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Haemochromatosis

Haemochromatosis is an inherited (genetic) disorder which causes the body to absorb too much iron from the diet. This excess iron causes damage to organs in which it builds up. 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?

People with haemochromatosis have inherited a faulty gene, which directs the body to absorb too much iron from what they 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, ovaries, skin and joints. It causes damage to these tissues, which leads to a variety of symptoms.

Haemochromatosis symptoms

Symptoms of haemochromatosis usually start between the ages of 30 and 50 years. The first symptoms are usually vague and may include feeling weak and tired, and having joint pains or abdominal pains.

As haemochromatosis progresses, more specific symptoms develop due to organ damage but these are significantly less common nowadays due to earlier diagnosis. However there are still many cases each year in people who have not yet been diagnosed.

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.

Causes of haemochromatosis

Haemochromatosis is a genetic condition. It is passed on through families through codes inside cells called genes. Genes come in pairs. In haemochromatosis, one of the 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 an autosomal recessive disorder. This means that haemochromatosis will only occur if both copies of the gene are abnormal (one copy from the mother and one from the father). If only one copy is defective, a person will be 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.

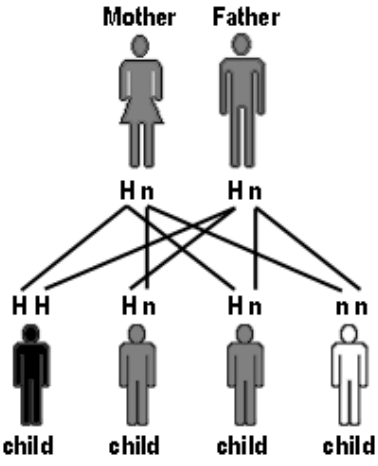
There is now a gene test which can help to diagnose most people who have haemochromatosis.

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):



How common is haemochromatosis?

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 150 of people of UK origin have haemochromatosis though it is much more common in Northern Ireland with closer to 1 in 100 people affected. Approximately 1 in 10 people carries one copy of the abnormal gene in Northern Europe and is therefore a carrier of haemochromatosis; this is 1 in 5 people in Northern Ireland. Haemochromatosis is now recognised as being one of the most common genetic disorders.

How is haemochromatosis diagnosed?

Blood tests

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 in 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.

Gene test

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.)

Scans

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. In 2018, insurance companies in the UK agreed not to require positive genetic tests to be revealed when offering life insurance. See the separate leaflet called Genetic Testing for more details.

Haemochromatosis treatment

Treatment of haemochromatosis is simple and effective. It 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. Once levels are normal, regular blood tests monitor the iron levels. Further blood may then need to be 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 because the faulty gene cannot be removed. However, with early diagnosis and the right treatment, symptoms and complications can be avoided.

A haematologist will usually monitor the blood levels and advise on treatment. If there are very high levels of iron, or any evidence of liver damage, a liver specialist (a hepatologist) will also be involved in the management.

Foods to avoid if you 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 which can help to control iron levels. These are to:

- Avoid vitamin supplements or tonics containing iron. This includes multi-vitamins and minerals. A balanced diet should provide all the vitamins and minerals needed.
- 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.

Eating a healthy balanced diet is still important. A very restricted or unusual diet will not significantly affect iron levels, and could cause other symptoms.

Further information on diet is available from the Haemochromatosis Society - see the 'Further reading and references' section at the end of this leaflet.

Alcohol and haemochromatosis

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

When drinking alcohol, it is important to 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'. It is sensible to try to avoid drinking alcohol with meals.

Alcohol should be avoided completely if there is liver damage from haemochromatosis or if there is any other liver disease.

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. This depends on which complication develops and how severe it is.

Further reading

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