leukaemia & blood cancer NEW ZEALAND

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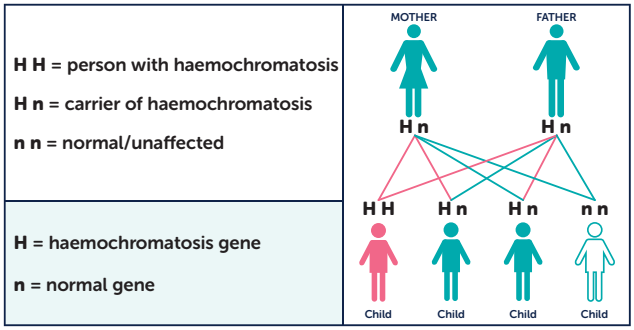
HAEMOCHROMATOSIS

A fact sheet for patients, families and whānau

WHAT IS HAEMOCHROMATOSIS?

Haemochromatosis (heem-o-krome-a-toe-sis) is an inherited disorder that results in too much iron being absorbed and stored. Too much stored iron can cause damage to major organs such as the liver, heart, pancreas and joints.

The most common form of haemochromatosis is caused by mutations in the HFE gene. Haemochromatosis is a recessive condition, which means an affected HFE gene is inherited from both parents. A person who only inherits one affected gene is known as a carrier. Approximately one in 200 New Zealanders have haemochromatosis (two affected genes), however not all of these people will have symptoms or need treatment.



Genetic inheritance

The most common HFE gene mutations are C282Y and H63D. The risk of developing iron overload depends on which gene mutations are present and whether one or two copies are affected. Table 1 shows which mutations affect a person's risk of iron overload.

Table 1.

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

*Iron overload in this case is only likely if additional risk factors (such as metabolic syndrome) are present.

Haemochromatosis can affect anyone; however, it is particularly common in people of European ancestry. There are also other forms of iron overload, but less is known about the genetic causes of these.

What is iron?

Iron is an essential mineral which is found in plants, soil and animals. In humans, iron can be found in the haemoglobin molecule in red blood cells, which carries oxygen from the lungs to all the tissues in the body.

All foods contain iron – some contain a little, while others contain a lot. Some sources of iron are absorbed more easily than others. During digestion the body absorbs the iron it needs through the intestines (bowels). In people with haemochromatosis, the absorption of iron is uncontrolled and too much iron is 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, causing damage.

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 a serum ferritin test. The result reflects the amount of stored iron in the body and is used to monitor patients with haemochromatosis.

What are the symptoms of haemochromatosis?

Many people with haemochromatosis have no symptoms and the condition is detected by testing the serum ferritin on a routine blood test. Symptoms of haemochromatosis appear as a result of iron accumulation (iron overload) over many years. They can appear in any order and may be different from person to person.

Some signs or symptoms of iron accumulation are:

- Tiredness, fatigue or weakness
- Joint pain or abdominal pain
- Low libido (loss of sex drive)
- General malaise (discomfort or physical unease)
- Changes in skin colour/complexion

These symptoms are non-specific and can easily be attributed to other unrelated health problems. This can make haemochromatosis difficult to diagnose, and the condition can go undetected. Serious damage can occur if iron stores reach very high levels.

How is haemochromatosis diagnosed?

Haemochromatosis is diagnosed through blood tests that check the amount of iron in your blood (iron saturation and serum ferritin). Other blood tests may be done at the same time to rule out other causes of high serum ferritin, such as blood glucose, fasting lipids and c-reactive protein (a marker of inflammation).

Your doctor may also order a blood test to check for liver damage (liver function tests, LFTs). If the LFTs are abnormal an 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 to exclude other disorders.

Your doctor will take a full medical history, including a family history of haemochromatosis. Genetic testing will also be carried out to confirm haemochromatosis and identify which genes are affected.

Once a diagnosis has been made, it is usually recommended that close blood relatives (siblings, children and parents) are screened for the condition. Aunts, uncles and cousins should also be notified that there is a chance they have inherited haemochromatosis. Most of the complications of haemochromatosis occur in adulthood, therefore it is recommended that those at risk should be tested in early adulthood (from the age of 18), ideally before the age of 30.

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

Blood tests results

Table 2 shows the normal ranges for serum ferritin and transferrin saturation (iron saturation) in adults. These tests are usually done on a fasting sample, which means that you will not eat or drink for a period of time before the blood test. If the results of these tests are above the normal range, they are often repeated to double check the results, before requesting testing for the gene mutations. Ferritin levels above 1000 µg/L indicate a severe issue.

Table 2.

Normal range for adults		
	Adult men	Adult women
Serum ferritin	20-300 µg/L	20-200 µg/L
Transferrin saturation	10-50%	10-45%

µg/L = micrograms per litre

Treatment and monitoring

Not everybody who is diagnosed with haemochromatosis will need treatment. However, you are likely to need regular blood tests. If you have a high ferritin level, then treatment will likely be required to prevent (further) organ damage.

Treatment for haemochromatosis involves removing blood, which is called venesection or therapeutic phlebotomy. Blood is removed from a vein in your arm into a bag or bottle. After the blood is removed from your body, some of the excess stored iron is used to make replacement blood cells in your body, reducing serum ferritin.

Treatment typically begins if serum ferritin levels are significantly raised. The exact level at which you begin treatment will depend on your symptoms, haemochromatosis genotype (which genes are affected), 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.

Venesection locations vary from region to region, and include hospital outpatient clinics, the New Zealand Blood Service (NZBS), community laboratories, and some GP practices.

Your serum ferritin levels will be monitored regularly, and once they have reached the desired level, you will move into a maintenance phase where your venesections are less frequent. In the maintenance phase, venesections may only be needed every 3-4 months, although this can vary from person to person, depending on how rapidly iron is absorbed.

Being a blood donor

Donating blood has the same effect as venesection. Some people might be referred to be a blood donor if they are:

- Generally healthy
- Have no other diseases or chronic infections
- Age 71 or less

This is a good option for people if a blood centre is convenient to them. The donated blood goes through the usual screening processes and is used to help other people who need it.

Tips for venesection:

- Do not have a venesection while fasting (not eating or drinking) 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 for blood to be taken 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.

Nutrition

Haemochromatosis cannot be treated by diet, but 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, you should not consume alcohol.
- Avoid vitamin C supplements and limit high-vitamin C foods (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. People with iron overload are at increased risk of infection with a bacteria (Vibrio vulnificus) which can sometimes be found in raw seafood.

Important information available online

For more information and to download other fact sheets, see our website **www.leukaemia.org.nz**

QUESTIONS & NOTES