HAX1 gene

HCLS1 associated protein X-1

Normal Function

The *HAX1* gene provides instructions for making a protein called HS-1-associated protein X-1 (HAX-1). This protein is involved in the regulation of apoptosis, which is the process by which cells self-destruct when they are damaged or no longer needed. Apoptosis is a common process that occurs throughout life. The HAX-1 protein is also thought to be involved in cell movement (migration). The HAX-1 protein is found primarily in the mitochondria, the energy-producing centers within cells.

Different versions of the HAX-1 protein can be produced from the *HAX1* gene by a mechanism called alternative splicing. This mechanism produces different version of the protein by cutting and rearranging the genetic instructions in different ways. The purpose of these multiple versions of the HAX-1 protein is unclear.

Health Conditions Related to Genetic Changes

Severe congenital neutropenia

At least 10 mutations in the *HAX1* gene have been found to cause severe congenital neutropenia, a condition characterized by a shortage (deficiency) of neutrophils. Neutrophils are a type of white blood cell that play a role in inflammation and in fighting infection. Most of the mutations that cause severe congenital neutropenia change single protein building blocks (amino acids) in the HAX-1 protein. *HAX1* gene mutations that cause severe congenital neutropenia result in the production of a nonfunctional HAX-1 protein. A lack of functional HAX-1 protein disrupts regulation of apoptosis, leading to the premature death of neutrophils. A deficiency of neutrophils results in recurrent infections, episodes of inflammation, and other immune problems in people with severe congenital neutropenia.

People with certain *HAX1* gene mutations have neurological problems such as seizures and developmental delay in addition to severe congenital neutropenia. These mutations disrupt two of the alternatively spliced versions of the HAX-1 protein. The gene mutations that lead to severe congenital neutropenia alone disrupt only one version of the HAX-1 protein. It is unclear how nonfunctional HAX-1 proteins cause neurological features seen in some affected individuals.

Other Names for This Gene

- HAX-1
- HAX1_HUMAN
- HCLS1-associated protein X-1
- HCLSBP1
- HS1 binding protein
- HS1-associating protein X-1
- HS1-binding protein 1
- HS1BP1
- HSP1BP-1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of HAX1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=10456[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28HAX1%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+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- HCLS1-ASSOCIATED PROTEIN X1; HAX1 (https://omim.org/entry/605998)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/10456)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=HAX1[gene])

References

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Genomic Location

The *HAX1* gene is found on chromosome 1 (https://medlineplus.gov/genetics/chromosome/1/).

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