ADNP Related Syndrome

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ADNP syndrome

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Description

ADNP syndrome is a condition that causes a wide variety of signs and symptoms. Its hallmark features are intellectual disability and autism spectrum disorder, which is characterized by impaired communication and social interaction. Affected individuals also have distinctive facial features and abnormalities of multiple body systems.Individuals with ADNP syndrome have mild to severe intellectual disability and delayed development of speech and motor skills such as sitting and walking. Some affected individuals are never able to speak. People with this disorder exhibit characteristics typical of autism spectrum disorder, including repetitive behaviors and difficulty with social interactions. ADNP syndrome is also associated with mood disorders or behavioral problems, such as anxiety, temper tantrums, attention-deficit/hyperactivity disorder (ADHD), obsessive-compulsive disorder, or sleep problems.Many people with ADNP syndrome have distinctive facial features, which most commonly include a prominent forehead, a high hairline, outside corners of the eyes that point upward or downward (upslanting or downslanting palpebral fissures), droopy eyelids (ptosis), a broad nasal bridge, and a thin upper lip. These individuals may

also have unusually shaped ears or hand and finger abnormalities. Eye and vision abnormalities, such as eyes that do not point in the same direction (strabismus) and farsightedness (hyperopia), also occur in ADNP syndrome. Some people with this condition have early appearance (eruption) of primary (baby) teeth.Some people with ADNP syndrome have weak muscle tone (hypotonia) and feeding difficulties in infancy. They may also have digestive system problems, such as backflow of stomach acids into the esophagus (gastroesophageal reflux), vomiting, and constipation. Other features that occur in ADNP syndrome include obesity, seizures, and heart abnormalities.

Frequency

The prevalence of ADNP syndrome is unknown. It is estimated to account for 0.17 percent of all cases of autism spectrum disorder, making it one of the most common genetic causes of this condition.

Causes

ADNP syndrome is caused by mutations in the ADNP gene. The protein produced from this gene 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.By regulating gene expression, the ADNP protein is involved in many aspects of growth and 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.Although it is unclear how mutations in the ADNP gene affect ADNP protein function, researchers suggest that the mutations result in abnormal chromatin remodeling. Disturbance of this process alters the activity of many genes and disrupts 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.

Learn more about the gene associated with ADNP syndrome

ADNP

Inheritance

ADNP syndrome is not inherited. It results from new (de novo) mutations in the ADNP gene that occur during the formation of reproductive cells (eggs or sperm) or in early embryonic development. The condition occurs in people with no history of the disorder in their family.

Other Names for This Condition

ADNP-related intellectual disability and autism spectrum disorder ADNP-related multiple congenital

anomalies-intellectual disability-autism spectrum disorder Helsmoortel-van der Aa syndrome

HVDAS Mental retardation, autosomal dominant 28 MRD28

Additional Information & Resources

Genetic Testing Information

Genetic Testing Registry: ADNP-related multiple congenital anomalies - intellectual disability - autism spectrum disorder

Genetic and Rare Diseases Information Center

ADNP syndrome

Patient Support and Advocacy Resources

National Organization for Rare Disorders (NORD)

Clinical Trials

ClinicalTrials.gov

Catalog of Genes and Diseases from OMIM

HELSMOORTEL-VAN DER AA SYNDROME; HVDAS

Scientific Articles on PubMed

PubMed

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