

Evidence Summary - Hereditary (Primary) Hemochromatosis (HH) and Other Iron Storage Diseases (AB 11.2)

The ACG Clinical Guideline indicates that MRI without contrast is the preferred modality for assessing hepatic iron concentration in iron overload conditions, including primary hereditary hemochromatosis (HH) as well as in secondary, multi-transfusion conditions, such as sickle cell disease, thalassemia, and in oncology patients and those with bone marrow failure, in whom it can be done annually. MRI offers several key advantages. MRI can distinguish between primary and secondary iron overload based on uptake in the reticuloendothelial system, is non-invasive, radiation-free, and has the ability to be performed on both liver and heart. In addition, it is useful for screening, as noted, in the appropriate populations.

CT has been used but presents the negatives of radiation exposure. Dual-energy scans are required to compensate for background attenuation, so its use is reserved for patients without access to MRI.¹⁹

Ultrasound-based elastography can assess the need for biopsy. However, Magnetic Resonance Elastography (MRE) is not preferred due to MRI signal degradation by excess iron and is not recommended by the ACG at any stage of the work-up.

For individuals with iron indices indicative of classic HH, iron mobilized by well-controlled phlebotomy can provide an alternative estimate of total body iron comparable to liver iron quantification.²¹ Serial MRI monitoring of hepatic iron concentration has not been defined; instead, serum ferritin levels are monitored during phlebotomy.

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