Hereditary Hemochromatosis: *HFE* Targeted Analysis

**Test Code:** MHEMO  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81256 x1

### Condition Description

Hereditary hemochromatosis is caused by too much iron being absorbed from the diet. The iron is then stored in various organs and tissues of the body and can cause damage as a person ages. People with this condition can experience tiredness along with joint and abdominal pain. Treatments are available to help reduce the amount of iron in the blood. If untreated, liver and heart disease can develop. However, most individuals with p.H63D and/or p.C282Y will never experience any physical symptoms of disease, but may develop high iron levels in their blood. Individuals with two copies of the p.Cys282Tyr variant are most likely to have significantly elevated blood iron levels, some of whom will go on to develop clinical symptoms. Up to 2% of individuals with one copy of the p.Cys282Tyr variant and one copy of the p.His63Asp variant will show clinical symptoms. Very few individuals with two copies of the p.His63Asp variant will ever show clinical symptoms.

### Genes

*HFE*

### Indications


### Methodology

DNA is analyzed for the p.C282Y and p.H63D variants within the *HFE* gene by multiplex PCR amplification and restriction enzyme digestion.

### Detection


### Specimen Requirements

* Preferred specimen type: Whole Blood

**Type: Whole Blood**

Specimen Requirements:

In EDTA (purple top) or ACD (yellow top) tube:
- Infants (2 years): 3-5 ml
- Older Children & Adults: 5-10 ml.

Specimen Collection and Shipping: Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.