

What is haemochromatosis? ‘Think Haemochromatosis’

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Haemochromatosis is a genetic disorder where too much iron is absorbed from the diet. This iron is stored mainly the liver, but also the pancreas, heart and the joints. Excessive iron stores can damage the liver, joints, heart and pancreas.

What are the symptoms?

Iron builds up slowly so the symptoms may not appear until the age of 30 to 40 years. Most people don't have any symptoms, as the iron builds up very gradually over years. However, symptoms may include:

- Chronic fatigue
- Arthritis
- Diabetes
- Liver disorders
- Sexual dysfunction
- Cardiomyopathy or irregular heartbeat
- Skin pigmentation

These symptoms are very common in many other disorders.

Arthritis affecting the first two fingers is highly suggestive of haemochromatosis, but uncommon. Chronic fatigue, weakness and lethargy may be ascribed to the after-effects of a viral infection or to psychological causes, and abdominal pain to irritable bowel syndrome. Similarly, liver disorders may be put down to excessive alcohol intake, even in a moderate drinker. Think haemochromatosis.

People with haemochromatosis may eventually develop some of the above symptoms, although possibly in a very mild form. The need for treatment does not depend on the presence of symptoms.

Diagnosis and Treatment

What are the tests?

These consist of a simple blood test for iron (ferritin), ideally performed after an overnight fast.

Serum Ferritin: Elevated ferritin may be due to iron overload, but there are many other causes of a high level

Genetic Tests: There is now a genetic blood test that can identify haemochromatosis. This allows adult family members to be tested: as we don't test children. Early detection before tissue damage has occurred is key.

Treatment

Treatment is most effective when begun early on in the disease as it can successfully prevent or stop organ damage. If damage has occurred then treatment should halt any further damage and in most cases will bring about an improvement.

The easiest way of removing excess iron from the body is by removal of blood. This is like giving a blood donation. Every pint of blood removed contains iron. The body then uses some of the excess stored tissue iron to make new blood cells which are removed in subsequent phlebotomy. The length of treatment depends on the amount of excess iron in the body. Treatment may mean weekly phlebotomy for one to two years, or until the iron levels have been reduced to a safe level. Once the initial treatment is completed and the iron levels are back to normal then they are monitored every three months.

Treatment for haemochromatosis is lifelong, usually 2-4 times each year and may require blood to be removed once or twice yearly depending on how quickly the iron is reaccumulating. This is called maintenance therapy.

Diet

Eat a normal healthy diet with:

- Modest alcohol consumption
- No iron medication or multi vitamins containing iron.
- No bread or cereals with fortified iron

Summary & Conclusion

- Haemochromatosis is common in Ireland
- Haemochromatosis (HHC) is often clinically 'silent': often no symptoms/signs until late
- Ask your GP to test your iron level at some stage: Don't assume it is a 'routine' blood test.
- Treatment is really easy once HHC is diagnosed
- All first-degree adult relatives of a patient with HHC should be tested.
- Think Haemochromatosis!