Haemochromatosis

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. It is caused by gene mutations. To be diagnosed with haemochromatosis you 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 in other races, 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 iron through the intestines (bowels). Under normal circumstances iron is not easily absorbed. In people with haemochromatosis, the absorption of iron is uncontrolled and excessive amounts are absorbed over a long period of time. The excess iron which is not used, gets deposited in body organs, especially the liver, and 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 body iron and this test is useful to monitor high and low body iron levels.

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 the 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 tissue iron level reaches a very high level, there can be a risk of serious problems occurring.

Symptoms of high levels of iron overload in organs:

Liver

- Enlarged liver (hepatomegaly) and/or pain in the area of the liver (under the right rib cage)
- Jaundice (yellow colour of the skin or the whites of your eyes from liver damage)
- Reduced body hair, itching and bruising easily
- Scarring of the liver (cirrhosis)

Heart

- Irregular heart beat or palpitations
- Shortness of breath, especially when active
- Fatigue
- Swollen ankles

Pancreas

- Thirst
- Increased need to urinate
- Tiredness
- Skin infections that do not heal well
- Blurry vision
- Dizziness
- Always feeling hungry, weight gain or weight loss

Joints

- Arthritis - most commonly in the joints of the hands and feet, wrists, shoulders, knees, ankles and hips

Sex organs in women

- Irregular periods
- Early menopause
- Loss of libido (low sex drive)

Sex organs in men

- Loss of libido (low sex drive)
- Shrinking testicles
- Enlarged breast tissue (gynaecomastia)
- Impotence (inability to get or maintain an erection)

How is haemochromatosis diagnosed?

Haemochromatosis is diagnosed through blood tests that check the iron saturation (how much iron is attached to the transport protein in the blood) and the serum ferritin levels (how much iron is stored).

Blood tests for liver function tests (LFTs) will show any liver damage. If the LFTs are abnormal a MRI liver 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 or not. It can also exclude other coexistent

disorders. A liver biopsy is done by an experienced medical person using a special needle; the small amount of liver tissue removed from the liver is then analysed under the microscope by a pathologist.

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.

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).

Reference ranges for Haemochromatosis Iron Tests

Male

Serum ferritin:	20-300 µg/L*
Transferrin saturation:	10-50%

Females

Serum ferritin:	10-200 µg/L*
Transferrin saturation:	10-45%

*µg/L = micrograms per litre

These tests are best done on a fasting sample which means you are told not to 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 the gene mutations.

Ferritin levels > 1000 µg/L indicate a severe issue.

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). If you are having venesections you will most likely go to the nearest blood centre or hospital and have an IV (intravenous) line inserted. Blood is then removed 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. This treatment is a safe, effective, and economical way of removing iron from the body.

How can removal of blood remove stored iron?

Your body contains about 10 mls of blood/kg of body weight. Your bone marrow is the blood cell factory of your body. The life-span of red blood cells is usually 100-120 days. If you lose blood by bleeding, venesection or donating blood, your bone marrow replaces the blood as long as you have enough iron, vitamin B12 and folate which are some of the ingredients used to make new red blood cells.

After blood is removed by venesection, iron that is stored in the body moves out of storage and is used to replace the new red blood cells.

At what level should treatment begin?

Treatment should begin if serum ferritin levels are raised above the normal range. The exact level where treatment is recommended will depend on your symptoms, 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 use 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?

Haemochromatosis is an inherited condition. To be affected you must receive a faulty (mutated) gene from each parent. We have about 25,000 pairs of genes in each cell. Haemochromatosis occurs due to a person inheriting gene faults called mutations. Mutations in a number of different genes can cause haemochromatosis.

Most European people with the condition have a mutation in the HFE gene.

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. Refer to the diagram below.

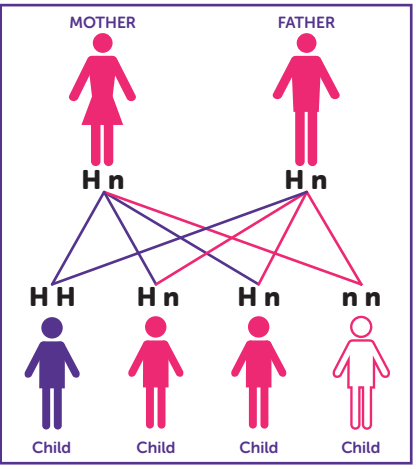
H H = person with haemochromatosis

H n = carrier of haemochromatosis

n n = normal/unaffected

H = haemochromatosis gene

n = normal gene



Genetic inheritance

Who should be tested?

If a person has haemochromatosis due to an abnormality in the HFE gene then their relatives (parents, siblings and children) are at increased risk of also having haemochromatosis.

Brothers and sisters have at least a 1 in 4 chance of inheriting a gene from each affected parent. Children of one parent who has haemochromatosis have around 1 in 20 chance of having the condition.

Children

It is recommended that, rather than testing young children directly, the other parent is tested in the first instance. There is no evidence that genetic haemochromatosis results in significant problems before adulthood and therefore there is no need to test young children. All those at risk though, should be tested before the age of 30 years old.

Other close relatives, including aunts, uncles and cousins, should also be tested.

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

Ensure that you have a healthy nutritious diet; however haemochromatosis cannot be treated by diet.

- 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

Helpful contacts

Leukaemia & Blood Cancer New Zealand
www.leukaemia.org.nz. Please contact Support Services on 0800 15 10 15 or supportservices@leukaemia.org.nz.

NZ Blood Service
www.nzblood.co.nz or 0800 448 325. To find a location near you where you can donate blood go to: https://www.nzblood.co.nz/give-blood/where-to-donate/. You may need a referral to the Blood Service from your consultant.

For more information please contact Support Services on 0800 15 10 15 or supportservices@leukaemia.org.nz

leukaemia.org.nz

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