
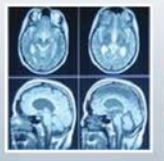


CLINICAL PRACTICE CHANGE

Updated Approach to *HFE*-Related Hereditary Hemochromatosis
June 13, 2016

Date Effective: For specimens received in the lab after June 15, 2016

Background Information:

- The most common form of hereditary hemochromatosis in the Caucasian population is associated with homozygosity or two copies of the p.C282Y variant in the *HFE* gene. This is an autosomal recessive condition with reduced penetrance causing iron overload typically in males between 40 and 60 years of age and females after menopause. Some individuals with iron overload are compound heterozygotes, having one copy of the p.C282Y *HFE* variant and one copy of the p.H63D *HFE* variant.
- Recent data has demonstrated that the p.H63D *HFE* variant has a poor association with *HFE*-related HH. Therefore analysis for this p.H63D *HFE* variant is no longer performed in absence of the p.C282Y *HFE* variant.

Change in or New Test Procedure:

- The p.H63D variant will only be reported for individuals who are identified to carry one copy of the p.C282Y allele. This analysis will not detect other variants in *HFE*.
- There will be four possible results reported.
    - This patient does not have the p.C282Y *HFE* variant.
    - This patient has one copy of the p.C282Y *HFE* variant and does not have the p.H63D variant.
    - This patient has two copies of the p.C282Y *HFE* variant.
    - This patient has one copy of the p.C282Y *HFE* variant and one copy of the p.H63D *HFE* variant.
- Patients will not be genotyped for p.H63D *HFE* variant in the absence of a p.C282Y variant.

Patient Impact:

- Minimal

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