Genetic Haemochromatosis

An Iron Overload Disorder

The Haemochromatosis Society

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Genetic Haemochromatosis (Iron Overload Disorder)

Genetic Haemochromatosis (GH) is an inherited disorder which causes the body to absorb too much iron from the diet. Excess iron gradually accumulates, usually in the liver, joints, heart, pancreas and other endocrine glands, causing serious tissue damage.

GH is one of the most common genetic disorders. Surveys have shown that as many as 1 in 200 people could be at risk of developing iron overload.

Simple and effective treatment is available, but if excess iron is not removed irreversible damage can occur, especially in the liver.

Diagnosis can be confirmed by a simple blood test and/or genetic test. Early diagnosis and treatment preserves normal life expectancy and quality of life

This leaflet tells you about GH and about The Haemochromatosis Society, your patient organisation and a valuable source of information and support.

Disclaimer: This leaflet has been approved by our medical advisers but is not a substitute for professional guidance. Please speak to your doctor if you need more information or are in any way concerned about your health or symptoms.

Symptoms

Symptoms develop as iron slowly builds up, so are more likely to occur during adult life. Some symptoms can also be caused by other illnesses, but a combination of two or more indicates that GH is possible.

Arthritis: Any joint, but particularly in the hands and feet
Fatigue: Chronic fatigue, lethargy, being continually tired or weak
Heart problems: Irregular heartbeat, shortness of breath and cardiomyopathy
Depression: Also mood swings, irritability and impaired memory
Sexual Problems: Impotence, loss of libido and early menopause
Skin colouration: Tanning (bronzing), greying or paling of the skin
Abdominal pain: In the stomach area or upper right, and often diffuse
Hair loss: From head and/or body
Liver disorders: Enlargement, cirrhosis and hepatoma (liver cancer)
Menstrual problems: Scanty, irregular or missing periods
Diabetes: GH is a known cause of late onset diabetes mellitus

A combination of symptoms, especially tiredness and joint pain, should be investigated for iron overload

Diagnosis

Diagnosis is through blood tests. Transferrin Saturation (TS) indicates how much iron is available for use in the body. The average is 30% (slightly higher in men than women). If on two occasions TS is over 50% in men (45% in women) GH is very likely. TS is the most specific and sensitive test for haemochromatosis. Serum Ferritin (SF) indicates the amount of iron stored in the body. Levels significantly over 300 micrograms/litre in men and post-menopausal women (200 in younger women) suggest GH. However in the early stages of iron accumulation SF may remain low.

Genetic Testing is positive in 95% of people with GH. A genetic test can also identify other family members at risk of loading iron before they actually do so.

A **liver biopsy** may be recommended if serum ferritin exceeds a certain limit (currently 1000 micrograms/litre as at January 2016) or if other tests suggest the liver is damaged. In any case, your consultant will guide you through this process.

Treatment

A referral to a haematologist (blood specialist) or to a gastroenterologist (digestive system specialist) is usually the next step. Treatment is simple and effective, and usually consists of regular removal of blood, known as *venesection* or *phlebotomy.* The procedure is the same as for blood donors and each pint of blood removed contains about a quarter of a gram of iron. The body then uses some of the excess stored iron to make new red blood cells.

Initially venesection may be performed once a week, and continues until the iron level is satisfactory, which may be a year or more. During the therapy iron storage is carefully monitored and treatment continues until the serum ferritin level reaches 50 micrograms/litre. Note this this is the current guideline figure (as at January 2016) but this should be discussed with your consultant on an individual basis.

After iron stores have been depleted a *maintenance phase* of treatment is usually required; iron levels are monitored regularly, and if necessary occasional venesections completed.

Venesection reduces the body's stores of iron to normal, and many symptoms will improve. Providing treatment begins before cirrhosis has developed life expectancy will be normal. However venesection will not cure some serious conditions caused by iron overload such as arthritis, diabetes or cirrhosis if they are already present.

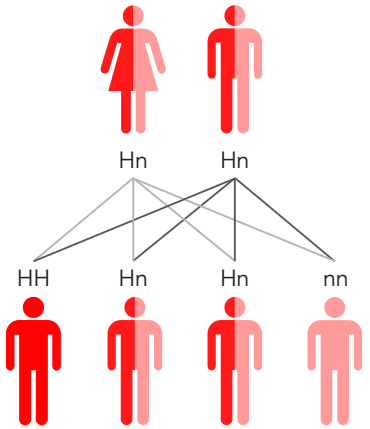
Early diagnosis and treatment is vital.

