



Appendix A: HFE-Associated Hemochromatosis Laboratory Testing Algorithm

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

- A. For those presenting with symptoms and signs of hemochromatosis, or with persistently elevated ferritin levels, the results of the serum iron studies will determine whether or not genetic testing will be done (see Figure 2).
- B. First-degree relatives of individuals with confirmed hemochromatosis due to C282Y homozygosity (i.e., genotype C282Y/C282Y) may or may not have iron studies performed prior to genetic testing (see Figure 2).

► **Figure 2: Laboratory Testing Algorithm**

