

## Hereditary Haemochromatosis

A leaflet for people with hereditary haemochromatosis and their family



**local care:**  
global impact

## What is hereditary haemochromatosis?

**Hereditary haemochromatosis (HH)** is a treatable inherited condition where the body absorbs too much iron from the diet. When too much iron builds up in the body this is known as **iron overload**. The excess iron is stored in the liver and other organs of the body such as the pancreas, heart, endocrine (hormone producing) glands and joints.

## Why is the amount of iron in the body important?

A small amount of iron is stored in the liver and is essential for health, as it is needed when new red blood cells are formed. However when too much iron is stored in the liver, the liver becomes enlarged and damaged. Excess iron may also be stored in other organs and joints, causing damage.

## What are the symptoms of hereditary haemochromatosis?

- Constant tiredness, weakness, lethargy
- Abdominal pain
- Joint pain (arthritis); this can affect any joint but commonly affects the knuckle and first joint of the first two fingers
- Late onset diabetes
- Cirrhosis of the liver (scarring of liver tissue that damages liver function)
- Bronzing of the skin, like a permanent tan
- Loss of libido
- Irregular heartbeat

People with HH can have no symptoms for many years.

## What age do people develop hereditary haemochromatosis?

The onset of HH is normally between 30 and 60 years, as the build up of iron takes many years. However, women tend to develop HH later in life than men. The reason for this is that before the menopause, having periods (menstruation) regularly removes blood, and therefore iron, from the body. So before the menopause women do not accumulate as much iron in their bodies as men do.

## What causes hereditary haemochromatosis?

Hereditary haemochromatosis is caused by changes in a gene known as HFE.

We all have about 25,000 pairs of genes inside every cell of our body. Our genes are the instructions that tell our body how to grow and develop. We inherit one copy of each gene from our mother and the other copy from our father. When we have children we pass on one copy of each of our genes and our partner provides the other.

The HFE gene was identified as the cause of HH in 1996. HH is a recessive condition which means people with HH have changes in both their copies of the gene. These changes, technically known as mutations, can be thought of as spelling mistakes in the gene. There are two common gene changes, known as C282Y and H63D.

## What is the difference between C282Y and H63D?

People with two copies of the C282Y version of the gene are the most likely to develop haemochromatosis. However many individuals with two copies of C282Y do not accumulate enough iron to become ill.

People with one copy of C282Y and one copy of H63D have a **very small chance** of developing HH. Studies have shown that only about 1 person in every 100 (1 per cent) with this combination go on to develop HH and the degree of iron overload tends to be less than for individuals with two copies of C282Y.

Having two copies of H63D does not generally cause iron overload. An individual with one altered and one unaltered copy of the gene is known as a **carrier** of HH. Carriers generally do not accumulate enough iron to cause any tissue damage.

As HH is genetic, the family members of an individual with HH are at risk of being carriers or being affected themselves. There is more information about at-risk family members below.

## How common are changes in the haemochromatosis gene?

The gene changes that cause HH are very common. Approximately 1 in 10 people of Northern European origin carries one copy of C282Y, which means about 1 in 400 people carry two copies of this version and are at high risk of developing HH. 1 in 5 people of Northern European origin are thought to carry one copy of the milder H63D version of the gene.

Studies have shown HH is slightly more common in people in people of Irish and Celtic descent and slightly less common in all other ethnic groups.

## How do you test if someone has haemochromatosis?

It is possible to do a genetic test for HH. This is a blood test that checks the individual's HFE genes and identifies which versions of the gene that person has. This test cannot say whether that person definitely has HH, but it can identify who is at risk and who is not at risk of developing HH.

Two other blood tests (known as serum ferritin and transferrin saturation) are commonly used to **check the amount of iron in the blood.**

## What is the treatment for haemochromatosis?

The excess iron can be removed simply and effectively by regularly removing blood. This is known as **therapeutic venesection** or **phlebotomy** and is the same process as donating blood. A pint of blood is removed, usually every week, until iron levels return to normal. This process can take up to two years. When levels are normal, venesection is only needed a few times a year.

The treatment works because each pint of blood removed contains iron in red blood cells, and the body uses stored iron to make new red blood cells.

## Is it possible to cure or prevent hereditary haemochromatosis?

If the diagnosis is made early enough so treatment begins before the individual develops symptoms of HH then it is possible to prevent any serious complications. However it is not possible to undo tissue damage, such as cirrhosis of the liver, if that damage has already occurred. For this reason it is important that immediate family members of an individual with HH are offered genetic testing to find out if they are at risk.

