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 [1]

> Serun ferritin (iron storage protein). An elevated result may be due to iron overload. The result should be interpreted in combination with transferrin saturation[2]

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 **National Reference Centre**.

A family case

If the test is positive, it is the responsability 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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www.european-haemochromatosis.eu

EFAPH targets

To raise awareness of Haemochro-matosis 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*
- Belgium
- France
- Germany
- Greece*
- Hungary
- Iceland
- Ireland
- Italy
- United Kingdom
- Norway
- Portugal
- Spain
- Switzerland*

** Planned*

non-contractual ordeal

04/2010 - Texte : Dr Françoise COURTOIS - Jean RAILLAND - J. LASSALE - Maquette : Imp. des Longchamps - Rennes

HAEMOCHROMATOSIS

Genetic iron overload diseases

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 diseases on target organs or vital organs such as liver, heart, pancreas, joints and in extreme cases may lead to death.

Haemochromatosis is hereditary. Parents and offsprings of the diagnosed one likely may 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

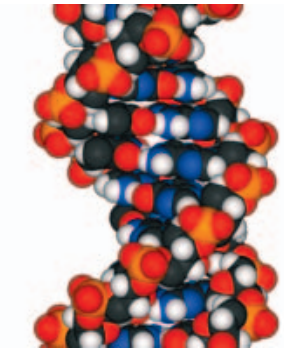
Children and teenagers can also have severe iron overload. In those very rare juvenile cases, the disease is due to a mutation in genes **others than HFE**, hemojuvenile, hepcidin, ferroportin... These rare forms **are found** world-wide.

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

An early diagnosis

Non specific warning signs could mislead one's interpretation, and thus delay diagnosis :

> Abnormal and chronic fatigue,
> Painful joints,
> Cardiac arrhythmia,
> Diabetes,
> Sexual disorder,
> Grey pigmentation of complexion...



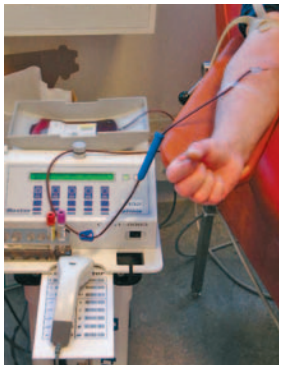
Provided that Haemochromatosis is diagnosed early and treated, the patient can have a normal life and normal life expectancy.

However, delayed diagnosis will 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 millions 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 **non related to HFE** are still misunderstood.

A quick and effective treatment



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

Phlebotomies are periodically repeated in order to eliminate iron overload.

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

Blood-lettings become 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 an alternative treatment to phlebotomy is being explored. It is vital to have a better understanding of iron metabolism.