HEREDITARY HAEMOCHROMATOSIS (HH)

UNDERSTANDING THE GENETICS

This leaflet is written for people with hereditary haemochromatosis or people with a family history of hereditary haemochromatosis.

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 may become enlarged and damaged. Excess iron may also be stored in other organs and joints, causing damage.

WHAT CAUSES HEREDITARY HAEMOCHROMATOSIS?

HH 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 and when we have children we pass on one copy of each of our genes and our partner provides the other.

HH is a recessive condition which means people with HH have changes in both their copies of the gene. These changes 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 **at risk** of developing HH. However many individuals with two copies of C282Y **do not** accumulate enough iron to become ill. Yearly blood tests are recommended to monitor iron levels.

People with one copy of C282Y and one copy of H63D have a **smaller risk** of developing HH and the degree of iron overload tends to be less than for individuals with two copies of C282Y.
It is recommended that iron levels are monitored at least every three years.

It is not clear if having *two copies of H63D* increases the risk of HH therefore occasional checking for iron overload is suggested.

An individual with one altered and one unaltered copy of the gene is known as a **carrier** of HH. Carriers do not accumulate enough iron to cause any tissue damage therefore monitoring of the iron levels is not required.

As HH is genetic, the family members of an individual with HH may **be carriers or be 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 9 people of Northern European origin carries one copy of C282Y, which means about 1 in 300 people carry two copies of this version and are at 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.

HOW DO YOU TEST IF SOMEONE HAS HAEMOCHROMATOSIS?

It is possible to do a genetic test for HH. This is a simple 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 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 total iron binding capacity) may be used to check the amount of iron in the blood. These tests can be carried out by your GP.

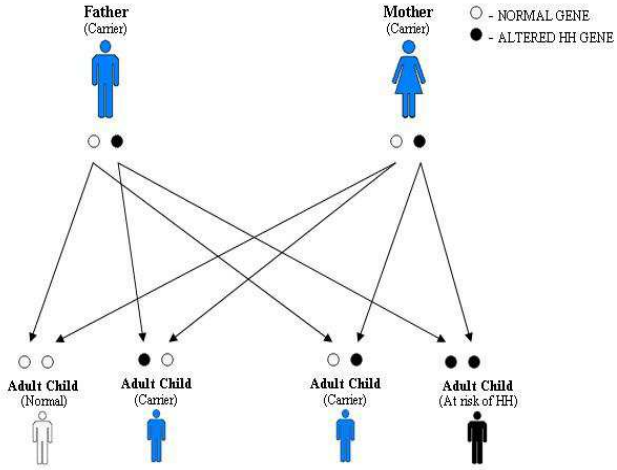
WHO IN THE FAMILY IS AT RISK OF HEREDITARY HAEMOCHROMATOSIS?

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.

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 **brothers** and **sisters** of an individual with HH have a 1 in 4 (25%) 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%) chance both parents will pass on the gene change so the child will be at risk of HH
- a 1 in 2 (50%) 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 %) chance neither parent will pass on the altered gene so the child will not be a carrier of HH.



The chances are the same for each brother or sister.

IF I HAVE ONE OR TWO ALTERED GENES FOR HH SHOULD MY CHILDREN BE OFFERED THE GENETIC TEST?

The answer to this question depends on how old your children are. Hereditary haemochromatosis is a condition that affects **adults**. Therefore testing is offered only when the child becomes an adult as there is no medical reason to be tested in childhood.

However if your children are 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.

WHERE CAN I FIND MORE INFORMATION?

The Haemochromatosis Society (www.haemochromatosis.org.uk)

If you have any questions you can contact your Clinical Genetics Team on

0131 537 1116.

Remember:

- **Carriers are NOT at risk of developing HH**
- **Not everyone with two altered copies of the gene develops symptoms of HH**