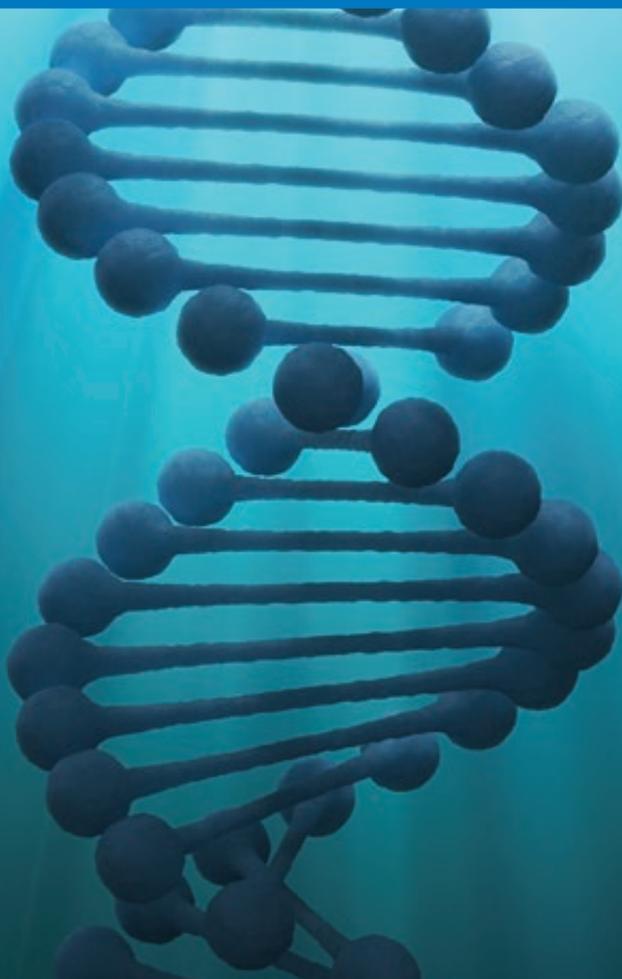


Hereditary haemochromatosis

Information for patients



This leaflet has been written for people who have hereditary haemochromatosis (HH) or have a family history of HH.

What is haemochromatosis?

Haemochromatosis is a disorder that causes the body to absorb too much iron from our normal diet. If no treatment is given, iron can build up in various organs in the body and cause problems with how the organs work.

Causes

There are many different causes of haemochromatosis. We know that some people will have inherited gene alterations (mutations) that increase the chances of developing haemochromatosis. This is called hereditary haemochromatosis (HH). Inheriting the gene alterations is not enough to cause HH, but you are more likely to develop it at some stage. With early diagnosis and treatment, symptoms can be prevented and you can expect to lead a normal life.

Symptoms

The symptoms of HH can all be explained by too much iron in the body. The iron levels build up very slowly over many years, so people do not usually notice any symptoms of HH until they are an adult.

Some of the symptoms that can result from too much iron storage are:

- arthritis
- chronic (long term) tiredness and lethargy
- abdominal pain
- changes in skin colour (typically a darkening, or sunburnt look)
- abnormal liver function or a condition called liver cirrhosis
- diabetes
- heart rhythm problems or enlargement
- shortness of breath
- male impotence.

If these signs are picked up early then they can be treated very successfully. It's very important to realise that not everyone who has a genetic tendency to HH will develop symptoms or need any treatment. Women are generally less affected than men because they lose blood through menstruation (periods) every month.

Treatment

The best treatment is regular removal of blood (venesection), which is like giving blood. Removing blood will lower the levels of iron in the body. Your doctor will need to check your iron levels regularly. This is a very effective treatment and you can expect to lead a normal life if the condition is managed correctly.

How are these genes inherited?

