Hemochromatosis Testing

INDICATIONS FOR TESTING
Adult patient presenting with clinical features suggestive of hereditary hemochromatosis (HH)

ORDER
Serum ferritin
AND
Iron and Iron Binding Capacity
Note: Test includes calculation of serum transferrin saturation

Transferrin saturation <45% and normal serum ferritin
No further testing at this point

Transferrin saturation ≥45% and/or unexplained elevated serum ferritin

ORDER
Hemochromatosis (HFE) 3 Mutations

C282Y homozygosity
HFE HH confirmed
Serum ferritin <1,000 µg/L and normal liver enzymes
Consider referral to hepatologist
Serum ferritin >1,000 µg/L and/or elevated liver enzymes
Refer to hepatologist for fibrosis assessment

Other genotype
Evaluate for iron loading (consider MRI, liver biopsy)
Iron loading absent
No further testing at this point
Iron loading present
Consider rare genotype or digenic inheritance

INDICATIONS FOR TESTING
Adult 1st-degree relative of a patient diagnosed with C282Y homozygous HFE HH or reproductive partner of individual diagnosed with HFE HH

References