

Information about

# Haemochromatosis

## What is Haemochromatosis?

Haemochromatosis is a genetic disorder in which too much iron is taken into the body potentially causing damage to organs, such as the liver, and resulting in serious disease.

Iron is essential to life, but only in small quantities. It plays an important role in our health, particularly for the function of haemoglobin which is the blood protein that carries oxygen to the tissues.

Normally, iron is taken into the body from food via the intestine (a process known as absorption). In people with haemochromatosis too much iron is absorbed and once this occurs the body has no way of eliminating excess amounts. The normal level of iron in the body is about 3-4 grams; in people with haemochromatosis this is sometimes more than 20 grams.

Health problems occur when the excess amounts of iron that are stored in various joints and organs increase over time. As this can damage vital organs, it is essential that this disease is diagnosed and treated at an early age.

## Prevalence

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

## Cause

Haemochromatosis is usually caused by changes in the HFE gene which have made it faulty. This is the most prevalent genetic disease in people of Northern European descent.

Individuals inheriting one haemochromatosis gene and one normal gene are known as carriers. Their iron absorption may be slightly higher than normal but most do not absorb enough iron to cause any significant health problems. If two carriers have children, each of their children has a 25 per cent chance of inheriting two haemochromatosis genes and a 50 per cent chance of inheriting one haemochromatosis gene.

Individuals inheriting two haemochromatosis genes often absorb too much iron. This iron slowly builds up in the joints, liver, heart, pancreas and other hormonal glands. It takes many years to build up iron to a level which causes damage to these organs, but by the time the damage occurs it is often too late for the organ to repair itself and some permanent damage may remain.

The good news is that carriers can be identified by a simple test for the HFE gene.



An information leaflet for patients and interested members of the general public prepared by the Digestive Health Foundation

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## Symptoms

Symptoms of haemochromatosis vary considerably among patients, and may resemble those of many other medical conditions, making diagnosis difficult.

In the majority of people with haemochromatosis, the first symptoms develop between 30 and 60 years of age.

Symptoms may include:

- Fatigue
- Weakness
- Weight loss
- Upper abdominal discomfort
- Joint pain, usually in the fingers
- A tan, not due to sun exposure.

Other symptoms may develop later as a result of organ damage to the liver, heart (e.g. palpitations, shortness of breath, chest pain), pancreas (e.g. thirst or frequent urination as a result of diabetes) or other hormonal deficiencies (e.g. loss of sex drive or body hair). However, most young people with haemochromatosis have no symptoms or only minor symptoms in the early stages of the disease.

## Risk factors

People who may be at risk of haemochromatosis include:

- Blood relatives of affected individuals (particularly close relatives - brothers, sisters and children).
- Individuals with symptoms of the disease.
- Individuals with diabetes, arthritis, certain heart problems or chronic fatigue
- Individuals with liver disease where the cause is unknown.

If you have some of the symptoms mentioned above, don't panic and assume that you have haemochromatosis - many other conditions may have similar symptoms. The best thing to do is see your family doctor and discuss your concerns.

## Diagnosis

### People with a family history of haemochromatosis

All close relatives - brothers, sisters, parents and children - should be screened for haemochromatosis. Cousins, aunts and uncles should also be screened, although the risk is much lower. Younger family members can usually have their screening deferred until adolescence.

Screening involves a simple blood test for the HFE gene to see if they are at high risk of haemochromatosis. This test is available (after discussion with your GP) through pathology

laboratories. Some people will need to have their iron levels checked every 2-3 years, as sometimes the excess iron does not become apparent until later life.

### Early diagnosis and treatment of family members with the disease is essential to prevent organ damage.

A liver biopsy (which involves the removal of a small piece of liver under local anaesthetic) is only required if the liver appears to be damaged.

### People without any family history of haemochromatosis

The two most useful initial blood tests for these individuals are:

- Serum transferrin saturation (the best test for detecting early stages of the disease)
- Serum ferritin (which may produce normal results in early stages of the disease).

Both tests can be done on the one sample of blood. If the tests are abnormal on at least two occasions, a further blood test looking for the HFE gene may be all that is required to confirm the diagnosis of haemochromatosis.

## Treatment

By removing about 500 ml of blood (which contains approximately 250 mg of iron), usually once or twice a week, the body is stimulated to make more blood and this uses up the excess iron. This is known as venesection treatment. Depending on the amount of iron in the body, the initial treatment may take 1-2 years.

Blood tests are done to monitor the iron removal. Once the excess iron has been removed, venesections are done about 3-4 times a year to prevent iron building up again, making it an important life-long treatment.

Venesection treatment must only be done by medical or nursing staff experienced in this technique. Venesections can be done at some hospitals, some pathology laboratories or some general practices. On occasions, venesections can be organised through blood banks, after referral from a specialist.

Immediately prior to a venesection, 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 for 5 minutes where the needle was inserted.

There is no need to follow a low iron diet, however people may choose to reduce red meat intake (e.g. to 90-120 g/day). Vitamin C supplements should be avoided as they can increase iron absorption. As with any liver disease, alcohol should be kept to a minimum (less than 20 g/day), but if there is liver damage your doctor may advise no alcohol.

## Results

There is a lot of encouraging news for people with haemochromatosis and the key is early diagnosis.

There is good evidence that shows with removal of excess iron people feel better and stronger, their tan lessens, liver size decreases, diabetes may improve and heart function improves.

If treatment has commenced early, damage to the liver and other organs may be completely prevented. If cirrhosis (liver scarring) is present it is usually not reversed by treatment, but should not get worse. There is an increased risk of liver cancer if a patient has cirrhosis, therefore a six monthly ultrasound surveillance program is recommended.

People with haemochromatosis who have been treated early and 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.

### Haemochromatosis Society of Australia

412 Musgrave Road

Coopers Plains QLD 4108

**Telephone** 1300 019 028

**Website** <http://www.haemochromatosis.org.au>

## Digestive Health Foundation

This information booklet has been designed by the Digestive Health Foundation (DHF) as an aid to people who have haemochromatosis or for those who wish to know more about it. This is not meant to replace personal advice from your medical practitioner.

The DHF is an educational body committed to promoting better health for all Australians by promoting education and community health programs related to the digestive system. The DHF is the educational arm of the Gastroenterological Society of Australia, the professional body representing the specialty of gastrointestinal (GI) and liver disease in Australia. Members of the Society are drawn from physicians, surgeons, scientists and other medical specialties with an interest in GI disorders.

Since its establishment in 1990 the DHF has been involved in the development of programs to improve community awareness and the understanding of digestive diseases. Research and education into gastrointestinal disease are essential to contain the effects of these disorders on all Australians.

Further information on a wide variety of gastrointestinal conditions is available on our website.

**dhf**

Digestive Health Foundation

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