



## Appendix E: Laboratory Test Procedures for *HFE*-HH Testing

This Appendix outlines the testing the laboratory will perform based on the requisition. Refer to [Table 2](#) for the indications for testing and associated instructions for filling out the requisition.

In all cases, the laboratory will collect an EDTA sample for *HFE* C282Y genetic testing and, where applicable, a serum sample for serum iron studies (serum ferritin, serum transferrin, TIBC).

- A. For those presenting with signs and symptoms of hemochromatosis, or with persistently elevated ferritin levels, or with a family history of hemochromatosis (genotype of relative unknown or not C282Y;C282Y), the results of the serum iron studies will determine whether or not genetic testing will be done (see Figure 1 below).
- B. Siblings and children of individuals with confirmed *HFE*-HH due to C282Y homozygosity (i.e., genotype C282Y; C282Y) will not have iron studies performed prior to genetic testing (see Figure 1).

**Note:** Requests for ferritin and/or TSAT do NOT constitute requests for *HFE*-HH testing.

### ► Figure 1: Hemochromatosis Test Procedure

