WHAT IS HAEMOCHROMATOSIS

Haemochromatosis (heem-a-krome-a-toe-sis) is a genetically inherited disorder involving excessive iron absorption and inappropriate storage of iron. The stored iron levels may potentially become high enough to cause damage to major organs, especially the liver, but also the heart, pancreas, joints and the organs that produce hormones.

Haemochromatosis is considered to be the most common genetic disease in people of European ancestry. To be diagnosed with hereditary haemochromatosis you will have inherited a faulty gene from each parent. People with one gene mutation are known as a carriers of haemochromatosis. Although there are other forms of iron overload, the incidence and/ or the genetic cause is not well known.

What is iron?
Iron, a mineral essential for life, is found in plants, soil and animals. All foods contain iron of varying concentrations, some contain more than others. Iron is part of the haemoglobin molecule in the red blood cells, which carries oxygen from the lungs to all the tissues in the body.

During digestion the body absorbs the iron it requires through the intestines (bowels). In people with haemochromatosis, the absorption of iron is uncontrolled and excessive amounts are absorbed over a long period of time. The excess iron gets stored in the liver and can accumulate in other organs such as the heart and pancreas.

What is ferritin?
Ferritin is the protein that stores iron. The amount of iron in the body can be measured by doing a blood test called the serum ferritin test. The result reflects the amount of stored iron in the body and is used to monitor patients with hereditary haemochromatosis.

What are the symptoms of haemochromatosis?
Many people with haemochromatosis have no symptoms and the condition is detected by testing the serum ferritin through a routine blood test. Symptoms of haemochromatosis do not appear in any particular order and may be different from person to person. As iron accumulates over many years, variable symptoms and signs may appear, such as:

- Tiredness/fatigue/weakness
- Joint pain or abdominal pain
- Low libido (loss of sex drive)
- General malaise (discomfort or physical unease)
- If the ferritin levels are very high, the skin/ complexion may darken

These symptoms are variable and non-specific and can easily be attributed to other unrelated health problems. Thus the condition can go unrecognised for some years. If the iron stores reach a very high level, there can be a risk of serious problems occurring.

How is haemochromatosis diagnosed?
Haemochromatosis is diagnosed through blood tests that check the iron saturation and the serum ferritin levels, followed by genetic testing (Haemochromatosis gene studies).

Blood tests for liver function tests (LFTs) will show any liver damage. If the LFTs are abnormal a MRI scan might be recommended which can show the degree of iron overload in the liver cells. A liver biopsy might occasionally be necessary to determine if the tissue has severe damage and can also exclude other coexistent disorders. Other blood tests may include blood glucose, fasting lipids and CRP (a test to exclude inflammation which can falsely elevate the ferritin).

The doctor will also take a full medical history including a family history of haemochromatosis.

Once a diagnosis has been made, it is usually recommended that close blood relatives (siblings, children, parents) are screened for the condition. Aunts, uncles and cousins should also be notified that they may be at risk. There is no evidence that genetic haemochromatosis results in significant problems before adulthood and therefore all those at risk should be tested in early adulthood, ideally before 30 years old.

Whilst haemochromatosis cannot be prevented, early diagnosis and treatment is likely to prevent the development of symptoms and organ damage (ideally before the damage is permanent).

Haemochromatosis – Blood test investigations
Ideally the initial blood tests are best done on a fasting sample, which means you are told not to eat & drink water only for a defined period before the blood test – usually overnight fast.

The Iron related tests – Iron, transferrin & ferritin, have age and gender dependent reference intervals, hence these results should be interpretated considering local laboratory reference intervals. These investigations, will assess:

- Amount of iron being transported – Transferrin saturation*
- Amount of iron stored – Ferritin*
- (Haemochromatosis gene status –investigation performed if unexplained abnormality)

*Note there are many causes for this investigation to be abnormal & your doctor will need to consider all information.

Monitoring and treatment
It is important to note that not everyone will need treatment. However once diagnosed with haemochromatosis you are likely to need regular blood tests. If you have a high ferritin level, then treatment will likely be required in order to prevent (further) organ damage.

Treatment for haemochromatosis is called therapeutic venesection or phlebotomy (taking blood). Blood is removed from the arm into a bag or bottle, similar to donating a unit of blood. Once the serum ferritin is lowered to the level requested by your specialist, there is likely to be a period of rest before moving to the maintenance phase. During the maintenance phase the frequency of venesection can vary greatly, depending on how rapidly iron is absorbed by each person. The usual frequency would be 3 or 4 monthly venesection, sometimes less.

