Iron is vital to health

Iron is an essential mineral used in many bodily processes. It is needed to form haemoglobin, a substance in the red blood cells that takes up oxygen from the lungs and releases it as the blood travels around the body. However, too much iron in the body can be damaging.

If you have haemochromatosis, your body is absorbing more iron that it needs, leading to iron overload. It is vitally important that you are diagnosed because if left untreated, the progressive build-up of iron can cause permanent damage to organs and tissues.

Where to have blood tests and venesections

Blood tests

Samples can be collected at any SNP collection centre – no prior preparation is required.

Venesections

These procedures may be performed by your doctor or the Red Cross, or you may be referred to SNP.

SNP venesection service

Before venesection can be started you will be assessed by one of our procedural doctors. An appointment is necessary. It is important you bring all notes and instructions from your doctor.

Venesection preparation: It is important that you drink plenty of water and eat as normal before a procedure.

Post-procedure care: For 24 hours after a venesection you should avoid strenuous exercise – it is possible to faint even if you are feeling well. Ideally, you should have someone drive you home.

For a list of centres that perform venesections, please go to our website: www.snp.com.au/locations/collection-centres.aspx

Locate your nearest collection centre

For a full list of collection centres and the opening hours, visit snp.com.au.

SNP collection centres locator app

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Haemochromatosis

Testing and Treatment

How the body controls iron levels

We get iron from the food we eat and the amount absorbed is determined by our body's needs. We do not have a biological mechanism for eliminating iron from the body. Instead, iron levels can only be regulated by controlling the amount taken in from the diet.

In haemochromatosis, there is a breakdown in the process that controls iron levels and the body behaves as if there is an iron shortage. It responds by absorbing more from food. This means there is more iron being taken in but no mechanism to get rid of it.

Inheriting haemochromatosis and genetic testing

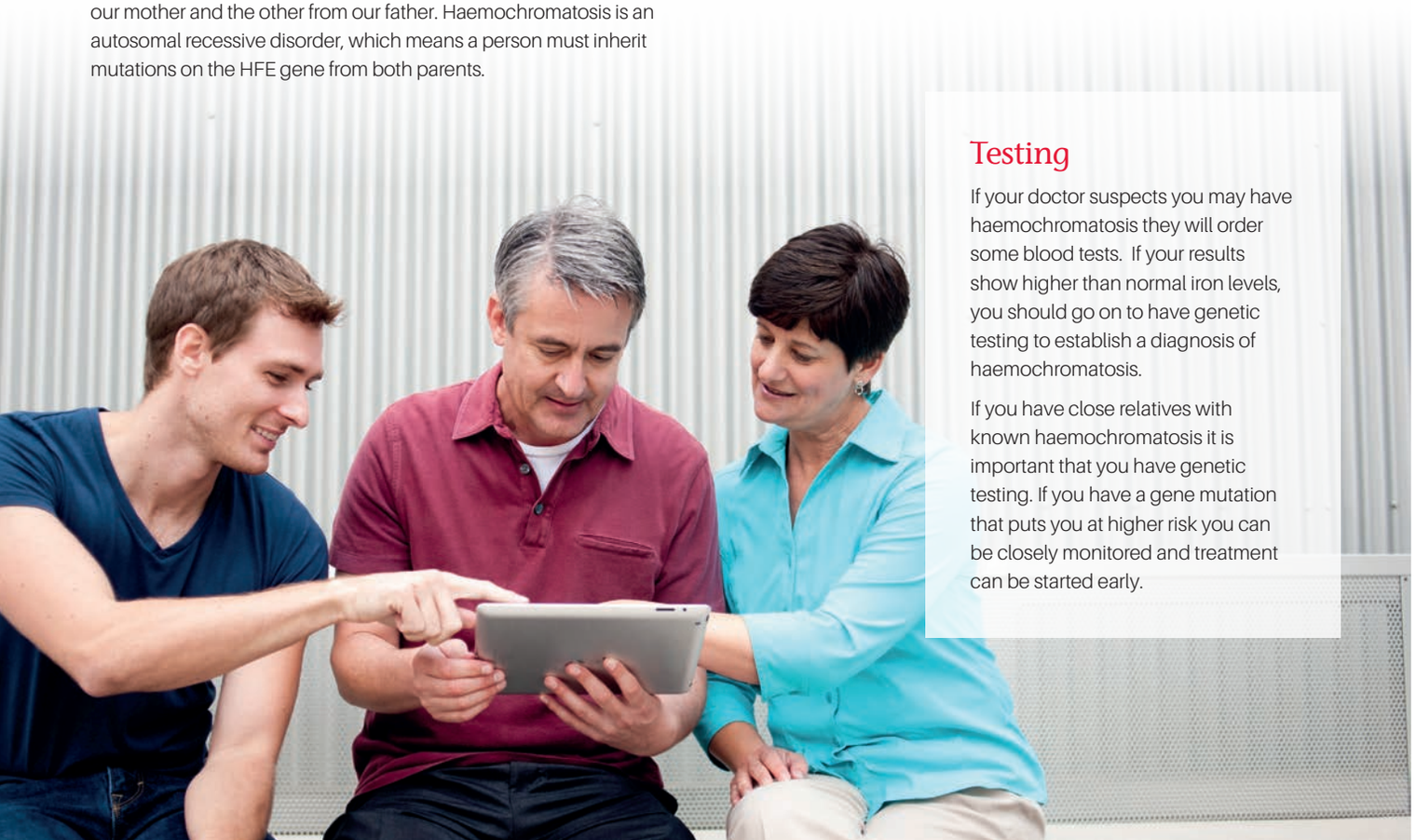
Haemochromatosis is associated with inherited gene mutations on the HFE gene, which is involved in the regulation of iron absorption. A definitive diagnosis of hereditary haemochromatosis can only be made by genetic testing.

