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Haemochromatosis: Patient Information

What is Haemochromatosis?

Hereditary haemochromatosis (HH) causes your body to absorb too much iron from the food you eat. The excess iron accumulates your organs, especially your liver, heart and pancreas. Too much iron in these organs can lead to life-threatening conditions such as cancer, heart arrhythmias and cirrhosis.

The disorder is due to a faulty gene. Many people carry the faulty genes that cause haemochromatosis — it is the most common genetic disease in Caucasians. But only a minority of people with the genes actually develop life threatening problems. Haemochromatosis is more likely to be serious in men, because women get rid of iron each month with their period, throughout their fertile years.

Signs and symptoms of hereditary haemochromatosis usually appear in midlife. The simplest way to remove iron down to safe levels is by regularly removing blood from your body using venesection.

What are the symptoms of haemochromatosis?

Many people with HH do not have symptoms. Often the earliest signs are rather vague, mimicking those of other common conditions. **Common symptoms** include:

- Joint pains or arthritis
- Fatigue or tiredness
- Weakness
- If iron overload is enough to cause organ damage, symptoms may include:
- Painful joints
- Diabetes (due to deposition of iron in the pancreas)
- Loss of sex drive (libido) – due to deposition of iron in the testicles
- Impotence
- Heart failure (due to deposition of iron in the heart muscle

When might I expect to see some signs and symptoms?

People with hereditary haemochromatosis have the condition from birth, but it takes years for iron to accumulate enough to cause symptoms. The first signs often occur between the ages of 50 and 60 in men and after age 60 in women.

When to see a doctor
If you are concerned that you may have this disorder, consult your doctor especially if you

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experience any of the signs and symptoms mentioned above. If there is a family history of this disorder, you can ask your doctor about specific genetic tests that can detect whether you carry the gene. The disorder itself is best evidenced by a high level of iron in your blood tests.

Hereditary haemochromatosis is caused by a mutation in a gene that controls the amount of iron your body absorbs from the food you eat. The mutations that cause hereditary hemochromatosis are passed from parents to children.

What are the gene mutations that cause haemochromatosis?

The commonest gene effected in hereditary haemochromatosis is called HFE. You inherit one HFE gene from each of your parents. There are two common mutations of the HFE gene, C282Y and H63D. More than 85% of people with hereditary haemochromatosis carry one of these mutations, and this can be detected on standard tenetic testing.

If you inherit 2 abnormal genes, you may develop haemochromatosis. This is called being "homozygous", and about 70 percent of people who inherit two genes will at some time develop the iron overload of hemochromatosis. You can also pass the mutation on to your children.

If you inherit 1 abnormal gene, you won't develop haemochromatosis. You carry the gene mutation and you can pass the mutation on to your children. They would not develop disease unless they also inherit another abnormal gene from another parent.

How hemochromatosis affects your organs

The hormone hepcidin controls how much iron is absorbed from your intestines. Hepcidin is made by the liver. Hepcidin also controls how iron is used in your body and how it is stored in your tissues.

Hepcidin fails to work normally in haemochromatosis and you absorb too much iron from your intestine. Excess stored iron can severely damage certain organs, leading to organ failure and chronic diseases such as cirrhosis, diabetes and heart failure. It is important to remember that only about 10% of people with haemochromatosis develop life threatening iron overload.

Other types of haemochromatosis

Juvenile haemochromatosis. This is a disorder of a different gene and causes much earlier onset of iron overload between the ages of 15 and 30.This disorder is caused by a mutation in the HJV gene.

Neonatal haemochromatosis. This is a very severe disorder where iron accumulates in a developing fetus.

Secondary haemochromatosis. This form of the iron overload is not inherited. Instead, iron accumulates because of too many blood transfusions, or disorders of blood cells where blood cells are destroyed too quickly, or in chronic liver disease or chronic infections.

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How is Haemochromatosis diagnosed?

Hereditary haemochromatosis is most often diagnosed these days because a high iron level is found on a blood test incidentally, or because a patient is aware of a family history of this disease.

Blood tests showing iron overload.
Serum transferrin saturation. Transferrin is an iron transporter and is normally less than 45% saturated with iron. If the transferrin saturation is higher than 45% this is suspicious for iron overload. Serum ferritin. The serum ferritin reflects how much iron is stored in your liver. Ferritin can be elevated in a number of conditions, so it is not used as the sole test for iron overload.

Other tests which may be ordered include:

Liver function tests. These tests can help identify liver damage.

MRI (magnetic resonance imaging) can be used to measure the degree of iron overload in your liver.

Gene mutation testing. Testing for the HFE gene is recommended if you have high levels of iron in your blood.

Liver biopsy. If liver damage is suspected, a biopsy of the liver can be sent to a laboratory to be checked for the presence of iron as well as for evidence of liver damage, especially scarring or cirrhosis. Risks of biopsy include bruising, bleeding and infection.

Screening healthy people for hemochromatosis
It is advisable to offer genetic testing to all first-degree relatives (parents, siblings and children) of anyone diagnosed with haemochromatosis. If a mutation is found in only one parent, then children do not need to be tested.

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