

About Haemochromatosis:

- Haemochromatosis is a genetic disorder that causes the body to absorb too much iron from the diet.
- Symptoms of haemochromatosis can include chronic fatigue, liver damage, diabetes, abdominal pain, heart problems, joint pain and 'bronzing' of the skin.
- Haemochromatosis can be treated by regular removal of blood, a procedure known as venesection therapy or phlebotomy. Patients are also advised to avoid too much iron in their diets.
- Haemochromatosis is caused by an inherited genetic change, called a mutation. People who inherit two copies of this mutation (one from each parent) are at risk of iron overload. People who inherit just one copy are healthy 'carriers' of the condition.
- Haemochromatosis can be diagnosed by measuring the iron levels in the blood (serum ferritin levels), and confirmed with a genetic test. A genetic test can also identify family members who may be at risk of storing excess iron, but do not yet have any symptoms.

Find out more

To find out more about Haemochromatosis, and for information and support contact:

UK Haemochromatosis Society
020 8449 1363
www.ghsoc.org



The Haemochromatosis Society

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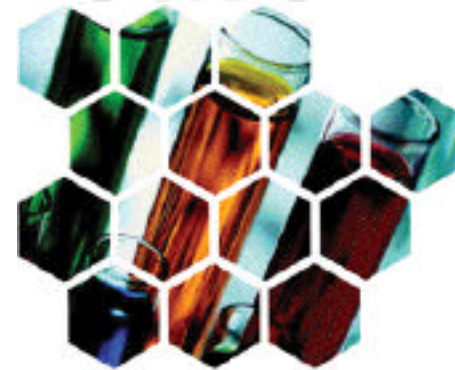
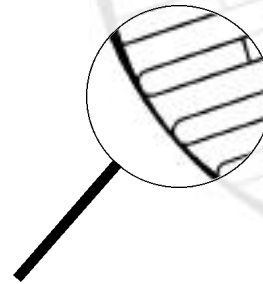


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Getting a diagnosis of...



Haemochromatosis



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Haemochromatosis

Who should have a test for Haemochromatosis?

Genetic or hereditary haemochromatosis is mainly associated with a defect in a gene that codes for a protein called HFE. HFE regulates the body's ability to absorb iron from food. Genetic testing is useful to assess the risk to members of a family once an individual in the family has been diagnosed with haemochromatosis.

There are two known important changes (mutations) in the HFE gene known to be associated with haemochromatosis. It cannot currently be predicted in advance who will develop significant clinical symptoms. Almost 90% of individuals with haemochromatosis inherit a HFE gene carrying this mutation from both parents, ie: they have two copies of the gene carrying a mutation. However, two copies of this gene alone do not predict with certainty that a person will develop haemochromatosis: only 90% of males and 50-70% of females who carry two copies of this mutation will actually go on to develop haemochromatosis. In other words, when two HFE mutations are identified in someone who has no symptoms of the condition, it is not currently possible to predict whether a person will go on to develop symptoms.

Additionally, there is a second mutation that is associated with only a slightly increased risk of haemochromatosis. About 10% of patients with haemochromatosis will have one or two other mutations. A small number of specialist research laboratories will search for other changes in the HFE gene by a process called DNA sequencing, which examines the sequence of letters in the entire DNA code for the gene. Other forms of haemochromatosis are known to exist that are not associated with the HFE gene.



Christine and Julia (one of her five children) live in North London, where Julia runs her own business. In 1996, both found out they had haemochromatosis, after Christine's brother Leonard was diagnosed with the condition.

Christine: *'I'm the eldest of three – all three of us were affected. In 1995 my brother went to the doctor complaining he was unwell.'*

Leonard was initially diagnosed with liver damage and diabetes, and later bladder cancer. But a specialist doctor found out that he also had haemochromatosis, which meant that his body stored too much iron. Sadly, Leonard's health problems were picked up too late for effective treatment, and he died of liver cancer in 1997.

Christine: *'Leonard is considered to be the martyr of the family, because if he hadn't died none of us would have known what to do about it at all.'*

Following his diagnosis, the rest of the family were tested and Christine, her younger brother Robert and two of Christine's children were found to have haemochromatosis too.

'They were tested once I was found to be positive – they were all adults, of course. Richard is the eldest, he is positive. He's been tested recently to see if he's still retaining any iron in his liver and he isn't, so they've cleared him completely. Then there's Julia who is positive, and has to go for venesection.'

Venesection treatment simply involves taking blood regularly, which prevents excess iron building up in the body. Christine had venesection therapy every week for a short while, until her iron levels (measured as her 'serum ferritin count') dropped to safe levels. Julia, her daughter, began treatment after she was found to have a ferritin count higher than usual:

Julia: *'I was going once every 6 weeks-2 months to start off with, to get bled down, but because I wasn't very high it wasn't a long process. Soon I was just put on to the '3 monthly maintenance'. It's a nuisance to just have to keep going and giving a pint of blood, basically. But obviously a vital nuisance!'*

Julia and Christine both stress the importance of having their condition diagnosed early, enabling them to have treatment that has prevented any symptoms appearing:

Julia: *'Extremely helpful, having seen a family member diagnosed with it when it's too late, and within two years die from it. Effectively, it's been caught early, because my first ferritin count was only the high end of normal. Therefore in theory, no damage has been done to me whatsoever by it.'*

As well as regular blood-taking, Julia and Christine also avoid eating iron-rich food like liver and spinach:

Julia: *'I have to watch what cereal I eat because of the added iron - not drinking so much vitamin C fruit juices is about the only thing we've actually changed, because we've never been big red meat eaters anyway. But no strict instructions, it was just awareness - we weren't so at risk as others anyway, because of the fact we were caught early'*