Haemochromatosis

Haemochromatosis is an inherited (genetic) disorder causing the body to absorb too much iron from the diet. The excess iron causes damage to an organ in which it collects. The main treatment is the regular removal of blood, which helps to remove the excess iron from the body. If treatment is started early, before complications occur, then the outlook for people with haemochromatosis is very good.

What is haemochromatosis?

If you have haemochromatosis, you have inherited a faulty gene, which directs your system to absorb too much iron from what you eat and drink. Excessive quantities of iron are then stored in various organs. This excess iron may be stored in the liver, pancreas, heart, testicles (testes)/ovaries, skin and joints. It causes damage to these tissues, which then leads to the symptoms below.

What are the symptoms of haemochromatosis?

Symptoms usually start between the ages of 30 and 50 years. The first symptoms are usually vague and may include feeling weak and tired, pain in the joints and pain in the tummy.

As haemochromatosis progresses, more specific symptoms develop but these are now much less common due to earlier diagnosis. These problems may include:

- Joint pain and swelling, especially the knuckle and the first joint of the first two fingers.
- Bronzing of the skin (looking like a permanent tan).
- Loss of sex drive.
- Loss of body hair.
- Impotence in men.
- Period changes in women. Women may have either no menstrual periods or very light menstrual periods. Early menopause may also occur in women with haemochromatosis.
- Poor memory.
- Feeling irritable.
- Depression.
- Developing diabetes.
- An increase in the size of the liver.
- ‘Scarring’ (cirrhosis) of the liver.
- Disease of the heart muscle (cardiomyopathy).

Most of these symptoms are found in other disorders and so diagnosis can be difficult. Arthritis found only in the knuckle and the first joint of the first two fingers is very suggestive of haemochromatosis.

The need for treatment to remove excess iron does not depend on the presence of symptoms. Because of the risk of developing a serious complication such as cirrhosis, treatment to remove iron buildup from the body is very important even if there are no symptoms.

How is haemochromatosis diagnosed?

The first tests are to see how much iron is stored in the body. This can be done by having a blood test for iron (ferritin) and transferrin saturations. Transferrin is a protein that transports iron in the blood. Transferrin is mostly made in the liver, and it regulates iron absorption. Transferrin saturation is the percentage of this protein which is already attached to iron. The levels of both iron in the blood and the transferrin saturation are usually high if you have haemochromatosis.

Initial tests will also check for any other possible causes of the symptoms. Blood tests will also be done for any possible complications of haemochromatosis such as problems with the liver or diabetes.

The test to confirm the diagnosis of haemochromatosis is the gene test for the HFE gene, which is abnormal in 9 out of 10 people with haemochromatosis. (See the section at the end of this leaflet to read more about this gene.)

A scan of the liver may help to detect how much iron is in the liver. Taking a liver sample (biopsy) used to be needed but is much less often done nowadays because a scan and a gene test can be done instead. However, a liver biopsy may be needed if the iron level in the body is very high or there seem to be other problems with the liver.

Other tests may be needed to check for complications of haemochromatosis, such as an ultrasound scan of the heart (echocardiogram).
Who should be tested?

Anyone with symptoms indicating possible haemochromatosis should be tested for the level of iron in the body. The gene test should be offered if high iron levels are detected.

Brothers, sisters and children of anyone who has haemochromatosis should be tested for the abnormal gene. The test should only be done after talking to a health professional about the possible benefits and problems of having the test.

The clear benefit of being tested is to have treatment early before any complications occur. Possible problems include the psychological impact of a positive test and difficulties obtaining insurance if the test is positive. See the separate leaflet called Genetic Testing for more details.

What is the treatment for haemochromatosis?

The simple and effective treatment consists of regular removal of blood, which is also known as venesection therapy or phlebotomy. This may need to be done frequently (every week) at first, depending on the level of iron overload in the body. The levels of iron in the body are monitored closely during treatment. Iron levels are reduced very slowly, often over quite a long period of time - it cannot be done very quickly. Once levels are normal, you will have regular blood tests to keep an eye on the iron levels. You may then need further blood removed every so often.

Regular blood removal will not cure some of the complications of haemochromatosis such as diabetes or liver 'scarring' (cirrhosis). Therefore, early diagnosis and treatment are very important.

Liver transplant may occasionally be needed if the liver is very badly affected.

Haemochromatosis cannot be cured as such, because you will always still have the faulty gene. However, with early diagnosis and the right treatment, symptoms and complications may be avoided.

You are likely to be under the care of a specialist - usually a haematologist, who will monitor your blood levels, and advise on treatment. If you have very high levels of iron, or any evidence of liver damage, you would also be referred to a liver specialist (a hepatologist).

