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Understanding Hereditary Hemochromatosis

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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 occurrence and distribution is worldwide, but it is most common in those of northern European origin. (Rivera and D’Palma, 2012). Occurrence is rare in other racial or ethnic groups. (Emanuele, Trahey, Galicich, & Edwards, 2014).

Signs and Symptoms

- **Low progression.** Many are asymptomatic.
- **Age of presentation typically 45-60 in men, post-menopausal women in women.**
- **Early signs and symptoms tend to be general and subtle.**

Underlying Pathophysiology

Hereditary hemochromatosis encompasses a range of iron overload disorders which are due to genetic miscalculation of iron accumulation. (Vujic, 2014). This genetic miscalculation which accounts for disease in 85-90% of cases of hemochromatosis occurs at the major histocompatibility complex (MHC) region on the short arm of chromosome 6. A missense mutation 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”. (Chehane, Hollenberg, Go, & Brown, 2013).

Slight weight loss
- Fatigue
- Gastrointestinal discomfort
- Abdominal discomfort
- Weakness
- Extensive leg syndromes
- Hair loss
- Loss of libido
- Changes in mental status/depression

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Implications for Nursing Care

Although patients may rarely present with the classic triad of symptoms described by Tyrer (liver, skin, diabetes mellitus, and hypogonadism), many people should consider hereditary hemochromatosis in patients with non-specific symptoms such as fatigue, arthralgia, weakness, altered moods, changes, or depression in conjunction with elevated transferrin saturation and ferritin levels. Medical professionals should be aware that expression of the disease is influenced not only by genetics but also by other factors such as alcoholism, iron rich diet, menopause in women, and diseases of other iron overload syndrome such as celiac disease. (Chehane, Hollenberg, Go, & Brown, 2013).

If iron begins to enter the plasma at a high rate, leading to increased transferrin saturation and ferritin levels, and over time builds up in body tissues and organs, causing the above signs and symptoms. (Emanuele et al., 2015). Most of the iron overload usually affects the liver, pancreas, heart, pituitary gland, testicles, and joints. Many of the presenting signs and symptoms are due to disruption of the normal function of these organs.

Manifestation of symptoms in the liver are fibrosis and cirrhosis. Iron overload may contribute to the risk of diabetes, as it is associated with increased hepatic glucose production and metabolic inflexibility, both characteristics of type 2 diabetes. (Huang, et al., 2013). About 75% of hemochromatosis patients have diabetes mellitus some time during their disease, while others may develop diabetes mellitus in the absence of iron overload. (Covey, 2013). About 50% of these patients will develop diabetes mellitus in the absence of iron overload. (Covey, 2013).

Conclusion

Hereditary hemochromatosis can impose significant disease affecting quality of life. As the number of persons who are homozygous for the disease rise, this will cause a significant increase in the number of patients with hereditary hemochromatosis who are asymptomatic. (Covey, 2013).

References


