

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.

Other Names for This Gene

- HIS
- histidase
- HSTD
- HUTH_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of HAL ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3034\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3034[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?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%29%29%29>)

Catalog of Genes and Diseases from OMIM

- HISTIDINE AMMONIA-LYASE; HAL (<https://omim.org/entry/609457>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/3034>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=HAL\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=HAL[gene]))

References

- Eckhart L, Schmidt M, Mildner M, Mlitz V, Abtin A, Ballaun C, Fischer H, MrassP, Tschachler E. Histidase expression in human epidermal keratinocytes: regulation by differentiation status and all-trans retinoic acid. *J Dermatol Sci.* 2008 Jun;50(3):209-15. doi: 10.1016/j.jdermsci.2007.12.009. Epub 2008 Feb 15. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/18280705>)
- Kawai Y, Moriyama A, Asai K, Coleman-Campbell CM, Sumi S, Morishita H, SuchiM. Molecular characterization of histidinemia: identification of four missense mutations in the histidase gene. *Hum Genet.* 2005 Apr;116(5):340-6. doi: 10.1007/s00439-004-1232-5. Epub 2005 Jan 27. Erratum In: *Hum Genet.* 2005 Dec; 118(3-4):531-2. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/15806399>)
- Suchi M, Sano H, Mizuno H, Wada Y. Molecular cloning and structural characterization of the human histidase gene (HAL). *Genomics.* 1995 Sep 1; 29(1):98-104. doi: 10.1006/geno.1995.1219. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/8530107>)

Genomic Location

The *HAL* gene is found on chromosome 12 (<https://medlineplus.gov/genetics/chromosome/12/>).

Last updated August 1, 2009