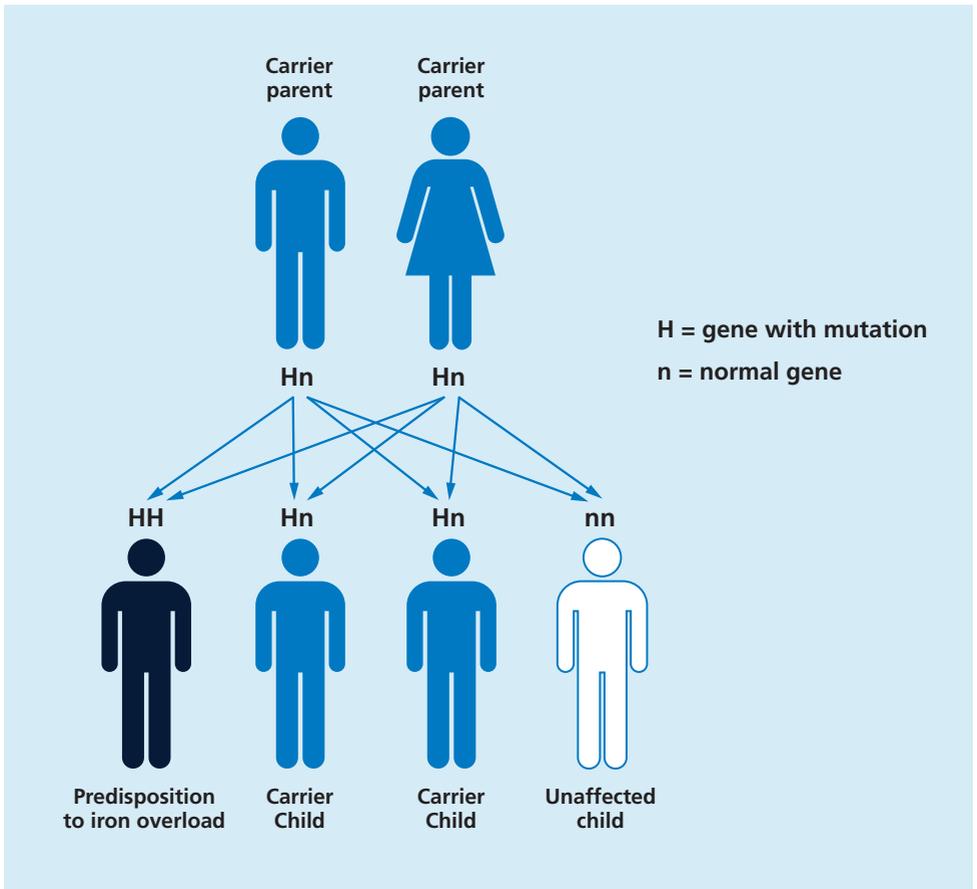
The inherited predisposition to haemochromatosis is caused by gene mutations. Almost every cell in our body contains about 25,000 genes which act like a set of instructions to the cell, controlling growth and how our bodies work. All our genes come in pairs, made up of one gene inherited from our mother and one from our father. When we have children, we pass on just one gene from each pair.

The particular gene that is associated with HH is the HFE gene. We all have two copies of this gene, and when we have a mutation in both copies, the chance of developing haemochromatosis increases.

For someone to inherit a tendency towards HH, they must inherit a mutation in both copies of the HFE gene (one from each parent). Having one normal copy and one mutation does not seem to cause any problems and these people are known as carriers of HH. This is quite common, with approximately one in eight of us being a carrier of HH.

If both parents are carriers of HH, then each child has a:

- one in four (25%) chance of inheriting two mutations. People who inherit two mutations will be more likely to develop iron overload, and therefore they need regular checks of their iron levels. However, not everyone who inherits two mutations will develop iron overload.
- one in two (50%) chance of inheriting one mutation and one normal gene from their parents. In this case they will be healthy carriers like their parents.
- one in four (25%) chance of inheriting two normal copies of the HFE gene. In this case the child will not have an inherited tendency towards HH and will not be a carrier.



These possible outcomes occur randomly. The chance remains the same in every pregnancy and is the same for boys and girls.

If only one parent is a carrier, then each child will have a one in two (50%) chance of being a healthy carrier but will not be at risk of HH.

Blood tests

Blood iron levels are measured by two tests:

- transferrin saturation, and
- serum ferritin.

Genetic testing may be suggested if you have higher than average blood iron levels, or if you have a family history of HH. A genetic test can be done on a small sample of your blood.

Genetic testing looks for the two most common mutations in the HFE gene, known as H63D and C282Y. This standard test picks up the vast majority of people with a predisposition to haemochromatosis, but not all. Occasionally, another test may be suggested.

Diet

As HH is a condition of iron overload, it's logical to consider that cutting down on iron in the diet would be an effective treatment. However, it's not possible to treat HH with a low iron diet. Only a small amount of iron is absorbed from a normal diet, and if you try to keep your iron intake very low you may not get enough nutrition.

You should aim to have as healthy and balanced a diet as possible. We also suggest you:

- avoid vitamin supplements or tonics containing iron
- avoid breakfast cereals heavily fortified with iron
- limit your intake of red meat
- avoid large doses of vitamin C, as it increases how much iron is absorbed from your diet
- limit your alcohol intake, as this can increase the risk of liver disease.

Further information

If you need any advice about hereditary haemochromatosis, please contact:

- **Wessex Clinical Genetics Service**

Princess Anne Hospital
Coxford Road
Southampton
SO16 5YA

Telephone: **023 8120 6170**

Website: **www.uhs.nhs.uk/genetics**

Useful contacts

- The Haemochromatosis Society

Website: **www.haemochromatosis.org.uk**

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**If you need a translation of this document,
an interpreter or a version in large print,
Braille or on audiotape, please telephone
023 8120 4688 for help.**

www.uhs.nhs.uk/genetics

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