Hereditary Haemochromatosis: The Celtic Disease

A factsheet explaining what Haemochromatosis is, how it can be diagnosed and how it can be treated.

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What is the Celtic Disease?

The Celtic Disease is a genetic disorder and the medical term for it is Hereditary Haemochromatosis (HH). The name 'Celtic Disease' comes from the fact that it is mostly found in Celtic Nations - Ireland, Scotland, Isle of Man, Wales, Cornwall, and Brittany. It will develop only if your mother and father both carry the defective gene. People of Irish heritage are more likely than any other ethnic group to carry the gene that leads to Haemochromatosis.

In Europe, between one in 300 and one in 400 people have the potential to develop the condition. In Ireland, by contrast, one in 83 people have the two genes and are predisposed to develop Haemochromatosis. One in five Irish people are carriers of the mutated gene and one in ten people are at risk of developing iron overload. This result culminates on the last research Haemochromatosis UK undertook in Northern Ireland. Scientists are not sure why the gene is more prevalent among people with Irish heritage, but it is thought that the gene mutated 50 generations ago, about 900 AD.

Haemochromatosis is a disorder that is characterised by iron overload. Iron is essential to all plants, animals, and human life. Normally, our body controls the amount we need to absorb to remain healthy and secretes the rest. If you have the mutated HH gene or have the condition, your body may store too much iron which is deposited in other vital organs such as the heart, liver, and pancreas. This affects the ability of the organ to function effectively. This absorption and storage of iron happens over several years, which can then be described as 'iron overload'.

Excess iron (caused by iron overload) is extremely toxic and can affect many parts of the body. Genetic Haemochromatosis (GH) affects everyone differently. Many people experience one or more symptoms. But some people don't experience any noticeable symptoms as a result of genetic Haemochromatosis.

How do I know if I have iron overload or HH?

As this is a genetic disease you are born with it. Everyone receives two sets of genes – one from their father and one from their mother. You're only at risk of Haemochromatosis if you inherit the faulty HFE gene from both of your parents. If you only inherit the faulty gene from one parent, you'll be at risk of passing it on to your children – known as being a "carrier" – but you will not develop Haemochromatosis yourself.

The symptoms of HH don't become apparent until middle age. It is therefore difficult to know without getting a genetic test done. The symptoms can have other causes in origin, which is why so few people get tested for the condition and get diagnosed with Genetic Haemochromatosis. The liver problems caused by Haemochromatosis may be wrongly attributed to excessive alcohol consumption and treatment may not be offered. We have examples of Irish men in particular not receiving treatment because the symptoms of Haemochromatosis have been wrongly diagnosed as alcohol related.

Symptoms of genetic Haemochromatosis can include the following:

- Arthritis; may affect any joint but particularly common in the knuckle and first joint of the first two fingers (the bronze fist). If arthritis is found only in the first two finger joints this is highly suggestive of GH
- Chronic fatigue, weakness, lethargy
- Abdominal pain; sometimes in the stomach region or the upper right-hand side, sometimes diffuse
- Neurological/psychiatric disorders; impaired memory, mood swings, irritability, depression
- Sexual disorders; loss of sex drive, impotence in men
- Absent or scanty menstrual periods and early menopause in women
- Bronzing of the skin, or a permanent tan, or grey tone
- Cardiomyopathy; disease of the heart muscle
- Diabetes (late-onset type)
- Pituitary or adrenal issues (eg Addison's Disease)
- Liver disorders: abnormal liver function, enlarged liver, cirrhosis, liver cancer
- Decrease in body hair

As you can see from these symptoms it is difficult to link it directly to HH as there can be many different reasons you have the above symptoms. However, if you do have one or more of these symptoms and are of Celtic origin you should consider bringing this leaflet to your GP and discuss possible testing for HH.

What should you do if you are worried?

Haemochromatosis is difficult to diagnose but it is important that the condition is diagnosed early. If the condition is treated before excessive build-up of iron occurs, life expectancy should not be affected. So, it is important that if you are worried you should go and see your GP as soon as possible and explain the symptoms to them. It is likely you will then be referred to a heart or liver specialist. Haemochromatosis is usually diagnosed through blood tests. If a member of your family is diagnosed with primary Haemochromatosis, the genetic condition, it is likely that you will need to be tested too.

What is the test?

