



AQP2 gene

aquaporin 2

Normal Function

The *AQP2* gene provides instructions for making a protein called aquaporin 2. This protein forms a channel that carries water molecules across cell membranes. It is found in the kidneys in structures called collecting ducts, which are a series of small tubes that reabsorb water from the kidneys into the bloodstream.

The aquaporin 2 water channel plays an essential role in maintaining the body's water balance. The placement of these channels is controlled by a hormone called vasopressin or antidiuretic hormone (ADH). When a person's fluid intake is low or when a lot of fluid is lost (for example, through sweating), the body produces more ADH. This hormone triggers chemical reactions that ultimately insert aquaporin 2 water channels into the membrane of collecting duct cells. These channels allow water to be reabsorbed into the bloodstream, which makes the urine more concentrated. When fluid intake is adequate, less ADH is produced. Without signals from ADH, aquaporin 2 water channels are removed from the membrane of collecting duct cells. At these times, less water is reabsorbed into the bloodstream and the urine is more dilute.

Health Conditions Related to Genetic Changes

Nephrogenic diabetes insipidus

At least 40 mutations in the *AQP2* gene have been identified in people with nephrogenic diabetes insipidus.

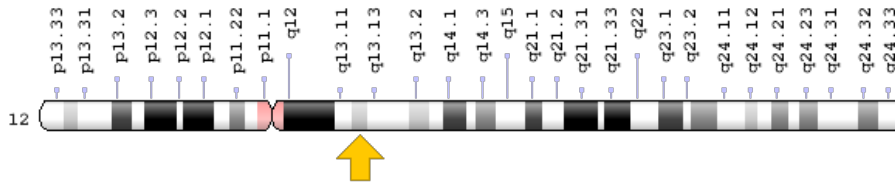
Most of the known *AQP2* gene mutations cause the aquaporin 2 protein to be misfolded into an incorrect 3-dimensional shape. The misfolded protein is trapped within the cell, where it is unable to reach the cell membrane to transport water molecules. A few mutations result in the production of functional aquaporin 2 water channels, but these channels are misrouted within the cell and do not reach the cell membrane.

If aquaporin 2 water channels are not inserted into the membrane of collecting duct cells, the kidneys are unable to respond to signals from ADH. As a result, collecting ducts do not reabsorb water as they should, and the body makes excessive amounts of urine. These problems with water balance are characteristic of nephrogenic diabetes insipidus.

Chromosomal Location

Cytogenetic Location: 12q13.12, which is the long (q) arm of chromosome 12 at position 13.12

Molecular Location: base pairs 49,950,741 to 49,958,881 on chromosome 12 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ADH water channel
- AQP-2
- AQP-CD
- AQP2_HUMAN
- aquaporin-2
- aquaporin 2 (collecting duct)
- aquaporin-CD
- collecting duct water channel protein
- MGC34501
- water-channel aquaporin 2
- water channel protein for renal collecting duct
- WCH-CD

Additional Information & Resources

Educational Resources

- Colorado State University: Antidiuretic Hormone (Vasopressin)
<http://www.vivo.colostate.edu/hbooks/pathphys/endocrine/hypopit/adh.html>
- Molecular Cell Biology (2000): Water Channels Are Necessary for Bulk Flow of Water Across Cell Membranes
<https://www.ncbi.nlm.nih.gov/books/NBK21739/#A4147>

Clinical Information from GeneReviews

- Nephrogenic Diabetes Insipidus
<https://www.ncbi.nlm.nih.gov/books/NBK1177>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28AQP2%5BTIAB%5D%29+OR+%28aquaporin+2%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- AQUAPORIN 2
<http://omim.org/entry/107777>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
<http://atlasgeneticsoncology.org/Genes/AQP2ID52230ch12q13.html>
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=AQP2%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report#!/hgnc_id/HGNC:634
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:359>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/359>
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