Genetic Heritance: which family members have to be monitored?

HH patients have inherited mutated genes from **both** parents. A ferritin and transferrin saturation check, together with the genetic test execution, is necessary in **patients' siblings**. Ferritin and transferrin saturation levels should also be monitored in **offspring** (after 18 years old).

5

Early diagnosis and treatment allow the patient to live a normal life.

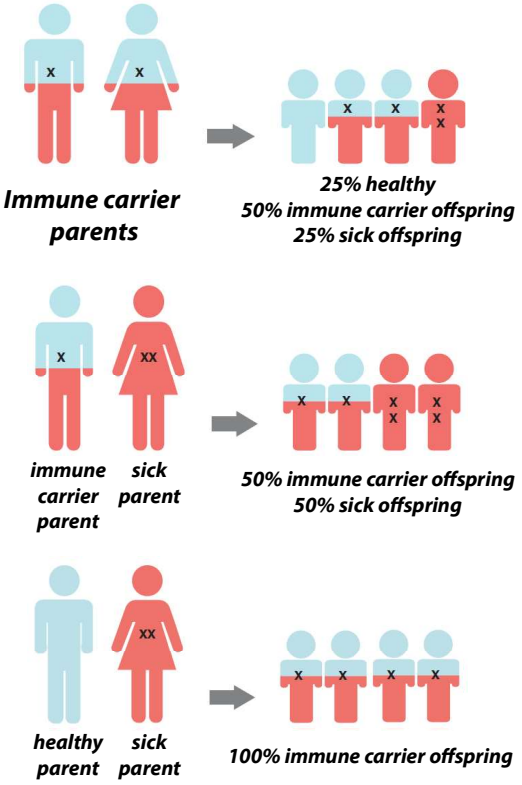

Image editing by: Sheahan O., O'Connell E. (2009).
Hereditary haemochromatosis: patient support and education, *Nursing Standard*

Nutrition

- A particular type of diet is not necessary

- **Reduce** or **avoid alcohol** consumption (**max 1 glass of wine at meal**)

- Avoid multi-vitamins or iron dietary supplements

- Reduce meat consumption (**no more than 3 times a week**).

6

Contacts:

Outpatient room for Iron Diseases:
University Hospital "Ospedale Policlinico B.go Roma",
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Reference person: Prof. Domenico Girelli

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Blood Transfusion Centres:
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Tel +39 045 812 4321
General Hospital "Ospedale Borgo Trento"
Piazzale Aristide Stefani, 1 – 37126 Verona, Italy.
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Patient Associations:
European Federation of Associations of Patients with Haemochromatosis (http://efaph.eu/)
Associazione per lo studio dell'Emocromatosi e delle Malattie da Sovraccarico di Ferro –ONLUS (http://www.emocromatosi.it).

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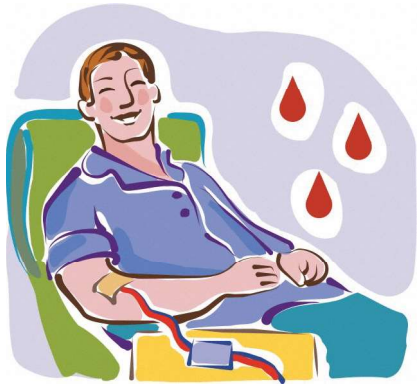
Azienda Ospedaliera Universitaria Integrata Verona

Patient's Guide

Hemochromatosis

Know the disease to learn how to deal with it...



Six steps to better understand the pathology and the cure

GRUPPO INTERDISCIPLINARE MALATTIE DEL FERRO
GIMFer
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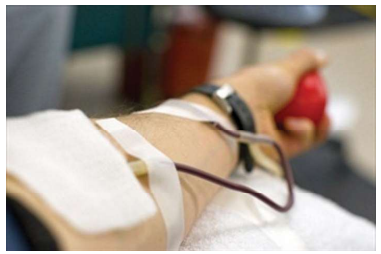
Hereditary Hemochromatosis (HH) Definition

Hemochromatosis is a hereditary genetic disease causing an **excess in body iron absorption**.
The most "common" form of Hemochromatosis is due to mutations on the HFE gene, the most frequent ones being the C282Y and the H63D.
As time goes by, iron progressively accumulates in the body leading to severe damages.

1



Symptoms and Common Signs

Early Symptoms:
- **Abnormal and chronic fatigue**
- **Abdominal ache**
- **Painful joints**

Late Symptoms
(associated to iron accumulation):
- **Hepatomegaly (enlarged liver)**
- **Grey pigmentation of complexion (skin darkening)**
- **Diabetes mellitus**
- **Progressive liver damage leading to cirrhosis**
- **Cardiac arrhythmia**
- **Amenorrhoea and loss of libido**

2

Iron Absorption and Iron Overload Diagnosis

Iron intake through the diet is essential for red blood cells production. Iron presence into the body is strictly regulated. If the mechanism of regulation is not properly functioning, constant iron absorption leads to the accumulation in different organs: **liver, pancreas, heart and skin**.

Serun ferritin
indicates the amount of accumulated iron

Transferrin saturation
indicates how much iron is present in the blood

If this value is above 50% iron accumulates

3

How is Iron Overload Treated?

At present, **phlebotomy** is the **first choice treatment**.
Phlebotomies consist of blood withdrawals customised for the single patient in frequency and volume (ca. 400 mL).
The procedure lasts 15-30 minutes and it's performed at the hospital **blood transfusion unit**.

Phlebotomy therapy is organized in two phases:

Induction Phase

Maintenance Phase

4

Induction Phase

The goal is to decrease iron deposit. It consists in doing blood phlebotomies every **7-14 days**.

The first phase duration is variable according to the individual answer.
It's over when ferritin levels in blood reach the value of about **100 mcg/L**.

Being HH a genetic disease, iron parameters' monitoring is essential even after the removing of accumulated iron.

Maintenance Phase

It follows the induction phase.

This phase aims at avoiding new iron accumulation. Ferritin and Transferrin saturation level are monitored and phlebotomies become less frequent, every **3-4 months**.

This phase lasts all-life long.

Preparation to Phlebotomy therapy

Drink at least two litres of water every day

Fasting is not necessary