

AVP Gene

Subjects: Genetics & Heredity

Contributor: Peter Tang

Arginine vasopressin

Keywords: genes

1. Normal Function

The *AVP* gene provides instructions for making a hormone called vasopressin or antidiuretic hormone (ADH). ADH starts out as a larger molecule called a preprohormone, which is cut (cleaved) and modified to produce the active hormone and several related proteins. The preprohormone is made in a region of the brain called the hypothalamus. It is then transported to the nearby pituitary gland, where active ADH is stored until it is needed.

The major function of ADH is to help control the body's water balance by determining how much water is excreted in urine. Normally, when a person's fluid intake is low or when a lot of fluid is lost (for example, through sweating), the pituitary gland releases more ADH 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 pituitary gland releases less ADH. Lower levels of this hormone cause the kidneys to reabsorb less water and to make more urine.

2. Health Conditions Related to Genetic Changes

2.1. Neurohypophyseal diabetes insipidus

At least 60 mutations in the *AVP* gene have been found to cause neurohypophyseal diabetes insipidus. Most of these mutations change single protein building blocks (amino acids) in the preprohormone or result in the production of an abnormally short version of this molecule. Studies suggest that the altered preprohormone becomes trapped inside the cells where it is produced instead of being transported to the pituitary gland. As the defective molecule builds up over time, it damages and ultimately kills these cells. The resulting shortage of ADH prevents the kidneys from reabsorbing water as they should, and the body makes excessive amounts of urine. These problems with water balance are characteristic of neurohypophyseal diabetes insipidus.

3. Other Names for This Gene

- ADH
 - antidiuretic hormone
 - ARVP
 - AVP-NP11
 - AVRP
 - NEU2_HUMAN
 - neurohypophyseal
 - vasopressin-neurophysin 2-copeptin
 - vasopressin-neurophysin 2-copeptin preproprotein
 - vasopressin-neurophysin II-copeptin
 - VP
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