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Abstract

The metabolism of iron is regulated by the peptide hormone hepcidin. Genetic alterations in the proteins involved in the signalling pathway and hepcidin transcription cause damage to the organism. Mutations and polymorphisms in the hepcidin antimicrobial peptide (HAMP), HFE, HJV, ferroportin and matriptase-2 genes influence serum hepcidin concentration. Genetic deficiency of hepcidin increases iron overload in tissues, leading to haemochromatosis. However, genetics changes in the TMPRSS6 gene promote an increase in serum hepcidin, with the development of severe anaemia and resistance to iron treatment, as observed in IRIDA. Making the flow and efflux of extracellular and intracellular iron is impossible. To date, no drug that works by inhibiting or enhancing hepcidin transcription is available, largely because of the cytotoxicity described in vitro models. The proposed therapeutic targets are still in the early stages of clinical trials, some are good candidates, such as heparin derivatives and mini-hepcidins.

Keywords: anaemia, ferroportin gene, HAMP gene, haemjuvelin gene, haemochromatosis hepcidin, HFE gene, IRIDA, iron homeostasis, polymorphism, TMPRSS6 gene, transferrin gene

1. Introduction

Disorders related to iron metabolism involve genetic alterations between hepcidin and the modulation pathway of the HAMP gene causing damage to the organism, and iron overload.
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