Patient information

Genetic Haemochromatosis

Digestive Diseases Care Group

Genetic Haemochromatosis

Haemochromatosis is an inherited (genetic) disorder causing the body to absorb too much iron from the diet. The excess iron is then stored in various organs, mainly the liver. The excess iron may also be stored in the pancreas, heart, testicles/ovaries, skin and joints. The main treatment is the regular removal of blood, which helps to remove the excess iron from the body. If treatment is started early enough and before complications occur then the outlook for people with haemochromatosis is very good.

How is it inherited and how common is it?

Haemochromatosis is a genetic condition affecting genes involved with iron storage. In nine out of ten people with haemochromatosis the abnormal 'HFE' gene is the cause of the condition.

Haemochromatosis is a 'recessive' disorder. This means that haemochromatosis will only occur if both copies of the gene are abnormal. If only one copy is defective, a person will be perfectly healthy but will be a 'carrier'. This means their children could inherit the defective gene.

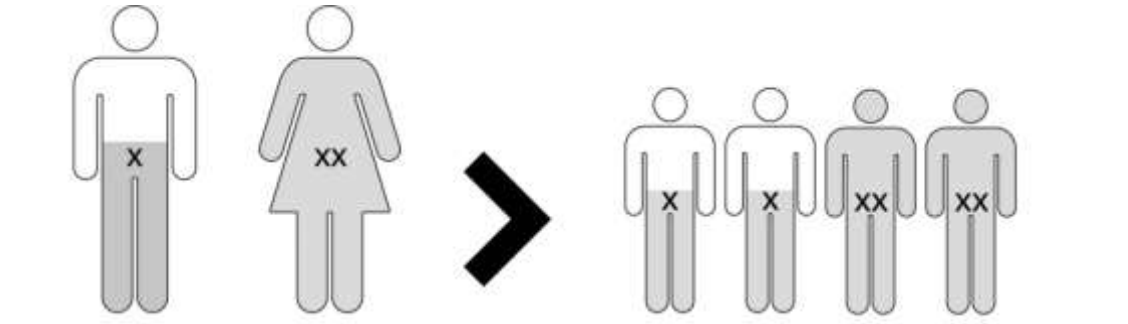
Genetics

1. If both parents are carriers

About 12.5% of the population are carriers, so 1.6% of partnerships will be between carriers;

- ¼ will then develop GH
- ¼ will be normal
- ½ will be carriers

2. One parent has GH and the other is a carrier



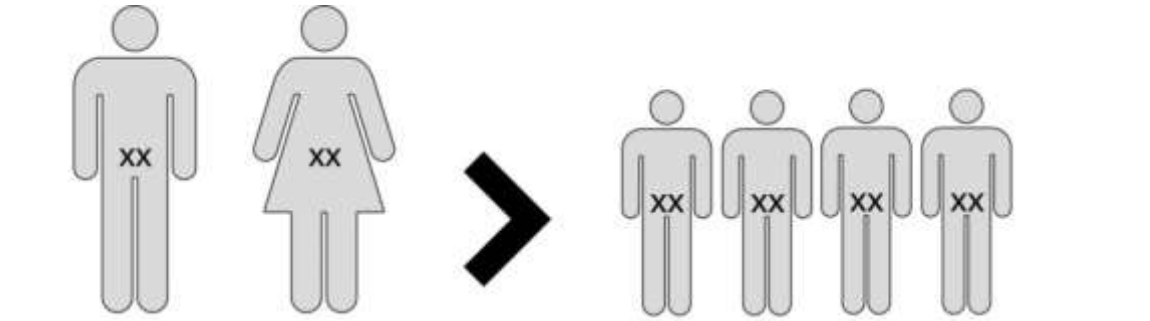
Father – Heterozygous carrier
Mother – Homozygous for C282Y

50% chance – Heterozygous carrier
5-% chance – Homozygous for C282Y

This occurs in about 1 in 2000 partnerships. On average

- ½ the children will develop GH
- ½ the children will be carriers

3. If both parents have GH



Father – Homozygous for C282Y
Mother – Homozygous for C282Y

100% chance – Homozygous for C282Y

- This is rare occurring in about 1 in 100,000 partnerships.
- In this situation all children will inherit two defective genes and all children will have GH

How does it present?

