Dr Michael Dickinson
MBBS (Hons) DMed Sci FRACP FRCPA
Assoc Prof Max Wolf
MBBS FRACP FRCPA

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.