Our genes come in pairs, one of each pair being inherited from our mother and the other from our father. Haemochromatosis is an autosomal recessive disorder, which means a person must inherit mutations on the HFE gene from both parents.

The majority of people with haemochromatosis have a mutation called C282Y on both of their HFE genes. Another mutation, H63D, is less common and associated with a lower risk of developing the condition. It is possible for someone to have a C282Y mutation on one gene and an H63D mutation on the other. There is a lower risk of developing iron overload with this combination.

If someone inherits an HFE mutation from only one parent and a normal HFE gene from the other parent, they are called a carrier. A carrier is unlikely to develop high iron levels, because they have one working gene.

Condition severity varies widely from one person to another. Someone can have mutations on each HFE gene yet not have iron overload, or only slightly elevated iron levels. Other people have elevated iron levels although they do not have the common mutations. Research into haemochromatosis continues as our understanding of the genes involved in iron regulation is incomplete and many genes are known to play a role.



Testing

If your doctor suspects you may have haemochromatosis they will order some blood tests. If your results show higher than normal iron levels, you should go on to have genetic testing to establish a diagnosis of haemochromatosis.

If you have close relatives with known haemochromatosis it is important that you have genetic testing. If you have a gene mutation that puts you at higher risk you can be closely monitored and treatment can be started early.

Measuring iron levels

Along with a Full Blood Count (FBC) and a Liver Function Test (LFT) your doctor will order a group of tests called Iron Studies. They measure blood iron levels and two proteins associated with transporting and storing iron.

Serum ferritin

Ferritin is the iron storage protein. Iron is absorbed through the gut and stored in ferritin around the body, especially in the liver and the bone marrow, where new red blood cells are made. Ferritin concentration reflects the amount of iron stored in the body.

Transferrin saturation and Total Iron Binding Capacity (TIBC)

Transferrin is the iron transport protein. Its job is to bind to iron and take it around the body. The body produces just enough transferrin according to how much iron is needed. If someone has iron overload, the body senses it and downregulates the amount of transferrin being produced. Conversely, if someone has iron deficiency, the body pumps out more transferrin.

The TIBC test measures the level of iron in a blood sample (serum iron test) and how much more iron the transferrin in that blood sample can bind. In someone with haemochromatosis, iron levels are high and the amount of transferrin is low and highly saturated.

Treatment and what to expect

Treatment for haemochromatosis is the regular drawing-off of blood, a procedure called venesection. In the process of losing blood, the body sends signals to the bone marrow to make more blood cells and in doing so, draws down on its iron stores.

Your treatment will be in two phases. The first is an intensive removal of blood that continues until body iron levels are at the lower end of normal. This may mean weekly venesections and it may take many months to unload excess stored iron.

Next is the life-long maintenance phase. You will need to have a sufficient number of venesections every year to keep your iron stores at a safe level. This varies from person to person. Women of childbearing age usually need fewer venesections because their iron levels are lowered each month through menstruation.