OPEN ACCESS Freely available online

Journal of Hematology & Thromboembolic Diseases

Perspective

Iron Metabolism, Biochemistry and Pathophysiology of Hemochromatosis

Sergio Della Sala*

Department of Medical Oncology, University of Western Brittany, Brest, France

DESCRIPTION

The development of genetics has significantly altered the way we perceive hemochromatosis and other iron-related disorders. Historical ideas regarding the disease, such as its intestinal origin, monogenicity, or complete phenotypic penetrance, have been challenged by new findings. A novel notion of hemochromatosis is introduced, one that is based on the hypothesis that, despite genetic differences, all known hemochromatoses has a particular metabolic abnormality: A genetically determined inability to stop extra iron from entering the circulatory pool. The central pathogenic event in all forms of hemochromatosis appears to be abnormal levels of the iron hormone, hepcidin. Depending on the protein involved and its impact on hepatic production of hepcidin, the phenotype varies, ranging from massive early-onset iron loading with severe organ disease (e.g., associated with homozygous mutations of hemojuvelin or hepcidin itself) to the milder late-onset phenotype characterizing. To analyses the effects of each hereditary hemochromatosis allele and improve our knowledge of the precise contribution of each gene to the hereditary hemochromatosis phenotype, *in vitro* and *in vivo* investigations will be required.

Iron metabolism, biochemistry and pathophysiology

The body's overall iron status typically affects how much iron is absorbed from food. Dietary intake is absorbed only to constitute for loss and, to a lesser extent, storage in the liver due to the body's effective mechanism for recycling iron. Tissue ferritin binds and stores iron in the enterocyte when dietary iron is taken up by enterocytes from the lumen of the gastrointestinal tract, preventing iron from going straight into the bloodstream. This is crucial if your body's iron reserves are already high. Iron can be moved out of enterocytes and released into plasma with the help of ferroportin. Iron is transported to several organs, most notably the bone marrow for the production of haemoglobin, once it has been bound to transferrin in the plasma. The final common metabolic pathway in hereditary haemochromatosis involves inadequate synthesis or sensitivity to hepcidin, a hepatic peptide that plays a vital role in the regulation of iron absorption. The iron export protein ferroportin interacts with hepcidin and is rendered inactive. This reduces the amount of dietary iron that enterocytes, macrophages, and hepatocytes store and release into the plasma. Hepcidin synthesis is governed by the bioavailability of iron; as a result, it rises with increased liver iron storage, preventing additional iron absorption. Similar to this, an iron deficit causes the liver to produce less hepcidin, which increases iron absorption. Due to a downstream hepcidin shortage in adult hereditary haemochromatosis brought on by faulty *HFE or TFR2* genes, increased ferroportin expression is seen at the cell surface. Parenchymal iron overload is caused by the resulting elevated plasma iron and elevated Transferrin Saturation (TS). It is yet unclear how exactly the mutant *HFE* gene affects hepcidin synthesis.

CONCLUSION

Regular blood removal (phlebotomy), which lowers blood iron levels, enhances liver function, lessens fatigue, and lowers the risk of liver and cardiac issues, is the first line of treatment for individuals with HH. Unless the blood ferritin level is low, phlebotomy is often conducted weekly for several years. At that point, it can be reduced to 3-4 times per year. Individuals who do not improve with phlebotomy or who cannot tolerate phlebotomy (due to severe anaemia or heart failure) may be given an oral iron-binding medication, albeit there is little research to support this course of treatment. A liver transplant should be considered for people with HH and end-stage liver disease or hepatocellular cancer. Although those with HH should refrain from using iron and vitamin C supplements, a low-iron (vegetarian) diet is not required. The risk of cirrhosis and hepatocellular carcinoma is increased in patients with HH who are overweight and who drink alcohol, thus they should limit or abstain from alcohol consumption.

Correspondence to: Sergio Della Sala, Department of Medical Oncology, University of Western Brittany, Brest, France, E-mail: saladella.s@edu

Received: 02-Jan-2023, Manuscript No. JHTD-23-22396; **Editor assigned:** 04-Jan-2023, Pre Qc No. JHTD-23-22396 (PQ); **Reviewed:** 18-Jan-2023, Qc No. JHTD-23-22396; **Revised:** 25-Jan-2023, Manuscript No. JHTD-23-22396 (R); **Published:** 01-Feb-2023, DOI: 10.35248/2329-8790.23.11.524.

Citation: Sala SD (2023) Iron Metabolism, Biochemistry and Pathophysiology of Hemochromatosis. J Hematol Thrombo Dis.11:524.

Copyright: © 2023 Sala SD. This is an open-access article distributed under the terms of the Creative Commons Attribution License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original author and source are credited.