



The Four Types of Hemochromatosis

Blog Categories:
HH Information
Science

Did you know that there are four 'types' of Hemochromatosis - called types 1, 2, 3 and 4?

Background



Hereditary Hemochromatosis (HH) is the most common genetic disease so far identified, with around 1 in 200 people severely affected. Most people with Hemochromatosis have mutations in the HFE gene, discovered in 1996.

Classifications

Hereditary Hemochromatosis is classified by type depending on the age of onset and other factors, such as genetic cause and mode of inheritance.

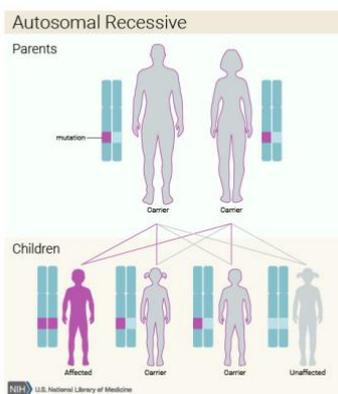
Type 1 Hemochromatosis is the most common form of the disorder, caused by a mutation in the HFE gene and begins in adulthood. Men with type 1 Hemochromatosis typically develop symptoms between the ages of 40 and 60, and women usually develop symptoms after menopause.

Sometimes referred to as 'The Celtic Curse' it is most prominent in people of Northern European/ Celtic ancestry.

Type 2 Hemochromatosis is a juvenile-onset disorder, caused by mutations in the HAMP or HJV genes. Iron accumulation begins early in life, and symptoms may appear in childhood. By age 20, decreased or absent secretion of sex hormones is evident. Females usually begin menstruation in a normal manner, but menses stop after a few years. Males may experience delayed puberty or symptoms related to a shortage of sex hormones. If the disorder is untreated, heart disease becomes evident by age 30.

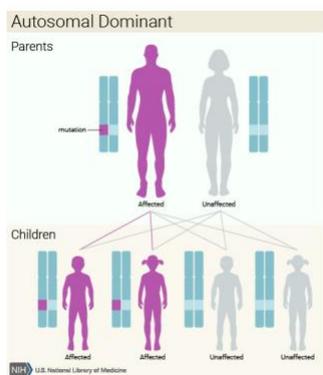
Type 3 Hemochromatosis is characterised by intermediate onset, between types 1 and 2 and is caused by mutation in the TFR2 gene. Symptoms of type 3 Hemochromatosis generally begin before age 30.

Type 4 Hemochromatosis - Ferroportin disease - is caused by a mutation in the SLC40A1 gene and begins in adulthood. Men with type 4 Hemochromatosis typically develop symptoms between the ages of 40 and 60, and women usually develop symptoms after menopause.



Modes of Inheritance

Types 1 to 3 Hemochromatosis are inherited in a recessive pattern, which means both copies of the gene in each cell have mutations. Most often, the parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene but do not show signs and symptoms of the condition.



Type 4 Hemochromatosis is distinguished by its dominant inheritance pattern. With this type of inheritance, one copy of the altered gene in each cell is sufficient to cause the disorder. In most cases, an affected person has one parent with the condition.

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