Are there any foods I should avoid if I have haemochromatosis?

The increased levels of iron in the body cannot be treated by diet alone. Removing blood has a much bigger effect on reducing the levels of iron in the body. However, there are some recommendations of foods to avoid:

- Avoid vitamin supplements or tonics containing iron. This includes multi-vitamins and minerals. A balanced diet should provide all the vitamins and minerals you need.
- Avoid breakfast cereals which are heavily fortified with iron.
- Large doses of vitamin C should also be avoided because it increases the amount of iron absorbed from food eaten. Vitamin C also increases the amount of iron stored in the body.
- Reduce intake of offal (eg, liver and kidney) and red meat.

Tea, coffee and all milk products taken with a meal reduce the amount of iron absorbed from food. So you may find having tea, coffee or a milky drink with meals helps a little with iron levels.

Eating a healthy balanced diet is, of course, still the main thing to do. A very restricted or unusual diet will not make that much difference to iron levels, and could make you unwell in other ways.

Further information on diet is available from the Haemochromatosis Society - see the 'Further reading and references' section at the end of this leaflet. Your doctor may also be able to refer you to a dietician for advice.

Can I drink alcohol if I have haemochromatosis?

Alcohol can increase the rate at which iron is absorbed, particularly if you have alcohol with a meal. Excessive amounts of alcohol can also damage the liver itself, adding to the risk of liver problems.

If you wish to drink alcohol, keep it to sensible levels - an absolute maximum of 14 units per week. This should be spread out over the week rather than in ‘binges’. Try to avoid drinking alcohol with meals.

If your liver has been damaged by haemochromatosis, or if you have liver disease from any other cause, you should avoid alcohol altogether.

What are the complications of haemochromatosis?

The excess iron stored in body organs can cause damage. The possible complications of haemochromatosis include:

- Diabetes due to damage to the pancreas.
• Heart disease, for example it can lead to the heart muscles becoming weak (dilated cardiomyopathy) or stiff (restrictive cardiomyopathy), heart failure and abnormal heart rhythms.
• Liver ‘scarring’ (cirrhosis). People with haemochromatosis who develop cirrhosis are also at increased risk of liver cancer and should be checked regularly with ultrasound scans or magnetic resonance imaging (MRI) scans.
• Fertility problems.
• Joint damage.

What is the outlook (prognosis)?

If haemochromatosis is diagnosed and treated early before any complications develop the outlook is very good with no reduction in life expectancy. In most people, haemochromatosis is not a fatal condition and life expectancy is normal.

If complications do occur then the prognosis may be much worse. If this happens it will depend on which complication develops, and how severe it is. Your specialist can advise on your individual outlook.

How is haemochromatosis inherited and how common is it?

Haemochromatosis is a genetic condition. 'Genetic' means that you are born with it and it is passed on through families through special codes inside cells called genes. Genes come in pairs.

If you have haemochromatosis, one of your gene pairs does not work properly. A few different genes may be involved but 9 out of 10 people with haemochromatosis have an abnormal ‘HFE’ gene, which is on chromosome 6.

Haemochromatosis is a 'recessive' disorder. This means that haemochromatosis will only occur if both copies of the gene are abnormal. If only one copy is defective, a person will be perfectly healthy but will be a ‘carrier’. This means he or she will be able to pass on the abnormal gene to a son or daughter.

There is now a gene test which can help to diagnose most people who have haemochromatosis.

When two people who carry the abnormal gene have a child, there is a:

• 1 in 4 chance that the child will have haemochromatosis (by inheriting the abnormal gene from both parents).
• 2 in 4 chance that the child will not have haemochromatosis but will be a carrier (by inheriting the abnormal gene from one parent but the normal gene form the other parent).
• 1 in 4 chance that the child will not have haemochromatosis and will not be a carrier (by inheriting the normal gene from both parents).

These proportions are averages for the whole population and in any one family with both parents being carriers, it would be possible for all children to be affected, all to be carriers, or for all not to be affected or be carriers.

How recessive inheritance works when both parents are carriers (n = normal gene; H = gene for haemochromatosis):

![Diagram showing recessive inheritance in haemochromatosis](image)

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Haemochromatosis occurs in people from all parts of the world but is most common in people from Northern Europe. Surveys have shown that around 1 in 250 of people of Northern European origin have the abnormal gene and so are likely to be at risk of developing iron overload. Haemochromatosis is now recognised as being one of the most common genetic disorders.
Further reading & references

- Healthy eating and haemochromatosis. The Haemochromatosis Society.

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Document ID: 4857 (v44)  Last Checked: 16/12/2018  Next Review: 15/12/2021

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