



ADNP gene

activity dependent neuroprotector homeobox

Normal Function

The *ADNP* gene provides instructions for making a protein that helps control the activity (expression) of other genes through a process called chromatin remodeling. Chromatin is the network of DNA and protein that packages DNA into chromosomes. The structure of chromatin can be changed (remodeled) to alter how tightly DNA is packaged. Chromatin remodeling is one way gene expression is regulated during development; when DNA is tightly packed, gene expression is lower than when DNA is loosely packed. As part of the remodeling process, the ADNP protein attaches to DNA and interacts with groups of proteins called SWI/SNF complexes, which direct changes in the structure of chromatin.

By regulating gene expression, the ADNP protein is involved in many aspects of development. It is particularly important for regulation of genes involved in normal brain development, and it likely controls the activity of genes that direct the development and function of other body systems.

Health Conditions Related to Genetic Changes

ADNP syndrome

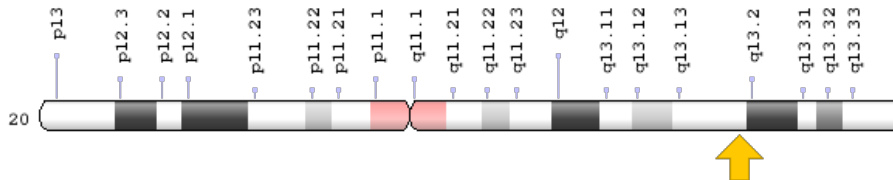
At least 22 *ADNP* gene mutations have been found to cause *ADNP* syndrome. This condition features intellectual disability and autism spectrum disorder, which is characterized by impaired communication and social interactions. Affected individuals can also have distinctive facial features and a wide variety of other signs and symptoms. Most *ADNP* gene mutations are thought to lead to the production of an abnormally short ADNP protein. Although it is unclear how these genetic changes cause *ADNP* syndrome, researchers speculate that the abnormally short protein can attach to DNA but cannot interact with SWI/SNF complexes. As a result, chromatin remodeling is impaired. Disturbance of this process alters the activity of many genes and disrupts the development or function of several of the body's tissues and organs, including the brain. These changes likely explain the intellectual disability, autism spectrum disorder, and other diverse signs and symptoms of *ADNP* syndrome.

Autism spectrum disorder

Chromosomal Location

Cytogenetic Location: 20q13.13, which is the long (q) arm of chromosome 20 at position 13.13

Molecular Location: base pairs 50,888,918 to 50,931,437 on chromosome 20 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- activity-dependent neuroprotective protein
- activity-dependent neuroprotector
- activity-dependent neuroprotector homeobox protein
- ADNP homeobox 1
- ADNP1
- HVDAS
- KIAA0784
- MRD28

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): ATP-driven Chromatin Remodeling Machines Change Nucleosome Structure
<https://www.ncbi.nlm.nih.gov/books/NBK26834/#A644>
- Molecular Biology of the Cell (fourth edition, 2002): Chromosomal DNA and Its Packaging in the Chromatin Fiber
<https://www.ncbi.nlm.nih.gov/books/NBK26834/>
- Undiagnosed Diseases Network: Gene Page
<https://undiagnosed.hms.harvard.edu/genes/adnp/>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ADNP%5BTIAB%5D%29+OR+%28activity+dependent+neuroprotector+homeobox%5BTIAB%5D%29%29+OR+%28%28ADNP+homeobox+1%5BTIAB%5D%29+OR+%28activity-dependent+neuroprotective+protein%5BTIAB%5D%29+OR+%28activity-dependent+neuroprotector+homeobox+protein%5BTIAB%5D%29+OR+%28activity-dependent+neuroprotector%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- ACTIVITY-DEPENDENT NEUROPROTECTOR HOMEBOX
<http://omim.org/entry/611386>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_ADNP.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=ADNP%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report#!/hgnc_id/HGNC:15766
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:23394>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/23394>
- UniProt
<https://www.uniprot.org/uniprot/Q9H2P0>

Sources for This Summary

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