moore barlow

Factsheet

Genetic Haemochromatosis

Genetic Haemochromatosis (GH), also known as hereditary haemochromatosis, is an inherited condition in which a faulty gene causes the body to absorb too much iron from the diet.

What is Genetic Haemochromatosis and who is at risk?

The body has no means to excrete excess iron, which is deposited within various organs and tissues, slowly building up over several years causing damage. The most susceptible organs to iron overload are the liver, heart, adrenal glands, pancreas and the joints.

The gene that controls haemochromatosis (also known as inherited iron overload disorder) is known as HFE. There are several different mutations to this gene but the C282Y mutation is associated with most cases of hereditary haemochromatosis.

Early diagnosis is difficult as the condition often shows no symptoms (asymptomatic). Those initial symptoms that do show can be vague and non-specific. This means that the diagnosis of GH is often made incidentally. For example, this could be following a finding on routine blood screening of abnormal liver function tests or elevated ferritin.

Screening criteria are difficult as not everyone with the faulty gene will go on to develop iron overload. However, family members of those who have been diagnosed should be tested. Screening should also be considered in patients with unexplained abnormal liver function tests.

What are the symptoms?

Many of the symptoms listed below are non-specific and may have other causes. However, these symptoms may also be the presenting main complaint in GH and therefore a diagnosis of GH should be considered if any of the following symptoms, especially in combination, are present:

- Chronic fatigue, lethargy and weakness
- Arthritis affecting any joints but particularly the second and third MCP joints (knuckle joints)
- Diabetes (late onset)

- Cardiomyopathy and abnormal heart rhythms
- Abdominal pain, often non-specific
- Abnormal liver function, enlarged liver, cirrhosis and cancer
- Psychiatric/neurological disorders such as depression, irritability, mood swings and impaired memory
- Bronzing of the skin (permanent tan)
- Erectile dysfunction, loss of sex drive
- Absent or irregular periods.

Treatment and management

If left untreated, iron overload leads to organ damage with potentially serious and irreversible complications. Therefore, early diagnosis is vital as treatment can lessen or even prevent the late effects of iron overload.

GH can be diagnosed via simple blood tests (known as transferrin saturation and serum ferritin) which provide a crude measure of iron stores in the body. These tests are then followed by a simple genetic test (another blood test) that tests for the HFE gene mutation.

Treatment is simple and effective and involves the regular removal of blood, known as venesection or phlebotomy. Depending on the degree of iron overload, initially this may need to be done every week. During this time, ferritin levels are monitored and once they are restored to normal levels (indicating depleted iron stores), venesection will continue as maintenance therapy every few months to ensure iron levels remain within the normal range.

Other tests will likely be required to assess whether there is any organ damage as well as the extent of any organ damage.

Treatment resulting in depletion of iron stores should lead to a decrease in some symptoms and may reverse some less serious conditions associated with GH such as mild fibrosis of the liver and heart arrthymias. However, it will not cure more serious conditions linked to the illness such as diabetes or liver cirrhosis. In these instances, on-going assessment and management of these conditions will be required.

If a mistake has been made leading to a delay in diagnosing and treating your GH, you may be entitled to pursue a claim for compensation

We all place our trust in doctors, nurses and other health professionals when we become ill. However, we all know many people receive their hereditary haemochromatosis diagnosis far later than they should have done, which can lead to significant and lasting consequences.

Sadly, mistakes do happen. If a doctor, nurse or other healthcare professional has failed in their duty to provide an appropriate level of care or skill and a reasonable opportunity to diagnose GH at an earlier stage was missed, resulting in injury, loss or damage – then you may be entitled to pursue a claim for compensation.

Further information and support

The Haemochromatosis Society is a patient-run UK charity that provides information and support to people living with haemochromatosis. It has a helpline as well as face to face support groups.

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