Lab Dept:	**Anatomic Pathology**
Test Name:	**HEMOCHROMATOSIS (HFE) SEQUENCING**

General Information

Lab Order Codes:	HHGA
Synonyms:	HFE Gene Analysis; HLA-H Gene; Hereditary Hemochromatosis
CPT Codes:	81256 – HFE gene analysis, common variants
Test Includes:	Detects 2 mutations: C282Y and H63D. Presence of the S65C mutation is reported only when it is observed as part of the C282Y/S65C genotype.

Logistics

Test Indications:	Useful for establishing or confirming the clinical diagnosis of HH (Hereditary Hemochromatosis) in adults, but because of concerns of the overall penetrance of these mutations, HFE genetic testing is not recommended for population screening. This test is also useful for testing of asymptomatic individuals with increase blood test of iron stores. With appropriate genetic counseling, predictive testing of individuals who have a family history of HH (careful consideration should be given to advantages and disadvantages of such testing before testing is performed).
Lab Testing Sections:	Anatomic Pathology – Sendouts
Referred to:	Mayo Medical Laboratories (MML Test: HFE)
Phone Numbers:	MIN Lab: 612-813-6280 STP Lab: 651-220-6550
Test Availability:	Daily, 24 hours
Turnaround Time:	5 – 14 days

Special Instructions:	Specimens must arrive within 96 hours of collection. Must include a "Molecular Genetics Congenital Inherited Diseases Patient Information Sheet" with information including relevant clinical and family history information in order to provide correct interpretation of test results. Also include "Informed Consent Form for DNA Testing" is available (Supply T576). Specimens will be retained at Mayo for 3 months.

Specimen

Specimen Type:	Whole blood
Container:	Lavender top (EDTA) **or** Yellow top (ACD) tube
Draw Volume:	2.5 mL (Minimum: 0.5 mL in microtainer)
Processed Volume:	Same as Draw Volume
Collection:	Routine venipuncture. Mix tubes by gentle inversion.
Special Processing:	Lab Staff: **Do Not** centrifuge. Specimen should be stored and shipped at ambient temperatures. **Specimen must arrive within 96 hours of draw.**
Patient Preparation:	None
Sample Rejection:	Frozen specimens should be avoided. No specimen will be rejected, please include a note to the lab with specimen issues at MML. Children's will reject specimens that are unlabeled or mislabeled.

Interpretive

Reference Range:	An interpretive report will be issued which will indicate whether or not results are consistent with a diagnosis of hereditary hemochromatosis.
Critical Values:	N/A

Limitations:

This assay will not detect all of the mutations that cause hereditary hemochromatosis. Therefore, the absence of a detectable mutation(s) does not rule out the possibility that an individual is a carrier of or affected with this disease.

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in our interpretation of results may occur if information given is inaccurate or incomplete.

Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.

In rare cases, DNA alterations of undetermined significance may be identified.

A previous bone marrow transplant fro an allogenic donor will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.

Because of concerns of the overall penetrance of HFE mutations, HFE genetic testing is not recommended for population screening.

Methodology:

Direct Mutation Analysis by Polymerase Chain Reaction (PCR)

References:

Mayo Medical Laboratories Web Page (August 2015)
(800) 533-1710

Updates:

2/6/2013: CPT update