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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. Its geographic distribution is worldwide, but it is most common in those of northern European origin. (Riach and Di Palma, 2012). Occurrence is rare in other racial or ethnic groups. (Emanuele, Tranzer, and Edelstein, 2014). Symptoms are due to significant iron overload, normally as a result of HFE gene mutation. (Centers for Disease Control and Prevention, 2010). The HFE gene plays an important role in regulating iron absorption in the G tract, transport, and storage. (Emanuele, et al., 2014). If excess iron accumulates in vital organs cirrhosis, bone and joint disease, diabetes, endocrinologic disorders and skin problems can result. (Ceveri and Coyne, 2013). Although genetically similar problems of iron overload, their phenotypic expression or, clinical symptoms varies in those with the genetic mutation, those who have the HFE mutation are much more genetically susceptible to iron overload as it manifests in hereditary hemochromatosis. Patients can present with nonspecific complaints such as weakness, fatigue, changes in mental status, and arthritis, so hereditary hemoschromatosis can be missed or misdiagnosed if iron testing is not performed. The most common route to diagnosis is through phlebotomy office visits or identification of common complaints. (Brown, Ball, Kline, and Cannon, 1996). In some cases, but not all, genetic testing may be appropriate. Other genetic testing must be considered as well when making a differential diagnosis.
Hereditary hemochromatosis is a common autosomal recessive trait. A carrier father and a carrier mother have a 1 in 4 chance of having a heterozygous (carrier) child. (Centers for Disease Control and Prevention, 2010). About 75% of homozygotes have expression of the disease, while those with the genetic mutation, those who are genetically susceptible to iron overload as it manifests in hereditary hemochromatosis.

Signs and Symptoms
- Slow progression. Many are asymptomatic.

- Age of presentation typically 40-60 in men, postmenopausal in women.

- Early signs and symptoms may be carriers of the affected HFE gene. (Rofail, et al., 2013). According to Centers for Disease Control and Prevention, 2011). The HFE gene plays an important role in regulating iron absorption in the G tract, transport, and storage. (Emanuele, et al., 2014). If excess iron accumulates in vital organs cirrhosis, bone and joint disease, diabetes, endocrinologic disorders and skin problems can result. (Ceveri and Coyne, 2013). Although genetically similar problems of iron overload, their phenotypic expression or, clinical symptoms varies in those with the genetic mutation, those who have the HFE mutation are much more genetically susceptible to iron overload as it manifests in hereditary hemochromatosis.

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Underlying Pathophysiology
Hereditary hemochromatosis encompasses a range of iron overload disorders which are due to genetic or metabolic iron accumulation. (Ouyj, 2014). Although, the mechanisms which account for iron overload in hereditary hemochromatosis occur at the major histocompatibility complex (MHC) region on the short arm of chromosome 6. A missense mutation may be found in 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 (CZ282Y). (Ouyj, 2014). This form of the disease is known as “hemochromatosis type 1”. (Cherwine, Hollenberg, Gu, & Brown, 2013).

Implications for Nursing Care
Although patients may rarely present with the classic triad of symptoms described by Trousseau (bromed, diabetes mellitus, and hypogonadism), healthcare providers should consider hereditary hemochromatosis in patients with nonspecific symptoms such as fatigue, arthralgia, weakness, skin changes, or depression in conjunction with elevated serum transferrin saturation and ferritin. Medical professionals should be aware that expression of the disease is influenced by factors such as race, gender, alcohol consumption, and iron rich diet. Identifying patients at risk for hereditary hemochromatosis and cirrhosis is reduced by 50 percent with those who do not have genetic testing. (Kari Davidson, RN, BSN, Cherfane, et al., 2014).

Conclusion
Hereditary hemochromatosis is the most common genetic disorder affecting whites in the United States. Ninety-five percent of those who are homozygous for the mutation are asymptomatic. Manifestations of hereditary hemochromatosis correlate with body iron content, with symptoms worsening as iron levels increase. Early recognition of disease progression, patients may experience a survival rate in those who have untreated hereditary hemochromatosis.

References
http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3949517/