ACADÉMIE NATIONALE DE MÉDECINE

Haemochromatosis: time for screening has come![1]

Press release of the French National Academy of Medicine

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While iron deficiency is well known, iron overload remains largely unrecognized. However, there are a lot of data supporting its impact on health. Haemochromatosis is a genetic disorder characterized by the insidious development of an iron excess linked to a deficiency in the iron hormone called hepcidin. This results in hyper-absorption of the iron normally contained in the diet. The genetic profile which predisposes to this disorder is very common, a recent large study (1) showing that it is present in more than 6 out of 1000 Caucasians.

The clinical consequences (2, 3) appear in adulthood and are a source of morbidity (fatigue, impotence, joint pain, osteoporosis, liver hypertrophy, diabetes, tanned complexion) and mortality (cirrhosis, liver cancer, cardiac damage), these different disorders being either isolated or variously associated. The diagnosis of haemochromatosis, provided it is thought of, has become very simple, based on the following triad: i) Clinic; ii) Biology: elevation in the blood of the transferrin saturation coefficient (efficient reflection of the iron level) and ferritin (reflection of the overall iron stock in the body), presence of the C282Y duplicate mutation, and iii) Imaging ("iron MRI"), which allows the overload to be visualized and quantified. This diagnostic approach is "non-invasive" as it avoids the need for a liver puncture.

At very rare occurrence for a genetic disease, haemochromatosis benefits from a simple, well-tolerated, effective and very inexpensive treatment: bloodletting. Applied early, repeated blood withdrawals allow a return to a normal life and preserves life expectancy. However, due to the insufficient financial value of bloodletting, the supply of care has been considerably reduced in recent years, with prolonged waiting times resulting in a loss of opportunity for a large number of patients. This development is all the more regrettable as haemochromatosis benefits from an institutional recognition through a national reference center working in close collaboration with several competence centers (4). It also benefits from a strong associative support (5). The medium term innovative therapeutic perspective is represented by hepcidin supplementation which will allow a return to a normal iron metabolism.

Apart from these "curative" therapies, we must stress the importance of the preventive approach, and particularly of family surveys, which are still insufficiently carried out.

On the occasion of the World Haemochromatosis Week (June 1-5, 2021), the French National Academy of Medicine insists on the following measures:

[1] Press release of the French National Academy of Medicine Rapid Communication Platform validated by the members of the Board of Directors on May 10, 2021.

- To improve the knowledge of health workers, in training and in practice, on haemochromatosis so as to detect the disease as early as possible;

- To urgently revalue the act of bloodletting, a treatment that is still very much in use, in order to allow an access to care, that is no longer satisfactorily provided in our country;

- To raise awareness of the public health issue by developing a real national network for carrying out family surveys, and by initiating a population screening strategy, from young adulthood, based on simple blood tests such as the combination of the transferrin saturation coefficient and the ferritin level.

References

1. Pilling L.C. et al. Common conditions associated with hereditary haemochromatosis genetic variants: cohort study in UK Biobank. BMJ 2019 Jan 16;364:k5222. doi: 10.1136/bmj.k5222

2 HAS (Haute Autorité de Santé). ALD n°17. Haemochromatosis. 2012 https://www.has-sante.fr/jcms/c_592230/fr/ald-n17-hemochromatose

3. Brissot P et al. Haemochromatosis. Nat Rev Dis Primers 2018 Apr 5;4:18016. doi: 10.1038/nrdp.2018.16

4. Haemochromatosis reference center. https://centre-reference-fer-rennes.org/

5. Association France Fer Hémochromatose. https://www.hemochromatose.org/