Handy Guide

TO HAEMOCHROMATOSIS TERMS

Haemochromatosis • UK

Saving lives and preventing ill-health

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Terms in the haemochromatosis world are often used differently, which can lead to misunderstandings – or even misdiagnosis.

This guide has simple explanations of the main words that are used. If you want more detailed info please look at some of our other guides.

Genetics

• Genetic Haemochromatosis – GH

At the charity, we describe GH as the condition you are born with if you have two 'faulty' genes associated with iron overload. This puts you at risk of developing iron overload at some point over time, but it does not mean you definitely will.

Others prefer to say you have GH only when you also have iron overload caused by the 'faulty' genes.

• Gene type

The most common gene types associated with GH diagnosis are C282Y and H63D. If you have two of these genes in any variation – two of C282Y, two of H63D or one of each then you are at risk of iron overload. The H63D gene has much lower risk of iron overload than C282Y.

GENETICS

- **Carrier**

You are a carrier if you have only one gene associated with GH. Typically, this is either C282Y or H63D.

- **Gene Test**

To find out if you have GH, you need to have a genetic test, not only a full iron panel test. Getting a genetic test before you have iron overload is an important way to prevent ill-health. Sometimes you are told you don't have haemochromatosis when they mean – your iron levels are fine. This may mean they have done a blood iron test rather than a genetic test.
Ask your GP for a print-out of your gene test results.

Cascade Screening

Cascade screening is the process of making sure that other family are genetically tested. When the first diagnosis of GH is made in the family it is important to tell parents, sisters, brothers and children (when old enough). There are NHS guidelines that recommend GPs test these relatives – but practice actually varies across the UK.

Iron Levels

Iron Overload

Iron overload is the state when someone with GH has iron levels that exceed the recommended range. Many doctors will use the term haemochromatosis to mean iron overload. This isn't wrong – but it can lead to confusion. We like to use the term 'iron overload' because you can move out of a state of iron overload with treatment, but you have the 'faulty' GH genes that risk overload for your whole life.

Serum Ferritin

Alongside Transferrin Saturation (TSat), Serum Ferritin (SF) is an important measure of iron levels in the body. SF is an indicator of your stored iron. It is not routinely checked – unlike Haemoglobin (Hb) which is another measure of iron. Sometimes you can be told your iron levels are fine – when they mean your Haemoglobin is fine.

To look for iron overload you need a full iron panel blood test. You will need to ask your GP for this.

Transferrin Saturation

Transferrin Saturation (TSat) is an important measure of iron levels in the body. TSat shows how much iron is moving around your body. It is routinely overlooked however – so you may need to ask your GP or nurse for this number each time it is tested.

Treatment

TREATMENT

Venesection or Blood Donation

Venesection is the removal of a small amount of blood regularly until iron levels are within a healthy range. Other terms for this include phlebotomy and blood-letting.

We recommend donor services are used when at all possible, so that your donated blood helps others too.

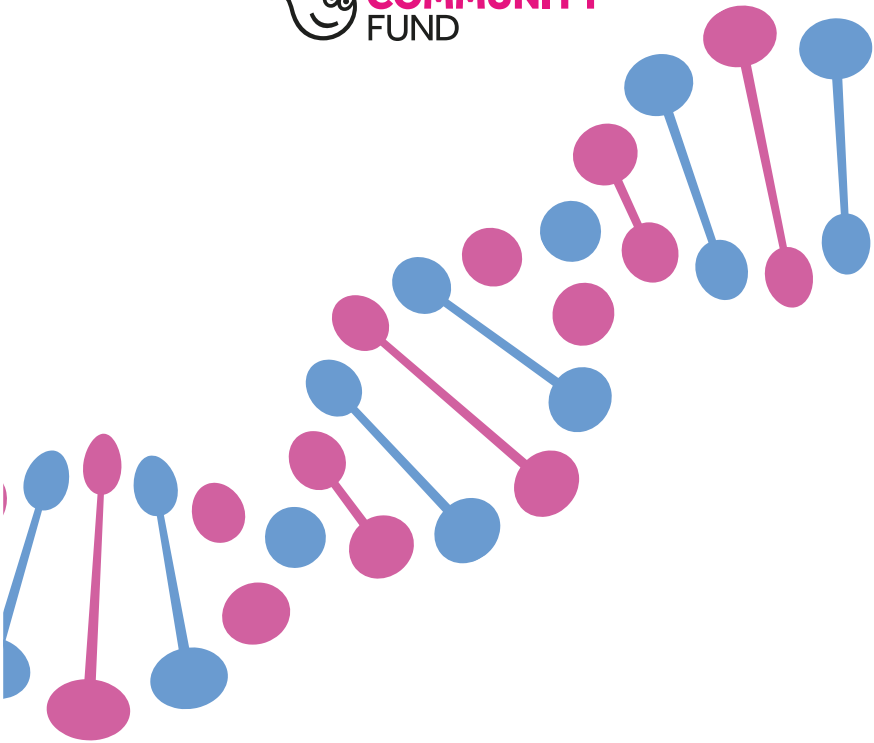
Maintenance

This is the state of keeping your iron levels within acceptable ranges – the target level can vary, unfortunately, from place to place. HUK recommends levels are kept close to target levels of under 50 Serum Ferritin (SF) and 50% Transferrin Saturation (TSat).

Referral to Specialist

When you are diagnosed with GH, your GP should refer you to a consultant. You may be referred to haematology, hepatology, rheumatology, cardiology - lets face it the list goes on! You also may need further investigations depending on levels of iron overload and symptoms.

COMMUNITY FUND



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