GESA
Gastroenterological Society of Australia

Information about

Haemochromatosis

What is haemochromatosis?

Haemochromatosis is a genetic disorder that causes too much iron to be stored in the body, potentially damaging organs like the liver and resulting in serious disease.

Iron is essential to life, but only in small amounts. It plays an important role in our health, particularly for helping haemoglobin, the blood protein that carries oxygen to the rest of the body.

Normally, iron from food is taken into the body through the intestine (a process known as absorption). In people with haemochromatosis, too much iron is absorbed. Once this happens, the body has no way of getting rid of the extra iron. The normal level of iron in the body is about 3 to 4 grams, but people with haemochromatosis sometimes have more than 20 grams.

When the extra iron is stored in various joints and organs and increases over time, it can cause health problems. This can include damage to vital organs, so it's essential to have this disease diagnosed and treated at an early age.

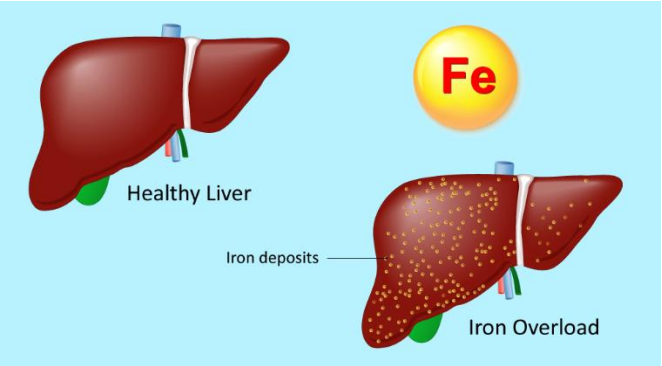
Prevalence

Haemochromatosis is one of the most common genetic diseases in our society, although many people are only mildly affected. About one in every 300 people has haemochromatosis, while about 12% of the Australian population are "carriers" of the haemochromatosis gene.

Cause

Haemochromatosis is usually caused by changes in a gene (the *HFE* gene) that have made it faulty. This is the most common genetic disease in people of Northern European descent.

People who inherit one haemochromatosis gene and one normal gene from their parents are known as carriers. Their iron absorption may be slightly higher than normal,



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but most of them don't absorb enough iron to cause any major health problems. If two carriers have children, each of their children has a 50% chance of inheriting one haemochromatosis gene and a 25% chance of inheriting two haemochromatosis genes.

People who inherit two haemochromatosis genes often absorb too much iron. This iron slowly builds up in their joints, liver, heart, pancreas and other hormonal glands. It takes many years to build up iron to a level that can damage these organs, but by the time the damage happens, it is often too late for the organ to repair itself, so there may be permanent damage. The good news is that carriers can be identified using a simple blood test for the *HFE* gene.

Symptoms

Diagnosing haemochromatosis can be hard because the symptoms can be very different between patients and are similar to symptoms of many other medical conditions.

Most people with haemochromatosis have their first symptoms when they are between 30 and 60 years old.

Symptoms may include:

- fatigue (tiredness)
- weakness
- weight loss
- upper abdominal (belly) discomfort
- joint pain, usually in the fingers
- a tan, not from sun exposure.

Other symptoms may develop later because of damage to the liver, heart or pancreas or from other hormonal deficiencies. These symptoms might include palpitations (where your heart races, flutters or thumps), shortness of breath, chest pain, thirst or frequent urination as a result of diabetes, loss of sex drive or loss of body hair. But most young people with haemochromatosis don't have symptoms or have only minor symptoms in the early stages of the disease.

Risk factors

People who may be at risk of haemochromatosis include:

- blood relatives of people with the disease (particularly close relatives, like brothers, sisters and children)
- people with symptoms of the disease
- people with diabetes, arthritis, some heart problems or chronic fatigue
- people with liver disease where the cause is unknown.

If you have some of the symptoms mentioned above, don't panic by assuming that you have haemochromatosis – many other conditions have similar symptoms. The best thing to do is see your family doctor and talk about your concerns.

Diagnosis

People with any family history of haemochromatosis

All close relatives of people with haemochromatosis – their brothers, sisters, parents and children – should be screened for it. Cousins, aunts and uncles should also be screened, although their risk is much lower. Children in the family can usually wait until adolescence to have their screening done.

Screening involves a simple blood test for the *HFE* gene. After discussing it with your GP, this test can be done through pathology providers. Some people will need to have their iron levels checked every 2 to 3 years, as sometimes the excess iron doesn't become apparent until later in life.

A liver biopsy (where a small piece of liver is removed under local anaesthetic) is only needed if the liver appears to be damaged.

People without any family history of haemochromatosis

The two most useful blood tests for these people to start with are:

- serum transferrin saturation (the best test for finding the disease in its early stages)
- serum ferritin (which may give normal results in early stages of the disease).

Both tests can be done on the same sample of blood. If the test results are abnormal at least twice, another blood test looking for the *HFE* gene may be all that is needed to confirm the diagnosis of haemochromatosis.

Treatment

Haemochromatosis can be treated by removing about 500 mL of blood (which contains about 250 mg of iron), usually once or twice a week. This stimulates the body to make more blood, which uses up the extra iron. This is known as venesection treatment. Depending on the amount of iron in the body, this treatment may take 1 to 2 years to start with.

Blood tests are done to monitor your levels of iron as it is removed. Once the extra iron has been removed, venesections are done about three or four times a year to stop iron from building up again. This makes it an important life-long treatment.

Venesection treatment must only be done by experienced medical or nursing staff. It can be done at some hospitals, pathology laboratories or general practices. Venesections can also be organised through the blood bank. The Australian Red Cross Lifeblood provides a therapeutic venesection service for people with Haemochromatosis. This service requires an online referral by your doctor, a specialist or a general practitioner, via the High Ferritin App. (https://highferritin.transfusion.com.au).

Just before a venesection, you should rest for 15 minutes and drink 500 mL of fluid. After the venesection, stand up

slowly and sit in a chair for 15 minutes, keeping pressure on your arm (where the needle was inserted) for 5 minutes.

You don't need to follow a low-iron diet, but you may choose to reduce how much red meat you eat (e.g. to 90 to 120 g/day). You shouldn't take vitamin C supplements because they can increase iron absorption. As with any liver disease, you should keep how much alcohol you drink to a minimum (less than 20 g/day), but if you have liver damage, your doctor may advise drinking no alcohol.

Results

There is a lot of encouraging news for people with haemochromatosis, and the most important thing is getting an early diagnosis.

There is good evidence that removing extra iron makes people feel better and stronger, lessens their tan, makes their liver smaller and improves their diabetes and heart function.

If you start treatment early in the disease, damage to your liver and other organs may be completely prevented. If your liver has scarring (known as cirrhosis), this is usually not reversed by treatment, but it should not get worse. As cirrhosis increases the risk of liver cancer, a 6-monthly ultrasound surveillance program is recommended for people with cirrhosis.

People with haemochromatosis who have been treated early in the disease and who do not have cirrhosis have a normal life expectancy.

Further information

Some large hospitals have support groups for patients and relatives with liver diseases and haemochromatosis.

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Acknowledgements
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