

## Haemochromatosis

Haemochromatosis (iron overload disorder) is one of the most common hereditary diseases. Around 1 in 300 people have a genetic predisposition to this disease - that is they may get it.

Haemochromatosis is characterised by the excessive absorption of iron. Normally, excess iron is safely stored in various joints and organs in the body, particularly the liver. In a person with haemochromatosis, iron stores keep rising and, over time, the liver enlarges and becomes damaged, leading to serious diseases such as cirrhosis. Other problems that can be caused by excessive iron include heart disease, diabetes and arthritis. Both sexes are at risk, but women tend to develop the condition later in life, since regular menstrual periods deplete the body of iron. Haemochromatosis tends to be under-diagnosed, partly because its symptoms are similar to those caused by a range of other illnesses.

### Iron is a vital trace mineral

Red blood cells contain a protein called haemoglobin, which carries oxygen. Iron is needed for production of this particular protein, and the iron in food is absorbed via the small intestine. The human body has no method of excreting excess iron, so any excess is normally stored in the liver, with no ill effects. The body typically stores around one gram or less of iron at any given time. However, a person with haemochromatosis absorbs a great deal more iron from their food than is necessary. Iron stores of five grams or more build up inside the body. Organs such as the liver, heart and pancreas are affected and ultimately damaged. Without treatment, haemochromatosis can cause premature death.

### Symptoms of haemochromatosis

Early haemochromatosis has no symptoms. However, in its later stages, haemochromatosis presents a variety of symptoms, and not all people will experience the same signs. Many symptoms are similar to those caused by other illnesses, which partly explains why haemochromatosis may be overlooked as a possible diagnosis. Some of the symptoms include:

- Weakness and lethargy
- Weight loss
- Joint pain, usually in the joints of the second and middle fingers
- Liver complaints, such as cirrhosis
- Sexual dysfunctions, such as impotence and low sex drive
- Disorders of the menstrual period, such as early menopause
- Loss of body hair
- Skin darkening
- Cognitive difficulties, such as poor memory.

### A recessive gene

Haemochromatosis is a recessive gene disorder caused by mutations of the haemochromatosis (HFE) gene. To develop a recessive gene disorder a person needs to inherit the gene mutation from both parents. If a person inherits only one mutated HFE gene, they are known as carriers. Around one in seven people carry the mutated HFE gene. A carrier won't develop the condition themselves, but may pass the mutation on to their children. If two carriers conceive, their child has a 50 per cent chance of inheriting one mutated HFE gene and becoming a carrier, and a one in four chance of inheriting both mutated HFE genes and developing the disease. A simple test can establish whether a person is carrying the gene.

### Treatment for haemochromatosis

A person with haemochromatosis is treated with venesection. This is a procedure similar to blood donation, where around 500mls of blood is removed until iron in the blood is reduced to normal levels. Depending on the severity of the condition, this may take around one and a half years of twice-weekly visits. Once iron levels are normal, venesection needs to be performed three or four times every year for life. If haemochromatosis is treated in its earliest stages before severe organ damage has occurred, there is no reduction in life expectancy - other things being equal.

## Lifestyle changes

A person with haemochromatosis can better manage their condition by making a few simple lifestyle changes, including:

- Reducing or limiting iron-rich foods such as red meat
- Not taking iron supplements
- Not taking vitamin C supplements, as vitamin C increases iron absorption.
- Reducing alcohol intake, as metabolising alcohol can stress an already compromised liver.

## Preventing organ damage

If a person is diagnosed before significant symptoms arise, they can prevent organ damage and disease symptoms by maintaining iron in the normal range. However, a person diagnosed with the condition should notify all blood relatives so they can be tested for the HFE genes and treated if necessary. Anyone with disorders including liver disease, cardiomyopathy, arthritis or impotence should be tested for haemochromatosis. This underlying condition (haemochromatosis) could be causing their secondary illnesses. Prompt treatment can reverse some organ damage and symptoms and prevent further damage.

## Where to get help

- Your doctor
- Haemochromatosis Australia information line Tel. 1300 019 028

## Things to remember

- Haemochromatosis is a common inherited disorder, which causes the body to absorb more iron than usual from food.
- Haemochromatosis tends to be under-diagnosed, partly because its symptoms are similar to those caused by a range of other illnesses.
- Treatment includes regularly removing blood until iron levels normalise.

**This page has been produced in consultation with, and approved by:**

Haemochromatosis Australia

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