King's College Hospital
NHS Foundation Trust

Genetic haemochromatosis (GH)

Information for patients

This information leaflet is for patients who have genetic haemochromatosis. It provides key information about the disorder, treatment and long-term management of the condition as well as key contacts and information resources.

Confirming your identity

Before you have a treatment or procedure, our staff will ask you your **name** and **date of birth** and check your **ID band**. If you don't have an ID band we will also ask you to confirm your address.

If we don't ask these questions, then please ask us to check. Ensuring your safety is our primary concern.

What is genetic haemochromatosis?

Genetic haemochromatosis (GH) is the most common genetic disorder in the Caucasian population. It is most commonly found in Northern Europeans of Celtic and Nordic descent.

GH causes the body to continue to absorb iron from the gut even when iron levels are normal or high, leading to increased iron deposition in the body (iron overload). Over time, iron overload can result in inflammation and damage to organs. This can result in arthritis, diabetes, heart disease and, in serious cases, liver cirrhosis and cancer.

GH is also referred to as hereditary haemochromatosis (HH) and familial haemochromatosis (FH). In this leaflet it will be referred to as genetic haemochromatosis (GH).

What causes genetic haemochromatosis?

GH is a genetically inherited autosomal recessive condition. Two mutated genes called high ferritin expression (HFE) need to be inherited (one from each parent) for you to have the disorder.

What are the symptoms of genetic haemochromatosis?

Iron overload is the main clinical feature in GH. There are three main stages:

1. predisposition – you have two copies of an HFE gene mutation, but no evidence of iron overload
2. iron overload is present but there is no evidence of organ damage
3. iron overload with organ damage

GH symptoms present in patients differently. Usually, symptoms will present slowly and are more likely to manifest in adulthood.

Some symptoms are more severe and may be associated with a later diagnosis or more severe iron overload.

Symptoms that may be associated with GH include:

- tiredness or feeling weak frequently
- abdominal pain
- changes to skin colour – skin appears 'bronze' or tanned or pale in colour
- hair loss from the head and/or body
- arthritis – this can be in any joint but typically affects the knuckles (second and third metacarpophalangeal joints) which can lead to a painful handshake
- loss of libido or impotence
- neurological or psychiatric disorders including dementia, impaired memory, mood swings, irritability, or depression
- diabetes
- menstrual irregularity
- cardiac issues that may result in irregular heartbeats, shortness of breath and cardiomyopathy
- liver disorders that may result in abnormal liver function, enlarged liver, cirrhosis, or liver cancer

Investigations

Genetic testing is required to diagnose GH. This involves testing for mutations in the HFE gene. There are two common mutations of the HFE gene at amino acid 282 (commoner) and 63.

Not all patients who inherit two copies of a genetic mutation will develop the disease.

Investigation of confirmed or suspected GH includes full blood count (FBC), liver function tests (LFT), ferritin and transferrin saturation.

Iron overload is characterised by high ferritin (greater than 300ug/l and 200 ug/l in males and females respectively) with transferrin saturation (Tsat) of 50% and 40% in males and females respectively.

As GH is a genetic condition, screening of family members is recommended. Family screening should include parents, siblings, partner, and children (over the age of consent). This can be done via their GP.

Consent

We must by law obtain your written consent to any operation and some other procedures beforehand. Staff will explain the risks, benefits and alternatives before they ask you to sign the consent form. If you are unsure about any aspect of the procedure or treatment proposed, please do not hesitate to speak with a senior member of staff again.

Treatment

Venesection (or bloodletting) is the most effective treatment for GH. Venesection helps to reduce excess iron stores through repeated blood removal. By regularly removing units of blood, the body uses its excess iron stores until the ferritin level returns to normal.

The standard amount for each venesection is 500ml of blood which contains around 200 to 250mg of iron.

There are two stages to venesection treatment.

1. **Iron removal phase (de-ironing)** – venesection is undertaken every week or two until the ferritin level is below 50ug/l and the transferrin saturation is less than 50% (as long at this does not cause symptomatic anaemia). **Individual ferritin and transferrin saturation thresholds may be agreed for patients**.