The condition is diagnosed through blood tests, which can be arranged by your GP. The tests come in two stages.

Stage One – initial blood tests

The first stage is to do a simple blood test for serum ferritin, transferrin-saturation, full blood count and liver function tests. This will give an indication if you have too much iron in your body.

If your initial blood test results are outside normal range, you should then be referred for a subsequent genetic test.

The normal ranges for these test results vary for men and women.

Men should be referred for a genetic test if their full blood count (FBC) is normal and serum ferritin (known as SF) is over 300µg/l and/or transferrin saturation (known as random TSAT) is over 50 percent.

Women should be referred for a genetic test if their full blood count (FBC) is normal and Serum ferritin (known as SF) is over 200µg/l and/or transferrin saturation (known as random TSAT) is over 40 percent.

Stage Two – genetic test

If your GP discovers that your initial blood tests are outside the normal range, they should arrange a genetic test. This will check for the common gene variants of genetic Haemochromatosis (known as c282y and H63d). The genetic test will show if you have genetic Haemochromatosis or not.

For a genetic test you will be asked to give a small blood sample. You will receive the results of this test within four to six weeks. If the test confirms that you have genetic Haemochromatosis, either your GP or a genetic counsellor will contact you to explain the results.

There are three common variations of genetic Haemochromatosis:

- C282y/c282y (also known as “c282y homozygous”). This is the most common variant of the condition in the UK, affecting over 9 in every 10 people diagnosed. People with this variation tend to load iron much more quickly compared to other people with genetic haemochromatosis.
- C282y/H63d (also known as “compound heterozygous”). This is a less common variant of the condition, affecting up to five in every 100 people diagnosed. People who are compound heterozygous tend to load iron less rapidly than other people with genetic haemochromatosis.
- H63d/H63d (also known as “H63d or non-c282y homozygous”). This is a less common variant of the condition, affecting up to five in every 100 people diagnosed. People who are H63d homozygous tend to load iron less rapidly than other people with genetic haemochromatosis.

Home genetic testing kits

The charity, Haemochromatosis UK, has introduced an at-home genetic testing service for members and families, for just £39 per test. This service is subsidised by the charity as a preventative health initiative. It is available at this discounted rate as a benefit of membership to HUK members who have been members for 3 months or more. The service is also available to new members and non-members for £119. Click here to order.

Can Hereditary Haemochromatosis be treated?

There's currently no cure for Haemochromatosis, but there are treatments that can reduce the amount of iron in the body and reduce the risk of damage. With treatment, many people live full and active lives with genetic Haemochromatosis.

There are two main treatments:

- Phlebotomy – a procedure to remove some of your blood; this may need to be done every week at first and can continue to be needed two to four times a year for the rest of your life. This is the most effective treatment.
- Chelation Therapy – where you take medicine to reduce the amount of iron in your body; this is only used if it's not easy to regularly remove some of your blood

The aim of the treatment is simply to reduce the stored iron in your body tissue. The treatment is the same as giving a blood donation. You may have to attend weekly or twice weekly sessions with your GP or nurse where they take a certain amount of blood from your body. If the condition is detected early before any organ is damaged, then you will have a normal life expectancy. However, if there is damage to the organs then the treatment can prevent further damage.

You do not need to make any big changes to your diet to control your iron levels if you're having treatment, but you'll usually be advised to avoid:

- breakfast cereals containing added iron
- iron or vitamin C supplements
- drinking too much alcohol

What happens if I don't get tested?

The continuous build up and storage of iron in the body can become toxic to your organs. If untreated it can cause serious health issues affecting vital organs such as your heart and liver and can cause diabetes and arthritis. However, if detected early the symptoms described above can be alleviated and, in some cases, resolved.

Where to get help

Irish in Britain is not a medical centre and the information supplied to you on this leaflet has been collated from research and from various Haemochromatosis Associations in the UK and Ireland. For detailed information and medical references, we recommend you visit the following websites or call the helplines.

- Haemochromatosis UK:

https://www.haemochromatosis.org.uk/

Telephone Advice Line: 03030 401 102 (weekdays 12-3pm)

Email Advice Line helpline@huk.org.uk

- The British Liver Trust

https://britishlivertrust.org.uk/

Irish in Britain

Email: health@irishinbritain.org

Twitter: @irishinbritain

Facebook: IrishinBritain

Irishinbritain.org