



## AHDC1 gene

AT-hook DNA binding motif containing 1

### Normal Function

The *AHDC1* gene provides instructions for making a protein whose function is not known. The AHDC1 protein is found in the nucleus of cells, and a region of the protein is thought to allow it to attach (bind) to DNA. Based on its location and possible DNA-binding ability, researchers suspect the protein may help control the activity of other genes.

### Health Conditions Related to Genetic Changes

#### Xia-Gibbs syndrome

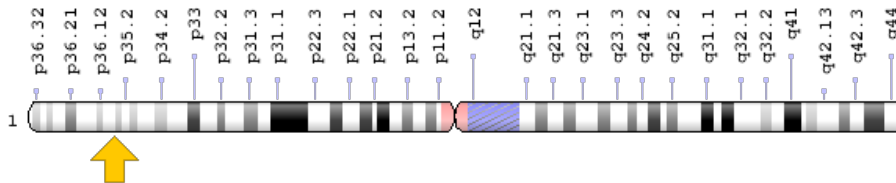
More than 25 mutations in the *AHDC1* gene have been found to cause Xia-Gibbs syndrome, a neurological disorder characterized by intellectual disability and delayed speech development. A variety of other signs and symptoms can also occur in this disorder, such as weak muscle tone, growth impairment, and unusual facial features. Most of the *AHDC1* gene mutations involved in Xia-Gibbs syndrome lead to production of abnormally short AHDC1 proteins. The effects of these changes in cells are unclear. The shortened proteins may be quickly broken down or be unable to function. Or, the abnormal proteins may interfere with the function of AHDC1 proteins produced from the normal copy of the gene. Some people with Xia-Gibbs syndrome have a mutation that removes (deletes) the *AHDC1* gene (and other nearby genes), although it is unclear if such mutations cause the condition.

Researchers suspect that a reduction in the amount of functional AHDC1 protein prevents normal brain development, leading to intellectual disability, speech problems, and other neurological features of Xia-Gibbs syndrome. Abnormal development of other body systems caused by a shortage of AHDC1 protein may account for additional signs and symptoms of the condition.

## Chromosomal Location

Cytogenetic Location: 1p36.11-p35.3, which is the short (p) arm of chromosome 1 between positions 36.11 and 35.3

Molecular Location: base pairs 27,534,245 to 27,604,178 on chromosome 1 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- AT-HOOK DNA-BINDING MOTIF-CONTAINING PROTEIN 1

## Additional Information & Resources

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28AHDC1%5BTIAB%5D%29+OR+%28AT-hook+DNA+binding+motif+containing+1%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+%22last+3600+days%22%5Bdp%5D>

### Catalog of Genes and Diseases from OMIM

- AT-HOOK DNA-BINDING MOTIF-CONTAINING PROTEIN 1  
<http://omim.org/entry/615790>

### Research Resources

- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=AHDC1%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/data/gene-symbol-report/#!/hgnc\\_id/HGNC:25230](https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:25230)
- Monarch Initiative  
<https://monarchinitiative.org/gene/NCBIGene:27245>

- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/27245>
- UniProt  
<https://www.uniprot.org/uniprot/Q5TGY3>

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