

Hemochromatosis - How Common is It?

The Most Common Genetic Disorder in the Western World

Hemochromatosis is the **most common genetic disorder in the western world**, affecting an estimated 1 in 300 Canadians of Northern European descent. That means about 100,000 Canadians have Type I hemochromatosis, the most common form of hereditary hemochromatosis (HHC).

Hereditary hemochromatosis is an autosomal recessive disorder, meaning that a person needs to inherit two defective copies of the gene, one from each parent, in order to be affected. **Men and women are affected equally**. People with just one copy of the mutated gene and one normal copy are referred to as carriers, or heterozygotes. While carriers only rarely develop hemochromatosis, children of two carriers may inherit the defective genes from both and develop the disorder. Current research estimates that **1 in 9 Canadians are carriers**, making the above scenario a very real possibility.

Genetics

The **most common gene related to iron metabolism is called HFE** (Human + Fe, the symbol for iron). In 1996, genetic researchers, searching for genes responsible for hemochromatosis, identified HFE as essential to iron metabolism and that mutations of this gene were responsible for Type I Hemochromatosis. The two most common mutations of this gene, named C282Y and H63D, account for 85% of all cases of hemochromatosis.

Although the vast majority of hemochromatosis patients have a combination of the 2 common mutations (C282Y and H63D) in the HFE gene, other genes associated with iron metabolism have been discovered. Mutations in these other genes have been linked to more rare types of hereditary hemochromatosis, including Juvenile Hemochromatosis, Hemochromatosis Type III and Type IV that account for iron overload in other race and age groups. In addition, more mutations of the original (HFE) gene have been identified, making the genetics of hemochromatosis a complex topic.

Complexity

Research into the various genes involved in the metabolism of iron has led to a better understanding of other disorders that may involve malfunctions of iron absorption, such as Alzheimer's and Parkinson's disease. The complexity of iron metabolism and the interrelation of several genes, some known, some yet to be discovered, may account for the fact that **some carriers develop hemochromatosis, and some people with two defective genes do not**. It also may explain why such **a wide range of severity and expression of disease exists** among individuals with hemochromatosis.

NOTE: Mutations of the HFE gene are found to cause hemochromatosis primarily in people of Northern European descent. These mutations are not typically found in African, Asian or Middle Eastern populations. Although uncommon, iron overload conditions do exist in these populations but are undoubtedly caused by other genes or environmental factors.

Other Iron Overload Conditions

For more information on other iron overload conditions, visit the Iron Disorders Institute web site at www.irondisorders.org.