

## 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 of haemochromatosis is caused by a gene change (mutation) in the protein that control iron absorption in the intestine. The gene change is called the C282Y mutation. This mutation causes one of the proteins in the lining of the intestine to not work properly causing too much iron to be absorbed.

There is another gene change called the H63D gene change that may be associated with a small risk of developing haemochromatosis.

### How did I get haemochromatosis?

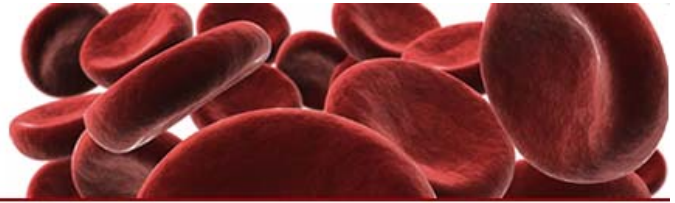
Haemochromatosis is a genetic condition – it is inherited from your parents. We all have two copies of each gene (one from our mother and one from our father). Haemochromatosis occurs when you have two copies of the abnormal gene (C282Y); you will have inherited a copy of the C282Y gene from each of your parents. Your parents may have only one copy of this gene mutation and so will not have developed haemochromatosis.

### What are the problems with having haemochromatosis?

The body needs a certain amount of iron to function properly. Iron is used by a lot of the body systems including the blood, bone marrow and brain. The amount of iron in the body is usually very closely regulated; too much iron is not good for the body. Too much iron can cause a number of body systems to not work properly including the heart, pancreas (organ in the abdomen that controls insulin production), hormone glands (pituitary gland) and the liver. Patients with too much iron in the body may develop problems in these organs. Some people with high iron levels have problems with tiredness, lack of energy, joint pains and the skin turning a darker color (hyperpigmentation).

### What are the normal levels of iron in the body?

We measure iron in the body using a blood test called serum ferritin. Ferritin is a protein in the body that carries iron around the body – a high serum ferritin is usually a sign of too much iron in the body. Normal levels of serum ferritin range from 20 – 300 mcg/l. People with very high levels of iron in the body have serum ferritin levels > 1000 mcg/l.



## How common is haemochromatosis?

The incidence of the genes that cause haemochromatosis are quite common – between 1 in 10 and 1 in 20 people will carry one copy of this gene (the other gene is normal). These people will not develop haemochromatosis because you need two copies of the abnormal gene to develop haemochromatosis. About 1 in 400 people carry two copies of the abnormal gene. The gene change is much more common in people who come from Northern Europe.

## What do I have to do now?

Once you have been diagnosed with having too much iron, you may need to discuss with your doctor having iron removed from your body using venesections (the opposite of a blood transfusion – blood being removed from your body). Blood contains a large amount of iron and venesections are the easiest way to reduce the amount of iron in the body.

It is also important to do tests (blood tests, x-rays and possibly ultrasounds) to determine if the increased iron has caused any problems in the organs known to be affected by high iron levels.

## How often do I have to have venesections?

This will depend on how high your iron levels are. Some people may initially need very frequent venesections – even every week if the iron levels are quite high. Once the iron levels are low, you may need regular venesections (e.g. every 3 – 4 months) to stop your iron levels increasing again.

## Can my blood be used at the blood bank?

The Australian Red Cross Blood Service ARCBS offers a venesections service for patients with haemochromatosis. The blood collected by the can be used in some circumstances. Your doctor may refer to the ARCBS venesections but you will need to fulfill the criteria for regular blood donors to take advantage of this service. Your doctor will need to fill out a form from the ARCBS; the medical team at ARCBS will then contact you to arrange your venesections.

## Should I do anything to change my diet?

Changing your diet will only have a limited effect on the iron levels of your body. It is recommended however that you reduce your intake of foods high in iron (especially red meat). It is also very important that you drink only small amounts of alcohol – alcohol has the capacity to increase the damage to the liver in patients with haemochromatosis.

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Resources used to produce this information sheet.

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