Some people might be referred to be a blood donor if they fit the criteria. People usually have to be generally healthy, under the age of 60 years old and have no other diseases or chronic infections. This is a good option for people if a blood centre or mobile collect is relatively close to their home or work location. It also helps other patients who need treatment at the hospitals and takes pressure off the public hospitals.

There is no cure for haemochromatosis, but treatments can keep the iron overload under control. Venesections are a safe, effective and economical way of removing iron from the body, as iron moves out of storage and is used to replace the red blood cells.

At what level should treatment begin?
Treatment should begin if serum ferritin levels are significantly raised. The exact level where treatment is recommended will depend on your symptoms, haemochromatosis genotype, your age and general health, particularly if you have any other diseases or conditions.

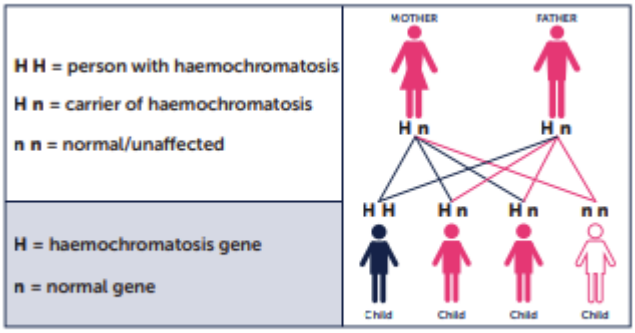
Talk to your health professional (GP or haematologist) who will set up a treatment schedule for you.

Where do I go for venesection?
Treatment takes place in New Zealand in a variety of settings depending on what region you live in. These include hospital outpatient clinics, the New Zealand Blood Service (NZBS), community laboratories, and occasionally in some GP practices.

Tips for venesection:
- Do not have a venesection while fasting or if you are unwell.
- Drink plenty of water the day before and on the day of the venesection to ensure you are well hydrated. This aids recovery and prevents your blood pressure dropping.
- Listen to your body – take time to rest after the procedure.
- Protect the arm used for venesection – avoid heavy lifting especially for that day.
- Ask to venesect from the arm that you do not use for writing and other tasks.

A venesection record book is available from Leukaemia & Blood Cancer New Zealand which will help you to track your blood test results and treatments. Phone 0800 15 10 15 to request a copy or visit our website www.leukaemia.org.nz.

Who gets haemochromatosis and how?
Our genes come in pairs, one inherited from each parent. To have haemochromatosis a person needs to have a fault in both of their HFE genes. If a person inherits just one haemochromatosis gene, they are a carrier and do not have the condition itself.



Genetic inheritance

There are a small number of different mutations that can cause haemochromatosis, and the risk of developing haemochromatosis varies dependent on combinations of these mutations.

Mutation	Risk of iron overload
Two *C282Y* mutations	Greatly increased risk
One *C282Y* mutation	No increased risk (carrier)
Two *H63D* mutations	No increased risk (carrier)
One *H63D* mutation	No increased risk (carrier)
One of each *C282Y* and *H63D*	Small increase in risk

Looking after yourself

Making good lifestyle choices are important for everyone but are particularly important for people with medical conditions and who are receiving treatment. Some of the lifestyle choices that are important include nutrition, exercise, sleep and reducing stress.

Nutrition

Haemochromatosis cannot be treated by diet, however there are small changes that can be made to reduce the risks of iron overload.

- Avoid iron supplements.
- Consume red meats in moderation.
- Consume alcohol in moderation. **Any alcohol consumed** can increase the liver damage that occurs with severe iron overload. If you have liver damage from excess iron (or another cause), you should not consume alcohol.
- Limit supplemental or other forms of vitamin C (such as orange juice) with your meals as this will enhance iron absorption.
- When your serum ferritin is very high, do not consume raw seafood (a bacteria called vibrio vulnificus can cause rapid and life threatening septicaemia. It is rare but not unknown in New Zealand waters).
- If you have fatty liver disease or diabetes, seek advice on specific dietary restrictions.
- Note that tannin (mainly in black tea and some coffees) binds to iron in the diet (other than that in meat) and helps to prevent further iron absorption.