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Juvenile hemochromatosis: *HAMP* mutation and severe iron overload treated with phlebotomies and deferasirox

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Abstract

Juvenile hemochromatosis (JH) is a rare condition classified as an autosomal recessive disorder that leads to severe iron absorption. JH usually affects people under the age of 30 and presents symptoms such as chronic liver damage, hypogonadotropic hypogonadism, cardiac diseases and endocrine dysfunctions. The present case reports a 29-year-old Brazilian woman with JH condition due to *HAMP* mutation (c.47C>A), treated with phlebotomies and

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Cases of HH due to HJV or **HAMP mutations** are denominated type 2 HH; those ... **Juvenile Hemochromatosis** or Type 2 hereditary hemochromatosis ... JH is characterized by **severe iron overload** usually associated with liver ... man: **treatment with phlebotomy and deferasirox** Acta Haematol 2010;124 (4) 204-205 [PubMed].

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file.scirp.org/Html/8-2770095_31119.htm

Keywords: **Juvenile Hemochromatosis**; **Deferasirox**; Iron Chelation; Child; **Iron Overload** ... early and marked iron deposition in tissues and **severe** clinical course [1,2]. ... JH is determined by homozygous **mutations** in two genes, **HAMP** and HJV, ... The mainstay of **treatment** in HJV is **phlebotomy**, but oral chelation could be a ...

[\[PDF\] Diagnosis and treatment of non-HFE-haemochromatosis](secure.esh.org/ChaptersIronHB/IRON2009_CAP.24(570-583).pdf)

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haemojuvelin (5), type 2B is due to **mutations** of **HAMP**, the hepcidin encoding gene. (6). TfR2 and **juvenile haemochromatosis** share with HFE- haemochromatosis ... finding of the same **iron overload** phenotype in the presence of a discordant ... **severe** disorders. 4. **Treatment**. 4.1 **Phlebotomy**. There is limited experience ...

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1 Feb 2012 ... Keywords: hemochromatosis, primary iron overload, HFE, high-resolution ... 4 additional disorders of primary iron overload: juvenile hemochromatosis (JH) or ... In contrast, mutations in HAMP are a very rare cause of JH: p. responsive to treatment and had more severe complications due to iron overload.

[Efficacy of deferasirox for the treatment of iron overload in a child ...](#)

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Keywords: Juvenile Hemochromatosis; Deferasirox; Iron Chelation; Child; Iron Overload ... early and marked iron deposition in tissues and severe clinical course [1,2]. ... JH is determined by homozygous mutations in two genes, HAMP and HJV, ... The mainstay of treatment in HJV is phlebotomy, but oral chelation could be a ...

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