JL06. Hereditary Hemochromatosis: What the Nurse Practitioner Needs to Know

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Background: Hereditary hemochromatosis is a disorder in which excessive iron absorption occurs, leading to an increase in iron deposits in major organs of the body and resulting in end organ damage. This is the most common genetic disorder in individuals of Northern European ancestry, and the history of this disease can be traced back to the time of the Vikings and Celtics. Although it was known for many years that this was a hereditary disease presenting with the classic triad of diabetes, bronze pigmentation of the skin, and cirrhosis of the liver, it was not until 1996 that the *HFE* gene was identified. Hereditary hemochromatosis is an autosomal recessive disorder where a mutation occurs at on chromosome 6 of the short arm of *HFE* resulting in two changes: the substitution of tyrosine for cysteina at position 282 (C282Y) and the substitution of aspartic acid for histidine at position 63 (H63D) of HFE protein. Hemochromatosis is easily treated if identified early, before it manifests itself as a disease. The treatment is therapeutic phlebotomy. *Discussion:* Individuals who have this mutation are asymptomatic until their ferritin levels are markedly elevated, at which point penetrance of the disease may present in affected organs. The disease may manifest itself as diabetes, arrhythmia, liver disease, arthritis, and sexual dysfunction. It is most important that primary care nurse practitioners have a familiarity with the hereditary hemochromatosis and how to screen, diagnose, and treat patients with the disease.