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Understanding Hereditary Hemochromatosis

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Introduction

One of the most common genetic diseases, hereditary hemochromatosis is a disruption of iron regulation in the body. Iron, the most abundant element in the earth's crust, is distributed worldwide, but it is most common in those of northern European origin. (Reich and DL Palma, 2012). Occurrence is rare in other racial or ethnic groups. (Emanuele, Tsai, and Emanuele, 2014)

Symptoms

Signs and Symptoms

- Slow progression. Many are asymptomatic.

- Age of presentation typically 45-60 in men, postmenopausal in women.

- Early signs and symptoms to be noticed and may include:
  - Tiredness: low weight loss
  - Fatigue
  - Abdominal discomfort
  - Weakness
  - Arthritis, particularly of the second or third metacarpophalangeal joints.
  - Hair loss
  - Gastrointestinal symptoms
  - Changes in mental status/depression
  - Loss of libido

- With untreated advanced disease, patients may present with:
  - Hepatitis
  - Hypothyroidism
  - Hepatocellular cancer
  - Cardiomyopathy
  - Arrhythmias
  - Arthritis, particularly of the second or third metacarpophalangeal joints.
  - Gastrointestinal disorders
  - Diabetes Mellitus
  - Hepatocellular carcinoma

- Laboratory findings:
  - Transferrin Saturation >45%
  - Serum ferritin >200 in premenopausal females

- Other forms of iron overload, normally as a result of HFE gene mutation, (Centers for Disease Control and Prevention, 2010)

Underlying Pathophysiology

Hereditary hemochromatosis encompasses a range of iron overload disorders which are due to genetic miscalibration of iron acquisition. (Vujic, 2014). The specific mutation which accounts for hereditary hemochromatosis occurs at the major histocompatibility complex (MHC) region on the short arm of chromosome 6. A missense mutation may be found at this locus in 80% of hereditary hemochromatosis patients. The mutation results in a cysteine to tyrosine substitution at amino acid 282 of the HFE protein (C282Y). (Vujic, 2014). This form of the disease is known as “hemochromatosis type 1”. (Chejhaj, Hollenbeck, Go, & Brown, 2013)

Iron is seen as essential for cell metabolism and is a constituent of hemoproteins, such as hemoglobin and cytochrome. (Crownover and Covey, 2013). There is debate among researchers about the number of those homozygous for the disease, this is a significant portion of the population.

Prognosis is worsened in those with diabetes, cirrhosis, or cardiomyopathy. (Emanuele, et al., 2014). Patients with hereditary hemochromatosis manifest in hereditary hemochromatosis in patients with non-specific symptoms such as fatigue, arthralgia, weight loss, or changes, or depression in conjunction with elevated transferrin saturation and ferritin levels. Medical professionals should be aware that many patients are diagnosed with hereditary hemochromatosis after the disease is in an advanced stage. Symptoms such as fatigue, joint pain, and other symptoms of iron overload such as cardiac disease may present in the absence of iron overload. (Crownover and Covey, 2013, p. 103)

Hereditary hemochromatosis can lead to irreversable organ damage.

Implications for Nursing Care

Although patients may rarely present with the classic triad of symptoms described by Trautwein (bremed skin, diabetes mellitus, and hepatosplenomegaly), patients should consider hereditary hemochromatosis in patients with non-specific symptoms such as fatigue, arthralgia, weight loss, or changes, or depression in conjunction with elevated transferrin saturation and ferritin levels. Medical professionals should be aware that many patients are diagnosed with hereditary hemochromatosis after the disease is in an advanced stage. Symptoms such as fatigue, joint pain, and other symptoms of iron overload such as cardiac disease may present in the absence of iron overload. (Crownover and Covey, 2013, p. 103)

Treatment for iron overload and iron loading is a challenge for patients with a disease that may require years of treatment. Hereditary hemochromatosis is the most common genetic disorder affecting whites in the United States, however, only 5-10% of those who are homozygous for the mutation are asymptomatic. Many patients continue to present with complaints, but may progress to sign organ damage and early death.

Conclusion

Hereditary hemochromatosis is the most common genetic disorder affecting whites in the United States; however, only 5-10% of those who are homozygous for the mutation are asymptomatic. Many patients continue to present with complaints, but may progress to sign organ damage and early death.

References


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Figure 1. The hand on the right shows hyperepigmentation, or bronzing of the skin, due to iron overload in hereditary hemochromatosis.

Image Courtesy of Consultant360.com