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Hemochromatosis

Hereditary hemochromatosis is the genetic form of hemochromatosis, a disease characterized by a progressive iron overload.

deCODEme can calculate your genetic risk for Hemochromatosis.

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Hereditary hemochromatosis is caused by known genetic mutations. Undiagnosed and untreated, it can have serious consequences over time.

Hereditary hemochromatosis is one of the few genetic disorders for which there is a relatively simple and effective therapy; iron levels are lowered by removing blood as directed by a physician.

This disorder has a very interesting geographic

In hereditary hemochromatosis (HH) more iron is absorbed than is needed. With the exception of menstruation in women, the human body has no natural means of getting rid of extra iron. As a result, iron builds up in individuals with HH and is deposited throughout the body. Over time iron can reach toxic levels in tissues, causing dysfunction and failure in major organs such as the liver, pancreas, heart, thyroid, pituitary gland, and joints.

Hereditary hemochromatosis can have a variety of symptoms, ranging from mild conditions like fatigue, weakness, abdominal and joint pain to more severe ones associated with failures in the aforementioned organs. Undiagnosed and untreated, HH can have serious consequences over time, including increased risk for liver diseases, heart problems, arthritis, depression, impotence, infertility, hypothyroidism, pituitary hormone deficiency, diabetes and even some forms of cancer.

The worldwide prevalence of HH in 18-70 year old individuals is estimated to range from 1.5 to 3 per 1000 individuals, it affects about one million individuals in the US. However, the disease is thought to

distribution that is thought to reflect the historical movements of people of northwestern Europe.

be significantly under-diagnosed and is more common in men than in women. The prevalence of HH also varies considerably between populations, being most common in individuals of European descent.

Hereditary hemochromatosis actually has a geographic distribution that is thought to reflect the historical movements of people of northwestern Europe. It is most frequent in populations surrounding the North Sea; Norway, Denmark, Iceland, Germany (west and south), France, the United Kingdom, and Ireland. It is less frequent in southern Europe, and almost non-existant in Africa. In the United States, the frequency in individuals of European descent is similar to that seen in northern Europe. The highest frequency of the disease is found in the Irish population. This and the geographic distribution of the disease, has led scientists to believe that hemochromatosis is the result of a relatively recent mutation originating in a Celtic population (indeed, HH is sometimes referred to as the “Celtic curse”). Some scientists have however proposed that the mutation is of Viking or Germanic origin.

Hereditary hemochromatosis is caused by mutations in the HFE gene on chromosome 6. There are more than 20 known mutations in the HFE gene, but the most important for HH identified to date are two sequence variants, C282Y and H63D. The C282Y mutation explains 80 to 90 percent of all diagnosed cases of HH in populations of northwestern European ancestry.

The deCODEme Complete Scan identifies the C282Y and H63D sequence variants in the HFE gene on chromosome 6 and gives an interpretation of the associated genetic risk for hereditary hemochromatosis.

risk factors

The main risk factors associated with hereditary hemochromatosis are:

- Genetics: Hereditary hemochromatosis shows an autosomal recessive pattern of inheritance. This means that an individual with the disease must have inherited a mutated and non-functional copy of the HFE gene from both parents. Such an individual is said to be homozygous for mutated copies of the HFE gene.

  In populations of northern European descent, one in 200-250 are homozygous (have two mutated copies) for C282Y. One in 50 are compound heterozygotes (have one C282Y mutation and one H63D mutation). One in 8-10 are heterozygotes for C282Y (carriers of the mutation).

  Although two copies of the mutated HFE gene are required to have the disease, not everyone who has two copies actually gets the disorder, meaning in genetic terms, that the genetic variants do not have full penetrance . Most studies report that 60-95% of C282Y homozygotes show symptoms of the disease.

- Ancestry: Hereditary hemochromatosis is found almost exclusively in individuals of northern European ancestry.
- Age: Older people are more likely to develop the disease than younger people. Symptoms do not usually appear in men until after the age of 40. In women, symptoms usually do not appear until after the age of 50 (i.e. after menopause). Individuals carrying two mutated and non-functional copies of the HFE gene rarely develop HH as young children.
- Other factors: The severity of the disorder varies between individuals homozygous for mutated HFE gene copies. Some people may never have symptoms or complications of the disorder while others can be severely affected. Certain factors, including both lifestyle and genetic factors, seem to affect the symptoms and progress of the disorder for those who have inherited mutated copies of the gene:
    - Other genes, besides the hemochromatosis gene, may modify the severity of the disease,
    - Vitamin C in the diet can increase the amount of iron the body absorbs from food and make hemochromatosis worse,
    - Alcohol use can increase the risk of liver damage and cirrhosis,
    - Certain other conditions, such as hepatitis (inflammation of the liver), can increase the effects of iron overload on damage to the liver.

prevention

The key to preventing hemochromatosis is early diagnosis and treatment. Diagnosis of HH is typically based on blood tests that measure transferrin, iron saturation and serum ferritin concentration, but also on molecular genetic testing for the C282Y and H63D mutations in the HFE gene.

Due to incomplete penetrance, the genotype provided by the deCODEme Genetic Scan is in itself insufficient for the diagnosis of HH, but should be considered as evidence of susceptibility to developing the disease. Individuals identified as C282Y homozygotes or C282Y/H63D compound heterozygotes should undergo further testing to identify or exclude iron overload.

As previously described, not everyone who inherits the hemochromatosis mutations develops the disease. For those who have confirmed iron overload, physicians may recommend preventive measures such as dietary changes aimed at reducing iron absorption, for example avoiding taking in extra iron (for example in multivitamins), limiting intake of vitamin C (as it increases iron absorption) and limiting alcohol intake to reduce risk of liver disease.

In terms of treatment, HH is actually one of the few genetic disorders for which there is a relatively simple and effective therapy; iron levels are lowered simply by removing blood (a procedure called phlebotomy).

When phlebotomy is started early in the course of the illness, it can prevent most of the complications associated with the disease. However, even if phlebotomy is started after complications have occurred, the treatment can still decrease symptoms and improve life expectancy.

more information

You can find out more information about hemochromatosis by talking to your doctor and visiting these Web sites:

- Centers for Disease Control and Prevention on Iron Overload and Hemochromatosis
- Iron Disorders Institute
- National Heart, Blood, and Lung Institute (NHBLI) article on Hereditary Hemochromatosis
- National Human Genome Research Institute on Hereditary Hemochromatosis
- MedlinePlus article on Hereditary Hemochromatosis

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