

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, which acts as a building block for many different proteins. Histidase is active (expressed) primarily in the liver and the skin. This enzyme breaks down histidine to a molecule called urocanic acid. During digestion, urocanic acid is broken down in the liver to form another amino acid called glutamate. In the skin, urocanic acid is involved in the response to ultraviolet (UV) light.

Health Conditions Related to Genetic Changes

Histidinemia

Several variants (also called mutations) in the *HAL* gene have been found to cause an inherited condition called histidinemia. People with histidinemia have high levels of histidine in the blood, urine, and the fluid that surrounds the brain and spinal cord (cerebrospinal fluid). Most of the variants change single amino acids in the histidase enzyme and are thought to decrease or eliminate the enzyme's activity. A decrease in histidase activity interferes with the body's ability to break down histidine. Histidine then builds up in the blood, urine, and cerebrospinal fluid. Rarely, people with histidinemia have intellectual disabilities, learning disabilities, or behavioral complications. Having a medical complication during or soon after birth might increase the risk of developmental problems in people with histidinemia.

Other Names for This Gene

- HIS
- histidase
- HSTD

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>)

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]))

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Genomic Location

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

Last updated June 3, 2024