HJV gene

hemojuvelin BMP co-receptor

Normal Function

The *HJV* gene provides instructions for making a protein called hemojuvelin. This protein is made in the liver, heart, and muscles used for movement (skeletal muscles). Hemojuvelin plays a role maintaining proper iron levels in the body by controlling the levels of another protein called hepcidin. Hepcidin is necessary for maintaining an appropriate balance of iron (iron homeostasis) in the body.

Health Conditions Related to Genetic Changes

Hereditary hemochromatosis

More than 30 *HJV* gene mutations have been found to cause type 2 hemochromatosis, a form of hereditary hemochromatosis that begins during childhood or adolescence. Hereditary hemochromatosis is a disorder that causes the body to absorb too much iron from the diet. The excess iron accumulates in, and eventually damages, the body's tissues and organs.

Most *HJV* gene mutations change one of the protein building blocks (amino acids) used to make hemojuvelin. Most frequently, the amino acid glycine is replaced by the amino acid valine at protein position 320 (written as Gly320Val or G320V). Other mutations create a premature stop signal in the instructions for making the hemojuvelin protein resulting in an abnormally small protein.

Mutations in the *HJV* gene lead to an altered hemojuvelin protein that cannot function properly. Without adequate hemojuvelin, hepcidin levels are reduced and iron homeostasis is disturbed. As a result, too much iron is absorbed during digestion, which leads to iron overload and damage to tissues and organs in the body that is found in hereditary hemochromatosis.

Other Names for This Gene

- hemochromatosis type 2 (juvenile)
- HFE2
- HFE2A
- JH

- RGM domain family, member C
- RGMC
- RGMC_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of HJV (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=148738[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28HFE2%5BTIAB%5D%29+OR+%28hemochromatosis+type+2%5BTIAB%5D%29+OR+%28hemochromatosis+AND+1q%5BTIAB%5D%29%29+OR+%28%28hemojuvelin%5BTIAB%5D%29+OR+%28HFE2A%5BTIAB%5D%29+OR+%28HJV%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- HEMOJUVELIN BMP CORECEPTOR; HJV (https://omim.org/entry/608374)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/148738)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=HJV[gene])

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Genomic Location

The *HJV* gene is found on chromosome 1 (https://medlineplus.gov/genetics/chromosome/1/).

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