Histidinemia

Subjects: Genetics & Heredity
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Histidinemia is an inherited condition characterized by elevated blood levels of the amino acid histidine, a building block of most proteins. Histidinemia is caused by the shortage (deficiency) of the enzyme that breaks down histidine. Histidinemia typically causes no health problems, and most people with elevated histidine levels are unaware that they have this condition.

Keywords: genetic conditions

1. Introduction

The combination of histidinemia and a medical complication during or soon after birth (such as a temporary lack of oxygen) might increase a person's chances of developing intellectual disability, behavioral problems, or learning disorders.

2. Frequency

Estimates of the incidence of histidinemia vary widely, ranging between 1 in 8,600 to 1 in 90,000 people.

3. Causes

Histidinemia is caused by mutations in the *HAL* gene, which provides instructions for making an enzyme called histidase. Histidase breaks down histidine to a molecule called urocanic acid. Histidase is active (expressed) primarily in the liver and the skin.

HAL gene mutations lead to the production of a histidase enzyme that cannot break down histidine, which results in elevated levels of histidine in the blood and urine. These increased levels of histidine do not appear to have any negative effects on the body.

3.1. The gene associated with Histidinemia

- HAL

4. Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

5. Other Names for This Condition

- HAL deficiency
- HIS deficiency
- histidase deficiency
- histidine ammonia-lyase deficiency
- hyperhistidinemia

References

1. Kawai Y, Moriyama A, Asai K, Coleman-Campbell CM, Sumi S, Morishita H, SuchiM. Molecular characterization of histidinemia: identification of four missensemutations in the histidase gene. Hum Genet. 2005 Apr;116(5):340-6.
2. Taylor RG, Levy HL, McInnes RR. Histidase and histidinemia. Clinical andmolecular considerations. Mol Biol Med. 1991 Feb;8(1):101-16. Review.
3. Virmani K, Widhalm K. Histidinemia: a biochemical variant or a disease? J AmColl Nutr. 1993 Apr;12(2):115-24. Review.
