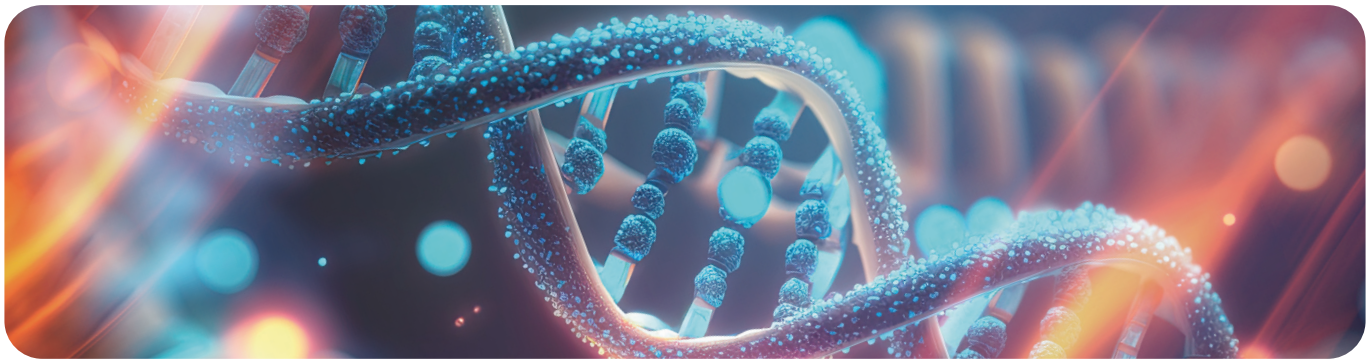
# PATHOLOGY TESTS EXPLAINED

Information about pathology tests to help everyone take control of their health and make the right decisions about their care.

## WHAT YOU SHOULD KNOW ABOUT
## GENETIC TESTING FOR HAEMOCHROMATOSIS

Haemochromatosis is a disorder that causes your body to absorb too much iron from your diet. This results in a condition called iron overload in which iron builds up in your body. The most common type of haemochromatosis is inherited, and it is caused by a fault in the *HFE* gene.



### What's going on?

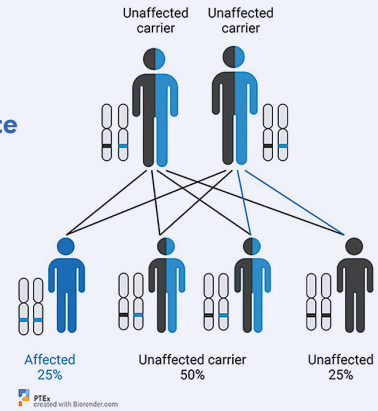
Our bodies cannot make iron. Iron must be absorbed from our diet or from supplements. The amount of iron we absorb is decided by our body's needs. We normally use just a little bit of iron each day and we don't have a way of getting rid of excess iron. Instead, the body closely controls iron levels by regulating how much iron we take in from our diet.

In haemochromatosis there is a breakdown in the process that controls iron levels and the body behaves as if there is an iron shortage. It responds by absorbing more iron from food than we need. Because there is no mechanism to get rid of it, the excess iron accumulates in organs and tissues where it can cause damage.

### How do we inherit an *HFE* gene mutation?

Everyone has two sets of genes. One inherited from their mother and one from their father. These genes in the form of DNA are packaged up in chromosomes which are found in most of the cells in our bodies.

**Recessive inheritance**




Humans have 23 pairs of chromosomes – one of the pair comes from the father and one from the mother.

The *HFE* gene is on chromosome 6.

#### Two gene mutations

If you inherit gene mutations from both your mother and your father, you are at risk of haemochromatosis, although many people never develop the disorder. You will pass the altered gene on to your children and all your children will be carriers of at least one copy.

#### One gene mutation

If you inherit a mutation in one gene from either your mother or father, you are not at risk of haemochromatosis although you have a 50 per cent chance of passing the mutation on to your children.

## How is haemochromatosis diagnosed?

Your doctor will start by ordering two blood tests:

- Full Blood Count (FBC) measures the amount of haemoglobin in your blood and looks at the size and shape of red blood cells.
- Iron studies is a group of several tests that assesses the amount of iron carried in your blood and stored in your body.

If your results indicate haemochromatosis, the next step is to order an *HFE* gene test. This will show if there is a mutation in the *HFE* gene and it is the only definitive way to make a diagnosis.

## What your results can tell you?

Mutations are named according to their location on the gene. Most cases of haemochromatosis are found in people who have two copies of the C282Y mutation, one on each of the pair of chromosomes. Another mutation H63D can slightly increase the risk of haemochromatosis and another one, S65C can lead to mild to moderate iron overload but not cause haemochromatosis.

About 100 mutations in the *HFE* gene have been recognised but most of these are very rare and the effects of many of them are not yet known. Only C282Y and H63D mutations are commonly tested for. In some circumstances, your doctor may request that the laboratory look for one of the rare mutations.

## Having a medical test

The choice of tests your doctor makes will be based on your medical history and symptoms. Make sure you tell them everything you think might help.

You play a central role in making sure your test results are accurate. Do everything you can to make sure the information you provide is correct and follow instructions closely.

Talk to your doctor about any medication you are taking. Find out if you need to fast or stop any particular foods or supplements. These may affect your results.

## What happens next?

Treatment of haemochromatosis is usually straightforward and requires regular removal of blood to get rid of the excess iron. This is known as venesection and is similar to a blood donation.

### Questions to ask your doctor

**Why does this test need to be done?**
**Do I need to prepare (such as fast or avoid medications) for the sample collection?**
**Will an abnormal result mean I need further tests?**
**How could it change the course of my care?**
**What will happen next, after the test?**

For more detailed information on these and many other tests go to **pathologytestsexplained.org.au**

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**www.pathologytestsexplained.org.au**

Please use this QR code to access more information

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Pathology Tests Explained is managed by a consortium of medical and scientific organisations representing pathology practice in Australia. More details at: **www.pathologytestsexplained.org.au/about**

### My Health Record

You'll find a direct link to the Pathology Tests Explained website embedded in the pathology results pages of your My Health Record.

Click on the link to find information about what your tests are investigating or measuring and what your results can tell your doctor.