

## Hereditary haemochromatosis

Hereditary haemochromatosis is a common condition in which too much iron is absorbed from food and stored in the body. It is usually picked up between the ages of 30 and 60 – earlier in men than in women.

The condition can cause tiredness, sore joints, loss of sex drive, impotence, abdominal pain, heart disease, diabetes, an enlarged liver and even, occasionally, cirrhosis of the liver and liver cancer.

If it is not detected for many years, hereditary haemochromatosis can be a severe condition.

But if picked up early and treated, the condition can be mild. People with treated hereditary haemochromatosis usually have a normal lifespan.

Haemochromatosis arises from an alteration to a particular gene known as the HFE gene. It follows an autosomal recessive pattern of inheritance (see fact sheet on '*How do genetic conditions occur?*'). This means that someone with the condition has an alteration in both copies of their HFE gene. The parents of an affected person will be carriers and there are likely to be other family members who are carriers. There may also be relatives with alterations in both copies of the gene who are healthy at present, but will develop the disorder in the future.

It is worth noting that having alterations in both HFE genes does not give you haemochromatosis – it only indicates that you have a high likelihood of getting the condition. At least a third of people with alterations in both their HFE genes will have no problems from it.

If someone in the family has hereditary haemochromatosis, then all members of the immediate family should have blood tests known as iron studies, which measure the amount of iron in the body. They should also have tests to see if they have two, one or no alterations in HFE.

Those who have abnormal iron studies will usually be referred to a specialist for follow up and treatment. Those with alterations in both HFE genes but normal iron studies need follow up by their doctor.

Depending on these results, your doctor may recommend that other adult family members also be tested. Because the condition does not generally affect children, it is usually best to wait to test until they are adults.

A word of caution about the gene test. The test picks up both HFE gene alterations in most people with hereditary haemochromatosis. But it does not always do so and it is possible to have hereditary haemochromatosis, but not have it show up on a genetic test.

Anybody with hereditary haemochromatosis, and their close relatives, should have genetic counselling, which may involve referral to a genetics service.

## Contacts and further information

- Your local genetic service, which you can contact through your nearest community health centre, public hospital or health department.
- Australasian Genetic Alliance at <http://www.australasiangeneticalliance.org.au>
- Haemochromatosis Society Australia at <http://www.haemochromatosis.org.au>
- Better Health Channel at <http://www.betterhealth.vic.gov.au>
- MyDr at <http://www.mydr.com.au>
- The Centre for Genetics Education at <http://www.genetics.edu.au>
- HealthInsite at <http://www.healthinsite.com>
- MedicineNet at <http://www.medicinenet.com>
- For other related fact sheets, you can contact the Gene Technology Information Service on **free call Australia-wide 1800 631 276** or email [gtis-australia@unimelb.edu.au](mailto:gtis-australia@unimelb.edu.au) or visit Biotechnology Australia's website at <http://www.biotechnology.gov.au>