HENRY FORD HEALTH
CENTER FOR PRECISION DIAGNOSTICS

Cystic Fibrosis

Cystic Fibrosis testing is used for the determining carrier status and confirmation of a diagnosis of cystic fibrosis. It also provides additional information in patients with features of atypical CF including equivocal sweat chloride values, congenital absence of the vas deferens and nasal polyps or pancreatitis.

Testing Method and Background

This test utilizes **Next Generation Sequencing (NGS) technology,** which provides coverage of all coding exons and noncoding DNA in exon flanking regions (on average 50 bp) enriched using hybrid capture methodology. This assay can detect >99% of described mutations in the included genes, when present, including single nucleotide variants (point mutations), small insertions/deletions (1-25 bp), larger deletions and duplication (<100 bp), complex insertions/deletions, splice site mutations, whole-gene deletions/duplications and exon-level intragenic deletions/insertions in each gene targeted for analysis. All reportable copy number variants are confirmed by independent methodology.

Cystic fibrosis (CF) is a multisystem disease affecting epithelia of the respiratory tract, exocrine pancreas, intestine, hepatobiliary system, and exocrine sweat glands. Pathogenic variants in CFTR gene are causative of classic CF as well as nonclassic CF phenotypes, including isolated congenital absence of the vas deferens (CAVD). The diagnosis of CF is established in individuals with elevated immunoreactive trypsinogen on newborn screen, signs and/or symptoms suggestive of CF, or family history of CF; and evidence of an abnormality in cystic fibrosis transmembrane conductance regulator (CFTR) function, biallelic CFTR CF-causing pathogenic variants, or nasal transmembrane epithelial potential difference measurement consistent with CF. The majority of CFTR pathogenic variants are sequence variants and small deletions and duplications (approximately 97-98%) while large deletions and duplications encompassing the entire gene as well as adjacent genes or intragenic deletions of one or more exons account for a smaller fraction of mutations in CFTR that are known to be clinically significant (approximately 2-3%).

Highlights of Cystic Fibrosis Testing

Targeted Region

CFTR

- **Wide-ranging Coverage of Variants**
  Detects and provides coverage of all coding exons and noncoding DNA in exon flanking regions.
- **Accurate Results Using Clinically Validated Computational Data Analysis**
  A variety of mutation types (point, indels and duplications) are confirmed using computational data analysis for sequence variant calling, filtering and annotation.

Ordering Information

Get started (non-HFHS): Print a Germline Testing requisition form online at www.HenryFord.com/HFCPD

Get started (HFHS): Order through Epic using test "Cystic Fibrosis" (DNA2100003)

Specimen requirements:

- Peripheral Blood - 1-3ml in lavender top tube (EDTA) **Specimen stability: Ambient - 72 hours; Refrigerated - 1 week**
- Extracted DNA - from a CLIA-certified Laboratory

Cause for Rejection: Clotted, hemolyzed, or frozen specimens, improper anticoagulant, tubes not labeled with dual patient identification, non-dedicated tubes.

TAT: 5-10 business days (after Prior Authorization obtained)

Mail test material to:
Henry Ford Center for Precision Diagnostics
Pathology and Laboratory Medicine
Clinic Building, K6, Core Lab, E-655
2799 W. Grand Blvd., Detroit, MI 48202

CPT Codes: 81220, 81224 (poly T allele, reflex), G0452

Contact us: Client Services, Account and Billing Set-up, and connect with a Molecular Pathologist at (313) 916-4DNA (4362)

For more information on Comprehensive Molecular Services, visit our website www.HenryFord.com/HFCPD

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