

HEMOCHROMATOSIS

DESCRIPTION

Mendelian Inheritance in Man number: [235200](#)

Click here for [GeneTests Review](#)

Hemochromatosis is one of the most common genetic disorders in the United States. About 1:8-12 individuals are carriers for this condition, and about .5% of the Caucasian population is homozygous for mutant alleles, making them much more likely to developing the disease. Hereditary hemochromatosis associated with the *HFE* gene is characterized by increased storage of iron in the liver and other organs. Individuals with *HFE* associated hemochromatosis may experience joint pain, lethargy, fatigue, abdominal pains, and cardiac problems. In men, symptoms first begin to manifest between the ages of 30 and 50, and in women over the age of 50. Men are also 5 times more likely to be diagnosed with hereditary hemochromatosis. *HFE* associated hemochromatosis is inherited in an autosomal recessive manner, meaning an individual must have both copies of the gene mutated for symptoms to appear.

INDICATIONS FOR TESTING

- Family history of hemochromatosis
- Adults with serum transferrin-iron saturation higher than 45%

TESTING METHODOLOGY

The hemochromatosis molecular genetic test is a multiplex PCR technique for the simultaneous detection of two mutations, C282Y and H63D in the *HFE* gene and is based on the method of [Stott et al., 1999](#). 87% of individuals of European descent with hemochromatosis have some combination of these two disease causing mutations. This technique allows complete determination of the patient's genotype in a single reaction.

SPECIMEN REQUIREMENTS

We require a minimum of 1 milliliter of whole blood. Blood samples must be collected in EDTA (purple topped) tubes

TRANSPORT

If the specimen is from clinics within UAB or Kirklin Clinic, please call 934-5562 for pickup. If specimens are being sent from some other location, please ship via **UPS Overnight Priority**. Specimens must be packaged to prevent breakage and absorbent material must be included in the package to absorb liquids in the event that breakage occurs. Also, the package must be shipped in double watertight containers (e.g. a specimen pouch + the shipping companies Diagnostic Envelope). Please contact us (Email – mgl@genetics.uab.edu, Phone – 205-934-5562) prior to sample shipment and provide us with the date of shipment and the tracking number of the package.

TURN AROUND TIME: 7-10 working days

CPT CODES AND PRICES: \$200 USD

83891, 83892, 83894, 83900, 83912

Please note that prices listed correspond to institutional rates; please contact the lab for insurance rates.

Payment may be made by Money order, Cashier's check, VISA or MasterCard. We will also bill requesting institutions. Please call for more information.

REQUIRED FORMS

Test Requisition Form

Note: Requests for Molecular Genetic testing will **not** be accepted for the following reasons:

- No label (patients full name and date of collection) on the specimens
- No referring physician's or genetic counselor's names and addresses
- No billing information if being paid for by an institution
- No informed consent

For more information, test requisition forms, or sample collection and mailing kits, please call: 205-934-5562.

REFERENCES

Stott, M.K. et al. Simple multiplex PCR for the simultaneous detection of the C282Y and the H63D hemochromatosis (HFE) gene mutations. *Clin Chem.* 1999 Mar;45(3):426-8