, sticky mucous in the lungs causes persistent coughing, wheezing and frequent lung infections, including pneumonia.

Laboratory Standards and Guidelines for Population-based Cystic Fibrosis Carrier Screening,*Genetics in Medicine*,March/April 2001,Vol3 No.2:149-154.

Watson, M.S., G.R. Cutting, et al. (2004). Cystic Fibrosis population carrier screening: 2004 revision of American College of Medical Genetics mutation panel. Genet Med 6(5): 387-391.

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