HAL gene

histidine ammonia-lyase

Normal Function

The *HAL* gene provides instructions for making an enzyme called histidase. Histidase breaks down the amino acid histidine, a building block of most proteins. Histidase is active (expressed) primarily in the liver and the skin. This enzyme breaks down histidine to a molecule called urocanic acid. In the liver, urocanic acid is broken down to form another amino acid called glutamic acid. In the skin, urocanic acid is involved in the response to ultraviolet (UV) light.

Health Conditions Related to Genetic Changes

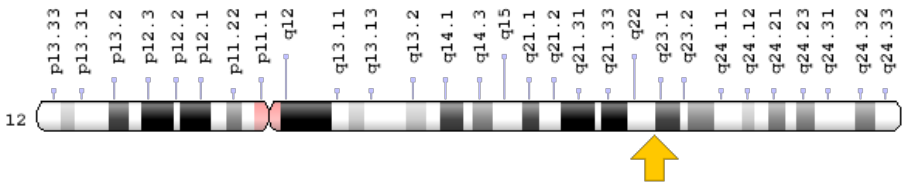
Histidinemia

At least four mutations in the *HAL* gene have been found to cause histidinemia. All of these mutations change single amino acids in the histidase enzyme. These mutations are thought to decrease or eliminate enzyme activity, resulting in an inability to break down histidine. Histidine that is not broken down accumulates in the blood, but it typically causes no health problems.

Chromosomal Location

Cytogenetic Location: 12q23.1, which is the long (q) arm of chromosome 12 at position 23.1

Molecular Location: base pairs 95,972,662 to 95,996,365 on chromosome 12 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- HIS
- histidase

- HSTD
- HUTH_HUMAN

Additional Information & Resources

Educational Resources

- Biochemistry (fifith edition, 2002): Histidine degradation
  https://www.ncbi.nlm.nih.gov/books/NBK22453/figure/A3247/

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28HAL%5BTIAB%5D%29+OR+%28histidase%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- HISTIDINE AMMONIA-LYASE
  http://omim.org/entry/609457

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=HAL%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:4806
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:3034
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/3034
- UniProt
  https://www.uniprot.org/uniprot/P42357

Sources for This Summary

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https://ghr.nlm.nih.gov/gene/HAL

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