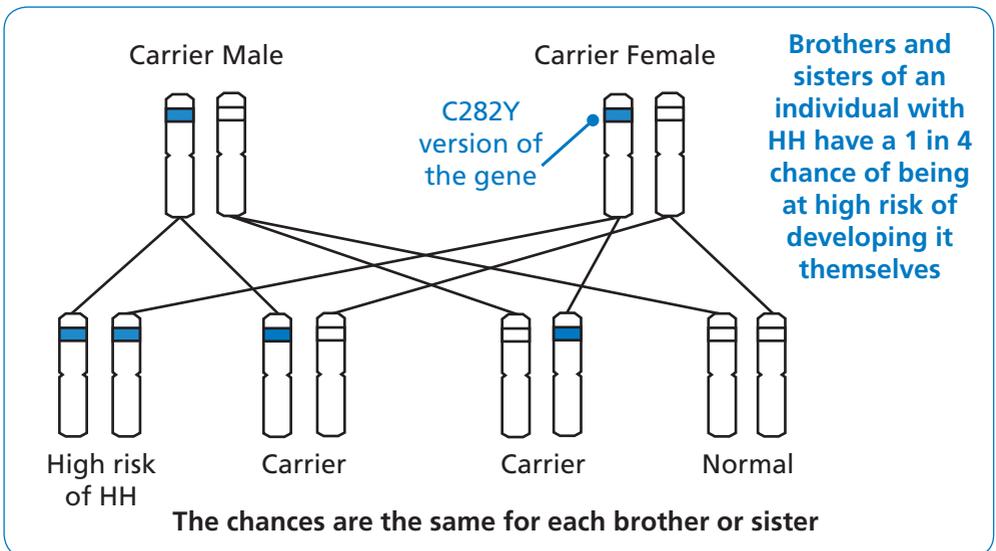
# Who in the family is at risk of hereditary haemochromatosis?

An individual with HH inherited each of their gene changes from their parents. This means the **parents** of an individual with HH will be carriers of HH. Occasionally the parent of someone with HH can have two altered copies of the gene so they are at risk of developing HH themselves and should have the iron levels in their blood checked.

When both parents are carriers, each of their children has a 1 in 4 chance of having two altered copies of the gene. This means that each **brother** and **sister** of an individual with HH has a 1 in 4 or 25 per cent chance of being at high risk of HH themselves.

In fact every time two carriers have a child there will be:

- a 1 in 4 (25 per cent) chance both parents will pass on the gene change so the child will be at high risk of HH
- a 1 in 2 (50 per cent) chance one parent will pass on the gene change and the other will pass the unaltered gene so the child will be a carrier of HH
- a 1 in 4 (25 per cent) chance neither parent will pass on altered gene so the child will not be a carrier of HH.

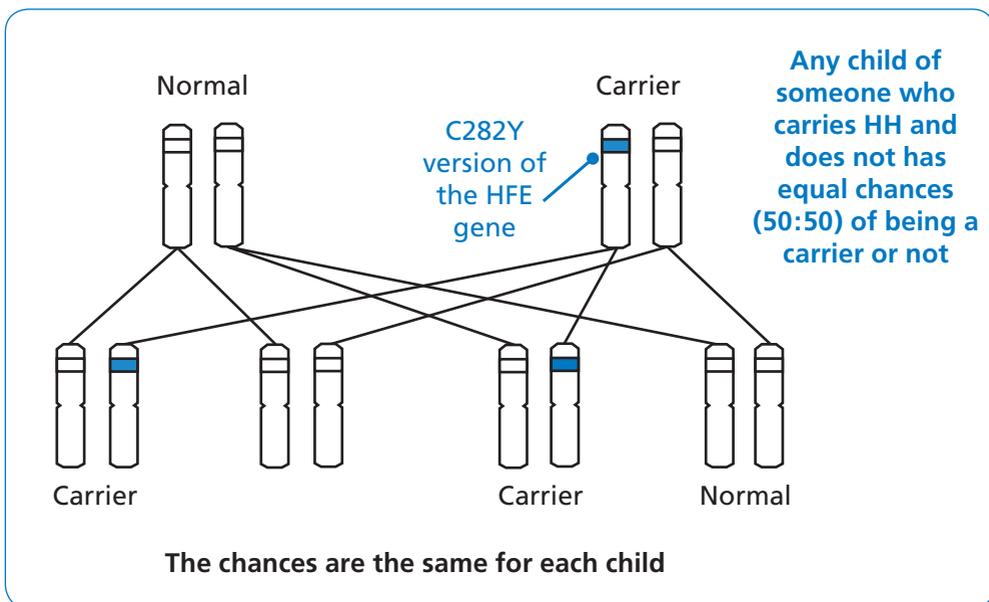


## Will my children be at risk of haemochromatosis if I am a carrier?

Individuals are only at risk of haemochromatosis if both of their parents are carriers. Therefore, the risk for your children will depend on whether their other parent is a carrier.

