

# Recent advances in genetic iron overload-related disorders

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## Recent advances in genetic iron overload-related disorders: with special focus on ferritin and *HFE*-related hemochromatosis

1. The liver iron index (liver iron concentration measured with MRI divided by age in years) is a helpful tool to interpretate liver iron concentration values in detecting major iron overload in patients with hyperferritinemia. (This thesis)
2. When *HFE* mutations are present, even in case of p.Cys282Tyr homozygosity, physicians must remain alert for the presence of liver disorders such as liver steatosis, viral or alcoholic liver disease to explain increased ferritin concentrations and elevated liver enzymes. (This thesis)
3. Despite innately low hepcidin levels, due to blockage of the BMP/SMAD pathway, patients with *HFE*-related hemochromatosis appear to be able to increase hepcidin levels in response to inflammation, probably via the JAK/STAT3 pathway. (This thesis)
4. Erythrocytapheresis is a promising treatment for patients with hereditary aceruloplasminemia to prevent peripheral and cerebral iron accumulation without resulting in a progressive anemia. (This thesis)
5. Our understanding of the principles of cellular and systemic iron homeostasis in the brain, in contrast to that of in the rest of the body, is still very limited. (Ward et al. Met. Ions Life Sci 2019)
6. Differentiation between the presence or absence of an associated iron overload in case of hyperferritinemia is essential but often proves to be complex. (Sandnes et al. J Clin Med 2021)
7. Phlebotomy should not be recommended as a treatment strategy for patients with DIOS and/or NAFLD. (Murali et al. Hepatol Res 2018)
8. The modified iron-avidity index can be used not only to predict the phenotypic expression of patients with *HFE*-related haemochromatosis in the maintenance stage but also to select those patients in whom erythrocytapheresis may be superior to venesection, when available. (This thesis, impact)
9. Trust your gut feeling, it knows what your mind hasn't figured out yet.
10. When the going gets tough, the tough get going. (Billy Ocean)