Hereditary Hemochromatosis

Description: Hereditary hemochromatosis (HH) is an autosomal recessive disorder of iron metabolism. Accumulation of excess iron results in damage to multiple organs including the liver, pancreas, heart, joints, and the brain. Individuals may develop cirrhosis of the liver, liver tumors, diabetes, arthritis, and/or heart disease. Two of the most common defects in the gene for hereditary hemochromatosis *(HFE)* are the C282Y and H63D mutations. The carrier frequency of HH is about 1 in 10 for individuals of Northern European descent.

Indications: Diabetes, bronze skin, liver disease, cardiomyopathy, iron storage disease. Family history of hereditary hemochromatosis.

Specimen: At least 2mLs whole blood in lavender top (EDTA) tube. Label tube with patient's name, birth date, and date of collection. Phlebotomist must initial tube to verify patient's identity.

Methodology: DNA is isolated from peripheral blood and is analyzed for two (2) mutations in the HFE gene, which have been associated with hereditary hemochromatosis. The analysis is performed by restriction analysis of PCR-amplified segments of the *HFE* gene: C282Y mutation is detected by restriction with Rsa1, and the H63D mutation is detected with Mbo1.

Sensitivity: The most common mutation, C282Y (Cys282Tyr) is found in the homozygous state in 89-90% of patients with typical hemochromatosis. The homozygous C282Y genotype confers an approximately 50% risk of abnormal serum iron binding. A second mutation, H63D (His63Asp) has also been associated with hemochromatosis. Less than 2 percent (<2%) of compound heterozygotes for C282Y/H63D also have signs of hemochromatosis, and account for approximately 5% of all cases.

Turn-Around Time: 7 days.

Billing and CPT Codes:

Hereditary hemochromatosis Mutation Analysis: 81256

Please call 1-866-450-4198 for any pricing or billing questions.

Results: Each test report includes a detailed interpretation of the genetic findings, the clinical significance of the result, and specific recommendations for clinical management and additional testing, if warranted. Results will be reported to the referring physician or health care provider as specified on the test requisition form.

Shipping Instructions

Please enclose **test requisition** with sample. **All information must be completed before sample can be processed**. Place samples in Styrofoam mailer and ship at room temperature by overnight Federal Express to arrive Monday through Friday.

Ship to:

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Cincinnati, OH 45229
513-636-4474

Cincinnati Children's®

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