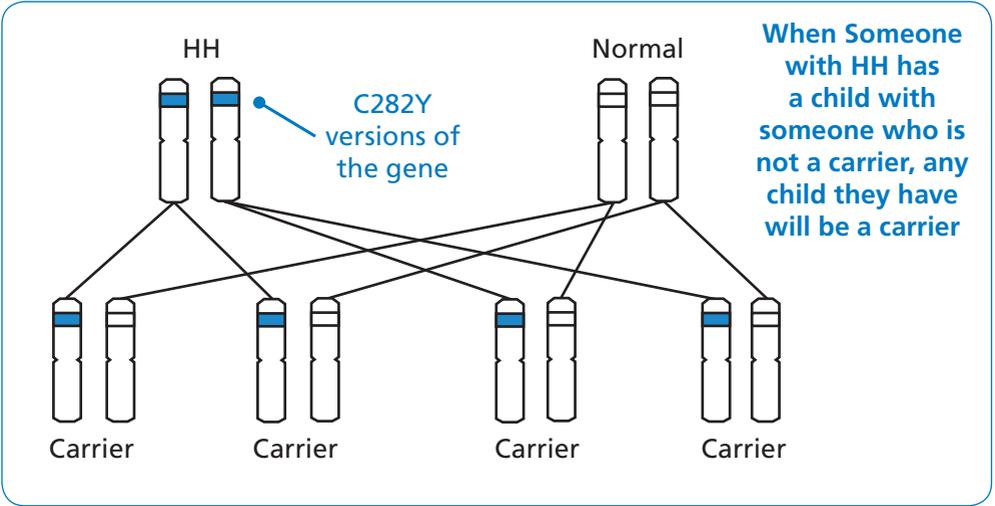
If both their parents are carriers of HH there is a 1 in 4 chance each of your children will be at risk of developing HH **when they are adults**. This is shown in the picture on the previous page.

If you are a carrier of HH but their other parent is **not**, you will not have a child with HH but your children will each have a 50 per cent chance of being a carrier. This is shown in the picture below.

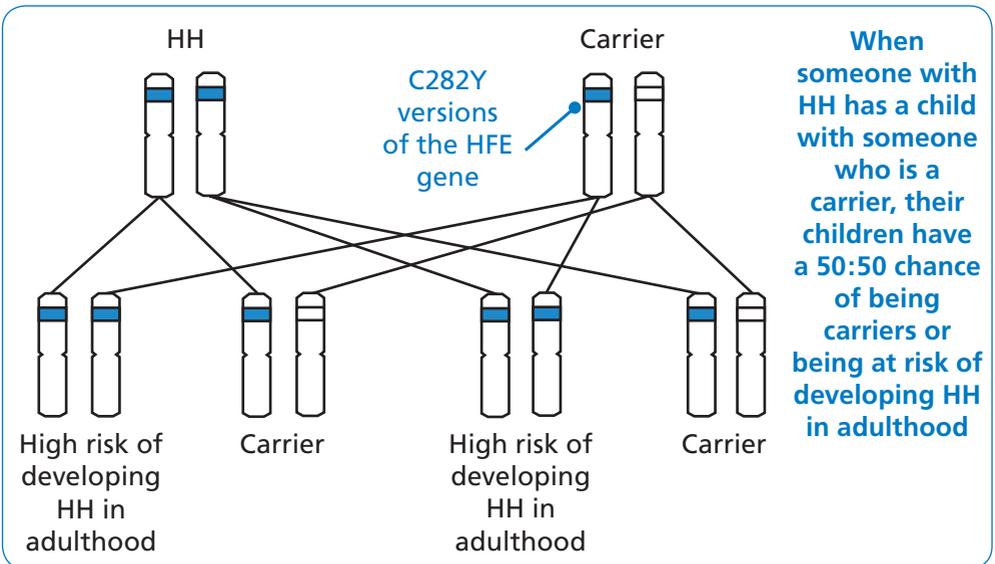


# Will my children be at high risk of haemochromatosis if I have the disease?

If your partner is not a carrier of HH you will not have a child with HH but all your children will be carriers, as shown below.



If your partner is a carrier of HH your children will have a 50 percent chance of being a carrier and a 50 percent chance of being at high risk of developing HH when they are adults.



## If I have one or two altered genes for haemochromatosis should my children have the genetic test?

The answer to this question depends on how old your children are. Hereditary haemochromatosis is a condition that affects **adults**. There is no medical reason for a child to have a genetic test for an adult onset condition.

However if your children are adults (or young adults) and would like to find out for themselves if they are at risk of developing HH then they should ask their GP (family doctor) about being tested.

## I am at risk of developing HH, how often should I have my iron levels checked?

If you have normal transferrin saturation and serum ferritin levels and you have been shown to have two copies of the C282Y gene, it is suggested that these levels are checked on a yearly basis.

If you have been shown to have the C282Y and H63D alterations and you have normal levels of serum ferritin and transferrin saturation levels, it is suggested they are repeated every three years.

## Should someone with haemochromatosis have a low iron diet?

An individual with HH can look after their health if they **avoid**:

- Vitamin pills and dietary supplements with iron
- Food that contain a large amount of iron, such as black pudding and food fortified with iron, such as some breakfast cereals

- Vitamin C supplements. This is because vitamin C can increase the absorption of iron from the diet
- Alcohol, as this can put strain on an already compromised liver

## Further information

### **Haemochromatosis Society contact details**

The Haemochromatosis Society  
Hollybush House  
Hadley Green Road  
Barnet  
Herts  
EN5 5PR  
Tel. 0208 449 1363  
[www.haemochromatosis.org.uk](http://www.haemochromatosis.org.uk)

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