# 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 the eSensor® Cystic Fibrosis Genotyping Test which uses a solid-phase electrochemical method for determining the genotyping status of a defined panel of CF mutations. In addition, PolyT (5/7/9T polymorphism) analysis is offered as an adjunct test to the cystic fibrosis mutation screen.

Cystic Fibrosis (CF) is a genetic disease in which defective chloride transport across cellular membranes causes dehydrated secretions. This leads to tenacious mucous in the lungs, mucous plugs in the pancreas and characteristically high sweat chloride levels. CF is inherited as an autosomal recessive disorder, caused by the CFTR gene. Poly T analysis is recommended for all individuals who are identified as positive for the R117H cystic fibrosis mutation, for patients with congenital absence of the vas deferens (unilateral or bilateral) who are either negative or heterozygous for a cystic fibrosis mutation, patients with mild or atypical symptoms of cystic fibrosis in whom one or no cystic fibrosis mutation has been previously identified and for individuals with a history of pancreatitis.

## Highlights of Cystic Fibrosis Testing

**Targeted Region**

**CFTR:** Panel includes mutations and variants recommended by the 2004 American College of Medical Genetics (ACMG).

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|---|---|---|---|---|---|
| 1717-1G>A | 1898+1G>A | 2184delA | 2789+5G>A | R560T | 3659delC |
| 3849+10KbC>T | 621+G>T | 711+1G>T | A455E | R117H | ΔI507 (DeltaI507) |
| R553X | G542X | G551D | G85E | ΔF508 (DeltaF508) | N1303K |
| R1162X | R334W | R347P | 3120+1G>A | W1282X | |

- PolyT (5/7/9T polymorphism) analysis is offered as an adjunct test.

## Ordering Information

**Get started (non-HFHS):** Print a Genetic Hereditary Disorder requisition form online at www.HenryFord.com/HFCPD

**Get started (HFHS):** Order through Epic using test "Cystic Fibrosis" (DNA2100003MOL)

**Specimen requirements:**

- Peripheral Blood - 1-3ml in lavender top tube (EDTA) **Specimen stability: Ambient - 72 hours; Refrigerated - 1 week**
- Saliva specimen - Oragene self-collection kit
- 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:** 5-7 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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