2. **Maintenance phase** (once the ferritin level is 50ug/l or below) – approximately 2 to 4 venesections are required a year. Venesection may be replaced by blood donation through the NHS Blood Donation service.

You may also require other treatments to manage symptoms related to organ-specific damage.

Dietary advice

You should aim to have a healthy balanced diet. People with GH are often worried about the amount of iron in their food. GH cannot be treated through diet alone. A restricted diet is not recommended as iron is an essential part of a balanced diet and diet alone cannot stop iron overload.

Iron is essential for:

1. the formation of haemoglobin – a protein in red blood cells that carries oxygen around the body
2. the production of myoglobin – a protein in muscle tissue, used for storage of oxygen
3. regulating normal energy metabolism
4. DNA synthesis and repair
5. supporting the immune system
6. brain development

There are two main sources of dietary iron. These are haem iron and non-haem iron.

Type of iron	Source
Haem iron	Meat, chicken and fish
Non-haem iron	Vegetable, cereals, beans and lentils

The body finds haem iron easier to absorb than non-haem iron. When choosing foods to eat, choices of foods that contain non-haem iron can help reduce the overall amount of iron absorbed each day. People with GH can still suffer from iron deficiency if there is not enough iron in the diet.

There are foods that also contain inhibitors of iron (such as tannins, oxalates, and phytates). These can reduce the amount of iron absorbed from meals – particularly non-haem iron. Consuming inhibitors with each meal can help to reduce iron absorption.

Inhibitor	Dietary source
Tannins	Tea, coffee, wine
Oxalates	Leafy greens, rhubarb, strawberries, beetroot, parsley, almonds, legumes, soy products, potatoes, dates
Polyphenols	Chocolate, walnuts, onions, apples
Phytates	Sesame seeds, lentils, wholegrain cereal

There are some foods and supplements that can increase the rate of iron absorption. This includes vitamin C (ascorbic acid) and multivitamins which contain iron. People with GH should avoid taking vitamin C supplements and avoid orange juice with meals. It is still ok to eat daily fresh fruit and vegetables.

Alcohol can also increase iron absorption. Patients who have liver damage secondary to GH are advised to reduce alcohol intake or abstain altogether.

Who can I contact for advice?

If you or your carers have any questions about your condition or any information in this leaflet, please do not hesitate to ask the nurses

in the Haematology Supportive Care Unit for advice. They can refer you to the appropriate person to discuss your queries with.

Patients with GH are cared for in different locations across the Trust sites.

Clinicians/nursing team: Please complete the box below with the relevant contact details based on the location of treatment:

Further information and resources can be found on the Haemochromatosis UK website
www.haemochromatosis.org.uk

Sharing your information

We have teamed up with Guy's and St Thomas' Hospitals in a partnership known as King's Health Partners Academic Health Sciences Centre. We are working together to give our patients the best possible care, so you might find we invite you for appointments at Guy's or St Thomas'. To make sure everyone you meet always has the most up-to-date information about your health, we may share information about you between the hospitals.

Care provided by students

We provide clinical training where our students get practical experience by treating patients. Please tell your doctor or nurse if you do not want students to be involved in your care. Your treatment will not be affected by your decision.

PALS

The Patient Advice and Liaison Service (PALS) is a service that offers support, information and assistance to patients, relatives and visitors. They can also provide help and advice if you have a concern or complaint that staff have not been able to resolve for you. They can also pass on praise or thanks to our teams.

PALS at King's College Hospital, Denmark Hill, London SE5 9RS
Tel: **020 3299 3601**
Email: **kch-tr.palsdh@nhs.net**

PALS at Princess Royal University Hospital, Farnborough Common, Orpington, Kent BR6 8ND
Tel: **01689 863252**
Email: **kch-tr.palspruh@nhs.net**

If you would like the information in this leaflet in a different language or format, please contact our Communications and Interpreting telephone line on 020 3299 4826 or email kch-tr.accessibility@nhs.net

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