**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).

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

**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).

**Contacts:**

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**Patient Associations:**
European Federation of Associations of Patients with Haemochromatosis (http://efaph.eu/)

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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.

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

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.

Serum 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.

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

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.

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