Genetics

To develop GH a defective gene is inherited from each parent. People who inherit a defective gene from one parent are carriers and are at less risk of iron overload (though it is not unknown).

For example, if both parents are carriers, children will have a 50% chance of being carriers and a 25% chance of being at risk of developing GH. It is estimated that up to 1 in 8 people in some areas of the UK may be carriers.

Inheritance when both parents are carriers



Key: H = haemochromatosis gene. n= 'normal' healthy gene.

Your relatives should also ask about testing. Discuss this with your doctor.

Diet

One of the most frequent areas of concern and discussion for people affected by haemochromatosis is diet, with understandable worries over the number of foodstuffs that include added iron. Some sensible but proportionate changes can help.

Changes to diet cannot prevent iron overload; many foods contain iron and a balanced diet is essential. The amount of iron absorbed from food is small compared with that removed by venesection. However, iron absorption can be slowed by:

- avoiding supplements and tonics containing iron and cereals fortified with iron
- avoiding large doses of vitamin C, which increases absorption of iron and hastens its deposition in some organs.
- limiting alcohol intake, particularly with meals, as it increases iron absorption and can also cause liver disease.
- taking tea and dairy products with meals, which reduces the rate of absorption.
- cutting down on red meat; iron is much more readily absorbed from meat than vegetables, fruit, cereals and beans.

More advice about diet and other lifestyle issues can be found on The Haemochromatosis Society website and in The Haemochromatosis Handbook which is distributed to members.

The Haemochromatosis Society

The society provides information and support for those affected by GH. Members receive treatment record cards, a handbook, regular newsletters and opportunities to meet each other and haemochromatosis specialists. We also provide the two GH advice lines.

PLEASE JOIN US. Your membership helps to make us stronger as a patient organisation and helps us to provide better services for people with haemochromatosis. Join us online or use the tear-off membership form at the back of this booklet.

The society was established to SUPPORT people with GH and their families, to promote AWARENESS amongst healthcare professionals, patients and their families so that GH can be diagnosed and treated as soon as possible, and to encourage and support RESEARCH.

The society is a registered charity and a member of the European Federation of Associations of Patients with Haemochromatosis (EFAPH)

The society is your patient organisation and provides information and support. Please join us

Information

Medical Advisors

Prof TM Cox University of Cambridge
Dr A Critchley Leeds Teaching Hospital
Dr D Das (retired) Stepping Hill Hospital, Stockport
Dr JS Dooley Royal Free Hospital, London
Dr E Fitzsimons Gartnaval Hospital, Glasgow
Dr P Kiely St George's Hospital, London
Prof R Williams Institute of Hepatology, London

Scientific Advisors

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Member Registration

Title ..... Full Name .....

Address .....

.....

Email .....

Daytime Phone ..... Mobile Phone .....

Date of Birth ..... Gender .....

Would you be happy to answer a short telephone questionnaire about your experience of haemochromatosis?

Yes ◯ No ◯

I am a UK taxpayer and would like any subscriptions/donations to be treated as qualifying for Gift Aid until further notice. I have paid an amount of income tax or capital gains tax equal to the tax reclaimed.

Yes ◯ No ◯ Unsure ◯ Prefer not to say ◯

How would you like to receive your newsletter?

By post ◯ Large print by post ◯ By email ◯ Newsletter not required ◯

Would you like to receive the following?

Monthly e-News Bulletin ◯ Volunteer Alerts by email ◯

Money box to collect your change for the charity ◯

How did you hear about the society? .....

The society is a membership organisation. Our constitution gives members voting rights at our AGM. The charity is a Company Limited by Guarantee; by joining you also agree to contribute the sum of £1 if the charity is ever wound up. See overleaf for subscription fees.

Sign ..... Date .....

The Haemochromatosis Society

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Name and full postal address of your Bank or Building Society.

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Instructions to your Bank or Building

Please pay The Haemochromatosis Society Direct Debits from the account detailed in this Instruction subject to the safeguards assured by The Direct Debit Guarantee. I understand that this Instruction may remain with the society and, if so, details will be passed electronically to my Bank/Building Society

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◯ £24 standard annual subscription ◯ £12 concessionary rate (benefits or state pension income only)
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Please return this form together with the membership registration overleaf to:
The Haemochromatosis Society, PO Box 6356, Rugby, CV21 9PA

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