

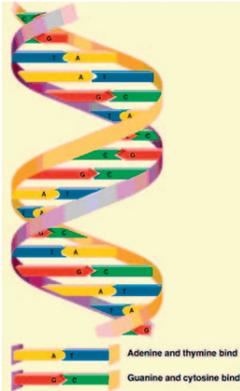
Diagnosis

Biochemical Test : A simple blood test called a serum iron profile measures both:

- > Transferrin Saturation (or iron transport protein). A fasting transferrin saturation above 50% is very suggestive of Haemochromatosis (HH) ⁽¹⁾
- > Serum Ferritin (iron storage protein). An elevated result may be due to iron overload. The result should be interpreted in combination with transferrin saturation⁽²⁾

⁽¹⁾ Iron transfer through the blood

⁽²⁾ Iron storage in the organs



A genetic test on blood or saliva will confirm the diagnosis. The genetic test for the double mutation C282Y/C282Y in HFE gene will detect the most common form of Haemochromatosis.

Others rare forms of HH (non HFE specific mutations) can be tested for in a specialised laboratory or a **National Reference Centre**.

A family case

If the test is positive, it is the responsibility of the patient to inform his/her relatives and to encourage biochemical and genetic testing.



European Federation of Associations of Patients
with Haemochromatosis

Fédération Européenne des Associations de Malades
de l'Hémochromatose

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EFAPH targets

To raise awareness of Haemochromatosis amongst the Medical Profession and the Public in general.

To assist in setting up national awareness associations in all countries and to promote best available treatment and research.

	Austria*		Ireland
	Belgium		Italy
	France		United Kingdom
	Germany		Norway
	Greece*		Portugal
	Hungary		Spain
	Iceland		Switzerland*

* Planned



Irish Haemochromatosis
Association *Iron Overload*

The Carmichael Centre
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non-contractual ordeal

HAEMOCHROMATOSIS

Genetic iron overload disorder



Information - Prevention
Treatment



What is Haemochromatosis ?

Iron Overload

Haemochromatosis is due to a progressive and excess iron storage in the body. If not treated it may lead to serious damage to vital organs such as liver, heart, pancreas, joints and in extreme cases may lead to death.

Haemochromatosis is hereditary. Parents, siblings and children of the diagnosed person are likely to carry a single C282Y mutation or may have the disorder.

A genetic disease

THE MOST FREQUENT FORM OF THE DISEASE

A common mutation called C282Y/C282Y in **HFE** gene is responsible for the most frequent form of Haemochromatosis. This mutation causes excessive iron in blood resulting in severe organ damage : chronic fatigue, cirrhosis, liver cancer, diabetes and disabling rheumatism.

The mutation is **very common** in populations that migrated from Northern European countries especially those from Celtic origin.

RARE FORMS OF HAEMOCHROMATOSIS

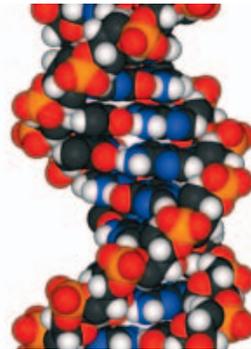
Children and teenagers can also have severe iron overload. In these very rare juvenile cases, the disease is due to a mutation in genes **others than HFE**, hemojuvenile, hepcidin, ferroportin... These rare forms **are found** world-wide.

National reference centres specialising in iron overload disease will help with diagnosis and appropriate treatment.

An early diagnosis

Non specific warning signs could be misinterpreted and thus delay diagnosis:

- > Abnormal and chronic fatigue,
- > Painful joints,
- > Cardiac arrhythmia,
- > Diabetes,
- > Sexual disorder,
- > Skin Pigmentation...



Provided that Haemochromatosis is diagnosed early and treated, the patient can have a normal life and normal life expectancy.

However, delayed diagnosis could lead to complications, organ damage and a much more serious prognosis.

At the present time, not enough is known about Haemochromatosis in most European countries.

One person in 300 is genetically predisposed to have iron overload. In Europe this would mean that about 2.2 million people may have the disorder. In Ireland HH is much more common and it is estimated that one in 83 people could be at risk of iron overload. The very rare forms of haemochromatosis **not related to HFE** are as yet not fully understood.

A quick and effective treatment



At present the treatment consists of blood-letting or phlebotomy.

Phlebotomy is performed on a regular basis in order to eliminate iron overload.

The physician organises the phlebotomy routine as required - frequency and volume. In the first phase (induction treatment) one blood phlebotomy may be performed each week until the overload disappears.

Phlebotomy becomes less frequent in the second phase (maintenance phase) each month to every six months, aiming at preventing iron build up in the body.

Medical research is ongoing and alternative treatment to phlebotomy is being explored. It is vital to have a better understanding of iron metabolism.

