Therapeutic recommendations in *HFE* hemochromatosis: specialist-patient knowledge transfer

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HEREDITARY HAEMOCHROMATOSIS (HH)

HFE-related haemochromatosis (HH) is an iron-overloading disease which, in people with Northern European ancestry, is one of the most common genetic conditions, with an estimated one in 150 to 200 individuals having *p.Cys282Tyr* homozygosity. Excess iron cannot be excreted by the body, and is taken up by the liver, pancreas, and heart.

The disease is polymorphic, and up to 50% of *p.Cys282Tyr* homozygotes may need venesection therapy. Diagnosed early, patients, treated by venesections, have a normal life expectancy. However, early symptoms such as tiredness and arthralgia, typically developing in mid-life, are non-specific and frequently overlooked. Patients often present too late to prevent shortened life-expectancy, typically presenting with liver cirrhosis or cancer, diabetes, or cardiac disease.

Early detection and treatment depend on increased awareness and proper information among health professionals and patients.

Although guidelines are available for HH, a large number of recommendations are not shared between those different guidelines.

OBJECTIVE

Our aim was therefore to provide an **objective, simple, brief, and practical set of recommendations on therapeutic aspects of HH**, understandable by patients/citizens without medical training.

METHODS

Haemochromatosis International (HI), an alliance of haemochromatosis patients associations, invited experts to produce a document based on published scientific studies and guidelines.

The definitive version of these recommendations was approved at the Haemochromatosis International Meeting, on May 12th, included in International Bioiron Society Meeting 2017, in Los Angeles.

RESULTS

1 WHAT IS THE TREATMENT?

Phlebotomy (venesection therapy) is the standard treatment for patients with HH having been used for more than 60 years. It is effective in reducing morbidity and mortality of HH [1].

2 WHO TO TREAT AND WHEN TO START?

Patients with *HFE* p.Cys282Tyr (C282Y/C282Y) homozygous genotype and biochemical evidence of iron overload, i.e., increased serum ferritin (>300 µg/L in male and postmenopausal female and >200 µg/L in premenopausal female) and increased fasting transferrin saturation (≥ 45%) [2, 3].

3 HOW TO TREAT?

<u>Induction Phase</u>

A phlebotomy schedule of the order of 400-500 mL, considering body weight, weekly or every two weeks has been proposed [2, 4].

The objective in this phase is usually to reach SF ≤ 50 µg/L. SF should be checked once a month until the values reach the upper normal limits, and every two weeks thereafter, until the final goal of SF is reached [2, 4] .

<u>Maintenance Phase</u>

The patient with HH needs lifelong follow-up.

One phlebotomy every 1 to 4 months, depending on the patient's iron status [2, 4].

Efficacy: the usual aim is to maintain ferritin levels around 50 µg/L [2, 3].

4 WHEN TO STOP?

Patients who have had iron overload should never stop having their iron status monitored and their treatment planned in the light of their iron status, general condition and age.

CONCLUSION

This document is an important and accessible source of information about HH that could be very useful for patients, and can also contribute to HH awareness.

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ACKNOWLEDGEMENTS

HI gratefully acknowledges the support of Novartis, DRG Germany and DRG USA with the start-up and running of this Charitable Incorporated Organization.