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Summary

EASL Clinical Practice Guidelines on haemochromatosis

Standards and Guidelines

Hepatobiliary

March 29, 2022

Haemochromatosis diagnosis and management depend on HFE genotype, with serum iron parameters sufficient for p.Cys282Tyr homozygotes but imaging or biopsy required for other genotypes.

KEY POINTS

- In p.Cys282Tyr homozygotes, haemochromatosis can be diagnosed using serum iron parameters alone: TSAT greater than 45% and ferritin greater than 200 µg/L in females, or TSAT greater than 50% and ferritin greater than 300 µg/L in males and postmenopausal women.
- Patients with high TSAT and elevated ferritin but other HFE genotypes require hepatic iron overload confirmed by MRI or liver biopsy for diagnosis.
- Early treatment by phlebotomy can prevent cirrhosis, hepatocellular carcinoma, diabetes, arthropathy and other complications.
- Assessment of liver fibrosis stage and end-organ damage at diagnosis determines disease management, with advanced fibrosis requiring hepatocellular carcinoma screening.
- This material is relevant to clinicians diagnosing and managing patients with suspected or confirmed haemochromatosis.

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