Hereditary Hemochromatosis (HFE)

This test is used to diagnose the hereditary form of hemochromatosis and hemochromatosis carrier state and to confirm diagnosis in equivocal cases. Testing includes molecular genotyping of the hereditary hemochromatosis mutations, C282Y and H63D. Most individuals with HFE-HHC are either homozygotes for the C282Y mutation (85%) or compound heterozygotes for the mutations C282Y and H63D (<10%).

Testing Method and Background

This test utilizes RFLP (Restriction Fragment Length Polymorphism). PCR is performed on genomic DNA and the amplicon is digested with restriction endonuclease (enzyme). DNA fragments are separated by gel electrophoresis to determine the genotype. The following HFE mutations are tested with this method: C282Y (synonyms: Cys282Tyr; nucleotide 845G>A) and H63D (synonyms: His63Asp; nucleotide 187C>G).

The Hemochromatosis gene is also known as HFE. HFE-associated hereditary hemochromatosis (HFE-HHC) is characterized by inappropriately high absorption of iron by the gastrointestinal mucosa, resulting in excessive storage of iron (particularly in the liver, skin, pancreas, heart, joints and testes). Abdominal pain, weakness, lethargy and weight loss are early symptoms. Without therapy, males may develop symptoms between 40 and 60 years of age and females after menopause. Hepatic fibrosis or cirrhosis may occur in untreated individuals after the age of 40 years. Other findings in untreated individuals may include progressive increase in skin pigmentation, diabetes mellitus, congestive heart failure and/or arrhythmias, arthritis and hypogonadism.

Highlights of Hereditary Hemochromatosis (HFE) Testing

Targeted Region

C282Y: Cys282Tyr; nucleotide 845G>A genotyping

H63D: His63Asp; nucleotide 187C>G genotyping

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 "Hereditary Hemochromatosis (HFE) Testing" (DNA2100007)

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

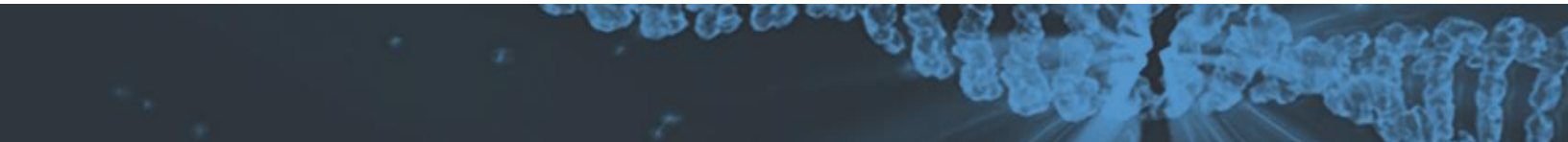
Cause for Rejection: Clotted, hemolyzed, or frozen specimens, improper anticoagulant, tubes not labeled with dual patient identification, non-dedicated tubes.

TAT: 5-7 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: 81256, 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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