Not all patients experience symptoms however some do and they usually start between the ages of 30 and 50 years. The first symptoms are usually vague and may include feeling weak and tired, pain in the joints and pain in the tummy. As haemochromatosis progresses, more specific symptoms develop but these are now much less common because of earlier diagnosis.

There problems include:

- diabetes
- increase in the size of the liver
- 'scarring' (cirrhosis) of the liver,
- bronzing of the skin (like a permanent tan),
- disease of the heart muscle (cardiomyopathy)

- joint problems, especially the knuckle and the first joint of the first two fingers
- poor memory
- feeling irritable
- depression.

Advanced haemochromatosis may cause loss of sex drive and less body hair. Impotence may occur in men. Women may have either no menstrual periods or very light menstrual periods. Early menopause may also occur in women with haemochromatosis. Most of these symptoms are found in other disorders and so diagnosis can be difficult. Arthritis found only in the knuckle and the first joint of the first two fingers is very suggestive of haemochromatosis.

The need for treatment to remove excess iron does not depend on the presence of symptoms. Because of the risk of developing a serious complication such as cirrhosis, treatment to remove excess iron from the body is very important even if there are no symptoms.

How is it diagnosed?

Haemochromatosis is usually diagnosed on blood tests (ferritin and transferritin saturation), if both of these are raised the diagnosis is confirmed with a gene test for the HFE gene. A scan of the liver may be used to assess the extent of liver disease.

Who should be tested?

Anyone with symptoms indicating possible haemochromatosis should be tested for the level of iron in the body. The gene test should be offered if the iron level is high.

Brothers, sisters and children of anyone who has haemochromatosis should be tested for the abnormal gene.

How is it treated?

The effective treatment consists of regular removal of blood, which is also known as venesection. Venesection may need to be done frequently (weekly) at first, depending on the level of iron overload in the body. Once the ferritin and transferrin levels are within normal range, you will only require venesection every three to six months. The procedure is likened to giving blood for the blood transfusion service. Venesection is done as a day case in the hospital and takes approximately 30 minutes. You will be advised to drink plenty of fluids in the 24hours before venesection and to make sure you have eaten normal meals.

The procedure will take place on a comfortable chair. A nurse will then insert a needle attached to a blood bag into the vein and drain approximately 500mls of blood. After venesection the needle will carefully be removed from your arm and a dressing will be applied.

Some people feel faint (light-headed or dizzy, hot, sweating, trembling or shaky) All the staff are trained to care for you and deal with this if it should happen.

Regular blood removal will not cure some of the complications of haemochromatosis such as diabetes or liver 'scarring' (cirrhosis). Therefore, early diagnosis and treatment are very important.

What about diet

The increased levels of iron in the body cannot be treated by diet alone. Removing blood has a much bigger effect on reducing the levels of iron in the body.

However, there are some recommendations:

- Avoid vitamin supplements or tonics containing iron; avoid breakfast cereals heavily fortified with iron.
- Large doses of vitamin C should also be avoided because it increases the amount of iron absorbed from food eaten. Vitamin C also increases the amount of iron stored in the body.
- Reduce intake of offal (eg, liver and kidney) and red meat.
- Reduce alcohol intake, especially with meals, as it may increase iron absorption and it can also cause liver disease.
- Tea and all milk products taken with a meal reduce the amount of iron absorbed from food.

What are the complications?

The possible complications of haemochromatosis include:

- diabetes
- heart disease
- liver 'scarring' (cirrhosis)
    - People with haemochromatosis who develop cirrhosis are also at increased risk of liver cancer and should be checked regularly with ultrasound scans or MRI scans.

What is the outlook (prognosis)?

If haemochromatosis is diagnosed and treated early before any complications develop the outlook is very good with no reduction in life expectancy. If complications do occur then the prognosis may be much worse.

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Further information

Hepatology Specialist Nurses
Tel: 0151 706 2805
Text phone number 18001 0151 706 2805

British Liver Trust
Tel: 0800 652 7330 or email helpline@britishlivertrust.org.uk

Haemochromatosis Society
Tel: 03030 401 101 or email helpline@huk.org.uk

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