

# Hemochromatosis

Diagnosis and evaluation, family screening, liver biopsy indications, phlebotomy therapy, secondary iron overload, and population screening.

STRENGTH 

● Strong (Class 1)

● Weak (Class 2)

## 1 Diagnosis & Evaluation

| STATEMENT                                                                                                                                       | STRENGTH          |
|-------------------------------------------------------------------------------------------------------------------------------------------------|-------------------|
| R1 – Evaluate patients with abnormal iron studies as hemochromatosis, even if asymptomatic                                                      | Strong<br>Level A |
| R2 – Evaluate all patients with evidence of liver disease for hemochromatosis                                                                   | Strong<br>Level B |
| R3 – Suggestive symptoms/history: obtain TS + ferritin (not single test); if either abnormal (TS ≥45% or ferritin >ULN) → HFE mutation analysis | Strong<br>Level B |
| R4 – Target iron-marker testing strategies at high-risk groups (family history, suspected organ involvement)                                    | Strong<br>Level B |

## 2 Family Screening

| STATEMENT                                                                                                                   | STRENGTH          |
|-----------------------------------------------------------------------------------------------------------------------------|-------------------|
| R5 – Screen first-degree relatives (iron studies + HFE mutation analysis) to detect early disease and prevent complications | Strong<br>Level A |

## 3 Liver Biopsy

| STATEMENT                                                                                                                               | STRENGTH          |
|-----------------------------------------------------------------------------------------------------------------------------------------|-------------------|
| R6 – Liver biopsy to stage disease in C282Y homozygotes/compound heterozygotes if ALT/AST elevated or ferritin >1000 µg/L               | Strong<br>Level B |
| R7 – Liver biopsy for diagnosis/prognosis in phenotypic iron overload without C282Y homozygosity/compound heterozygosity                | Weak<br>Level C   |
| R8 – Non-HFE HH: hepatic iron concentration + histopathologic iron staining useful to determine degree/cellular distribution of loading | Weak<br>Level C   |

## 4 Treatment — Phlebotomy

| STATEMENT                                                                                                                                | STRENGTH            |
|------------------------------------------------------------------------------------------------------------------------------------------|---------------------|
| R9 – Weekly therapeutic phlebotomy (as tolerated); target ferritin 50–100 µg/L                                                           | Strong<br>Level A/B |
| R10 – No significant liver disease (normal ALT/AST): C282Y homozygotes with ferritin <1000 µg/L may proceed to phlebotomy without biopsy | Strong<br>Level B   |
| R11 – End-organ damage from iron overload: regular phlebotomy to same endpoints                                                          | Strong<br>Level A   |
| R12 – Dietary adjustment unnecessary; avoid vitamin C and iron supplements during treatment                                              | Weak<br>Level C     |
| R13 – Monitor for iron reaccumulation; maintenance phlebotomy to target ferritin 50–100 µg/L                                             | Strong<br>Level A/B |
| R14 – Non-HFE iron overload with elevated HIC: treat with phlebotomy                                                                     | Strong<br>Level B   |

## 5 Secondary Iron Overload — Chelation

| STATEMENT                                                                                                                                             | STRENGTH          |
|-------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------|
| R15 – Iron chelation (deferoxamine mesylate or deferasirox) for iron-overloaded patients with dyserythropoietic syndromes or chronic hemolytic anemia | Strong<br>Level A |

## 6 Population Screening

| STATEMENT                                                         | STRENGTH          |
|-------------------------------------------------------------------|-------------------|
| R16 – Average-risk population screening for HH is NOT recommended | Strong<br>Level B |