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Hereditary Hemochromatosis: Defining the Disease

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Defining the Disease

Hereditary hemochromatosis has been traditionally defined as an inherited disorder characterized by inappropriately high absorption of dietary iron, which leads to abnormal accumulation of iron in parenchymal organs.^[2,3] In 1996, the HFE gene was isolated on chromosome 6. Since this finding hereditary hemochromatosis has been defined by some as the presence of a homozygous defect in the HFE gene with direct or indirect evidence of iron overload.^[8] The requirement for evidence of iron overload to establish the diagnosis is a point of controversy in the new era of genetic testing.^[3,6,9-12] For this article, hereditary hemochromatosis will be defined as the phenotypic expression of iron overload in the presence of the HFE mutations known to cause the disease.

Iron overload is not synonymous with hereditary hemochromatosis. Although hemochromatosis is the most common cause of primary iron overload, there are other inheritable causes of primary iron overload, some of which occur only in the setting of excessive iron intake. [Table 1](#) displays the differential diagnosis of iron overload.

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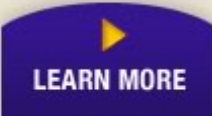
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