

# ADNP Related Syndrome

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## ADNP gene

activity dependent neuroprotector homeobox

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### 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.

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### Autism spectrum disorder

[MedlinePlus Genetics](#) provides information about Autism spectrum disorder

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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

Tests Listed in the Genetic Testing Registry

Tests of ADNP

Scientific Articles on PubMed

PubMed

## Catalog of Genes and Diseases from OMIM

### ACTIVITY-DEPENDENT NEUROPROTECTOR HOMEODOMAIN; ADNP

## Gene and Variant Databases

NCBI Gene

ClinVar

## References

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Genomic LocationThe ADNP gene is found on chromosome 20.

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