Arginine vasopressin deficiency

Description

Arginine vasopressin deficiency (previously called neurohypophyseal diabetes insipidus) is a disorder of water balance. The body normally balances fluid intake by releasing fluid in urine. However, people with arginine vasopressin deficiency produce an excessive amount of urine (polyuria), which depletes the amount of water in the body. This water loss also leads to excessive thirst (polydipsia).

People with arginine vasopressin deficiency can quickly become dehydrated if they do not drink enough water. Dehydration can cause dizziness and fatigue. Prolonged dehydration can lead to confusion, low blood pressure, seizures, and coma. People with this condition often develop high levels of sodium in the blood (hypernatremia) due to dehydration. Repeated cycles of dehydration can cause long-term health problems, particularly in children.

Arginine vasopressin deficiency can be either acquired or familial. The acquired form occurs when the brain is damaged due to head injuries, brain tumors, or other events, and this form can occur at any time during life. The familial form is caused by genetic changes; its signs and symptoms usually become apparent in childhood and worsen over time.

Researchers have recommended using the condition name arginine vasopressin deficiency because the previous name, neurohypophyseal diabetes insipidus, was often confused with a much more common disorder called diabetes mellitus. Arginine vasopressin deficiency and diabetes mellitus are separate disorders with different features, causes, and treatment.

Frequency

Arginine vasopressin deficiency occurs in approximately 1 in 25,000 individuals. The acquired form is much more common than the familial form.

Causes

The familial form of arginine vasopressin deficiency is caused by variants (also called mutations) in the *AVP* gene. This gene provides instructions for making a hormone called arginine vasopressin (AVP), which is sometimes also called antidiuretic hormone (ADH). This hormone, which is produced and stored in the brain, helps control the body's

water balance.

AVP works with the kidneys to manage the balance between fluid reabsorption and fluid release. Normally, when a person's fluid intake is low or when a lot of fluid is lost (for example, through sweating), the brain releases more AVP into the bloodstream. High levels of this hormone direct the kidneys to reabsorb more water and to make less urine. When fluid intake is adequate, the brain releases less AVP.

Variants in the *AVP* gene result in the production of an AVP hormone that does not get released from brain cells. Without this hormone, the kidneys do not reabsorb water as they should, and the body makes excessive amounts of urine. These problems with water balance are characteristic of arginine vasopressin deficiency.

The acquired form of arginine vasopressin deficiency occurs when the areas of the brain that produce or store AVP are damaged by head injuries, brain tumors, brain surgery, certain diseases and infections, or bleeding in the brain. A loss of AVP disrupts the body's water balance, leading to excessive urine production and the other features of the disorder.

In 30 to 50 percent of all cases of arginine vasopressin deficiency, the cause of the disorder is unknown. Studies suggest that some of these cases may be caused by a malfunctioning immune system that attacks the body's own tissues and organs.

Learn more about the gene associated with Arginine vasopressin deficiency

- AVP

Inheritance

Familial arginine vasopressin deficiency is almost always inherited in an autosomal dominant pattern, which means that one copy of the altered *AVP* gene in each cell is sufficient to cause the disorder.

In a few affected families, familial arginine vasopressin deficiency has had an autosomal recessive pattern of inheritance. Autosomal recessive inheritance means that both copies of the gene in each cell must have a variant to cause the disorder. The parents of an individual with an autosomal recessive condition each carry one copy of the altered gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- Central diabetes insipidus
- Diabetes insipidus secondary to vasopressin deficiency
- Diabetes insipidus, central
- Diabetes insipidus, neurogenic
- Diabetes insipidus, neurohypophyseal

- Diabetes insipidus, pituitary
- Neurohypophyseal diabetes insipidus
- Pituitary diabetes insipidus
- Vasopressin defective diabetes insipidus
- Vasopressin deficiency

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Neurohypophyseal diabetes insipidus (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0342394/)

Genetic and Rare Diseases Information Center

- Central diabetes insipidus (https://rarediseases.info.nih.gov/diseases/6015/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Neurohypophyseal diabetes insipidus%22)

Catalog of Genes and Diseases from OMIM

- DIABETES INSIPIDUS, NEUROHYPOPHYSEAL (https://omim.org/entry/125700)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=(Diabetes+Insipidus,+Neurogenic%5BMAJR%5D)+AND+((neurohypophyseal+diabetes+insipidus%5BTIAB%5D)+OR+(central+diabetes+insipidus%5BTIAB%5D)+OR+(vasopressin+deficiency%5BTIAB%5D))+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+5000+days%22%5Bdp%5D)

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