MAYO CLINIC LABORATORIES

NEW TEST

Notification Date: May 19, 2022
Effective Date: June 28, 2022

Hereditary Hemochromatosis, *HFE* Variant Analysis, Varies

Test ID: HFET

Useful for:

- Establishing or confirming the clinical diagnosis of hereditary hemochromatosis (HH) in adults
- Testing of individuals with increased transferrin-iron saturation in serum and serum ferritin
- Predictive testing of individuals who have a family history of HH, in coordination with appropriate genetic counseling
- This test is **not recommended for** population screening.

Methods:

Droplet Digital Polymerase Chain Reaction (ddPCR)

Reference Values:

An interpretative report will be provided.

Specimen Requirements:

Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.

Submit only 1 of the following specimens:

Specimen Type:	**Whole blood**
Container/Tube:	
Preferred:	Lavender top (EDTA) or yellow top (ACD)
Acceptable:	Any anticoagulant
Specimen Volume:	2.5 mL
Collection Instructions:	1. Invert several times to mix blood. 2. Send whole blood specimen in original tube. **Do not aliquot.**
Specimen Stability Information:	Ambient (preferred) 4 days/Refrigerated 14 days
Minimum Volume:	0.5 mL

Specimen Type:	**Saliva**
Patient Preparation:	Patient should not eat, drink, smoke, or chew gum 30 minutes prior to

	collection.
Supplies:	Saliva Swab Collection Kit (T786)
Specimen Volume:	1 Swab
Collection Instructions:	Collect and send specimen per kit instructions.
Specimen Stability Information:	Ambient 30 days
Additional Information:	Due to lower concentration of DNA yielded from saliva, it is possible that additional specimen may be required to complete testing.

Specimen Stability Information:

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Cautions:

- This assay only tests for the C282Y, H63D and S65C (reported as a part of the C282Y/S65C genotype) variants and will not detect all variants in the *HFE* gene that may be associated with hereditary hemochromatosis. Therefore, the absence of a detectable C282Y, H63D, or S65C variant 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 the interpretation of results may occur if information given is inaccurate or incomplete.
- Rare variants (ie, 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 variants of unknown significance may be identified.
- Because of concerns of the overall penetrance of *HFE* variants, *HFE* genetic testing is not recommended for population screening.

CPT Code:

81256-*HFE (*hemochromatosis*)* (eg, hereditary hemochromatosis) gene analysis, common variants (C282Y andH63D)

Day(s) Performed: Monday through Friday **Report Available:** 6 to 7 days

Questions

Contact Michelle Raths, Laboratory Technologist Resource Coordinator at 800-533-1710.