Hemochromatosis Testing

INDICATIONS FOR TESTING
Suspicion of hereditary hemochromatosis (HH)
(family history, compatible symptoms)

ORDER
Iron and Iron Binding Capacity
Note: Test includes serum transferrin saturation (STS)
AND
Ferritin (SF)

STSTests and SF tests
AND
Rule out secondary iron overload

STS tests at 2-year intervals

Secondary iron overload

Hemochromatosis (HFE) 3 Mutations gene testing
(accounts for >90% of mutations in Caucasians)

HFE2 gene testing
(accounts for >90% of cases)

HAMP gene testing
(accounts for <10% of cases)

Ferritin ≥1000 μg/L
Aspartate aminotransferase (AST) abnormal
Liver biopsy with hepatic iron concentration

Ferritin <1000 μg/L
Aspartate aminotransferase (AST) normal
Phlebotomy
Monitor STS

CONSIDER
Liver biopsy
Alternative gene testing (TFR2, SLC40A1)

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