melbourne haematology

fact sheet

Haemochromatosis

What is Haemochromatosis?

Haemochromatosis is a condition caused by too much iron in the body. People with haemochromatosis absorb too much iron from the intestine.

What causes haemochromatosis?

The most common form is caused by a gene change (mutation) in the protein that controls iron absorption in the intestine, called the C282Y mutation. This causes a protein in the intestinal lining to not work properly, leading to excess iron absorption.

Another gene change, called H63D, may be associated with a smaller risk of developing haemochromatosis.

How did I get haemochromatosis?

Haemochromatosis is a genetic condition, inherited from your parents. We all have two copies of each gene. Haemochromatosis occurs when you have two copies of the abnormal C282Y gene, one inherited from each parent. Your parents may have only one copy each and so may not have developed the condition themselves.

What are the problems with having haemochromatosis?

The body needs a certain amount of iron to function properly, used by many body systems including the blood, bone marrow, and brain. Iron levels are usually closely regulated; too much iron is not good for the body. Excess iron can affect the heart, pancreas, hormone glands (pituitary gland), and the liver. Some people with high iron levels experience tiredness, lack of energy, joint pains, and darker skin colour (hyperpigmentation).

What are the normal levels of iron in the body?

Iron in the body is measured using a blood test called serum ferritin, a protein that carries iron around the body — a high serum ferritin usually indicates too much iron. Normal levels range from 20–300 mcg/l. People with very high iron levels have serum ferritin above 1000 mcg/l.

How common is haemochromatosis?

The genes that cause haemochromatosis are quite common — between 1 in 10 and 1 in 20 people carry one copy (the other gene being normal). These people will not develop haemochromatosis, since two copies of the abnormal gene are needed. About 1 in 400 people carry two copies. The gene change is much more common in people of Northern European descent.

What do I have to do now?

Once diagnosed with excess iron, you may need to discuss having iron removed via venesections (the opposite of a blood transfusion — blood being removed from the body). Blood contains a large amount of iron, making venesection the easiest way to reduce body iron.

It's also important to have tests (blood tests, x-rays, and possibly ultrasounds) to check whether the increased iron has affected any organs.

How often do I have to have venesections?

This depends on how high your iron levels are. Some people may initially need very frequent venesections — even weekly if levels are quite high. Once levels are reduced, regular venesections (e.g. every 3–4 months) are usually needed to keep iron levels down.

Can my blood be used at the blood bank?

The Australian Red Cross Blood Service (ARCBS) offers a venesection service for patients with haemochromatosis, and the blood collected can be used in some circumstances. Your doctor may refer you to the ARCBS venesection program, though you'll need to fulfil the criteria for regular blood donors. Your doctor will complete a referral form, and the ARCBS medical team will then contact you to arrange venesections.

Should I do anything to change my diet?

Changing your diet has only a limited effect on body iron levels. It's recommended to reduce intake of foods high in iron (especially red meat) and to drink only small amounts of alcohol, since alcohol can increase liver damage in people with haemochromatosis.

References

1. Brissot, P and de Bels, F. Current approaches to the management of haemochromatosis. Haematology, 2006:36-41.

Further Questions?

The information presented in this fact sheet is intended as a general guide only.

Patients should seek further advice and information about **Haemochromatosis** and their individual condition from their treating haematologist or doctor.

For additional information about blood disorders and their treatment, or to contact one of our specialist haematologists, visit the Melbourne Haematology website:

www.melbournehaematology.com.au