ATP6V1B1 gene

ATPase H+ transporting V1 subunit B1

Normal Function

The *ATP6V1B1* gene provides instructions for making a part (subunit) of a large protein complex known as vacuolar H^+-ATPase (V-ATPase). V-ATPases are a group of similar complexes that act as pumps to move positively charged hydrogen atoms (protons) across membranes. Because acids are substances that can "donate" protons to other molecules, this movement of protons helps regulate the relative acidity (pH) of cells and their surrounding environment. Tight control of pH is necessary for most biological reactions to proceed properly.

The V-ATPase that includes the subunit produced from the *ATP6V1B1* gene is found in the inner ear and in nephrons, which are the functional structures within the kidneys. The kidneys filter waste products from the blood and remove them in urine. They also reabsorb needed nutrients and release them back into the blood. Each nephron consists of two parts: a renal corpuscle (also known as a glomerulus) that filters the blood, and a renal tubule that reabsorbs substances that are needed and eliminates unneeded substances in urine. The V-ATPase is involved in regulating the amount of acid that is removed from the blood into the urine, and also in maintaining the proper pH of the fluid in the inner ear (endolymph).

Health Conditions Related to Genetic Changes

Renal tubular acidosis with deafness

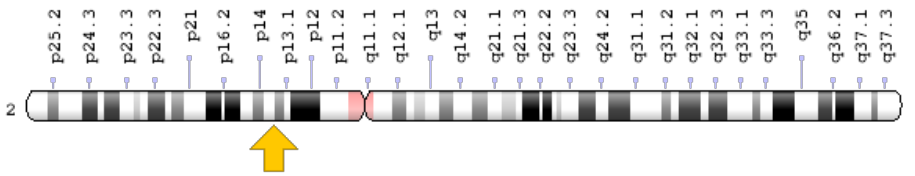
More than 25 *ATP6V1B1* gene mutations have been identified in people with renal tubular acidosis with deafness, a disorder involving excess acid in the blood (metabolic acidosis), bone weakness, and hearing loss caused by changes in the inner ear (sensorineural hearing loss).

Mutations in the *ATP6V1B1* gene impair the function of the V-ATPase proton pump. As a result, the kidneys are less able to control the acidity of the blood, which leads to bone weakness caused by loss of bone minerals (demineralization) and other consequences of metabolic acidosis. The body's capability to control the pH of the fluid in the inner ear is also impaired, resulting in sensorineural hearing loss.

Chromosomal Location

Cytogenetic Location: 2p13.3, which is the short (p) arm of chromosome 2 at position 13.3

Molecular Location: base pairs 70,935,900 to 70,965,431 on chromosome 2 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ATP6B1
- ATPase, H+ transporting, lysosomal 56/58kDa, V1 subunit B1
- endomembrane proton pump 58 kDa subunit
- H(+)-transporting two-sector ATPase, 58kD subunit
- H+-ATPase beta 1 subunit
- RTA1B
- V-ATPase B1 subunit
- V-ATPase subunit B 1
- V-type proton ATPase subunit B, kidney isoform
- vacuolar proton pump 3
- vacuolar proton pump subunit B 1
- vacuolar proton pump, subunit 3
- VATB
- VMA2
- VPP3

Additional Information & Resources

Educational Resources

- Molecular Cell Biology (fourth edition, 2000): Active Transport by ATP-Powered Pumps
  https://www.ncbi.nlm.nih.gov/books/NBK21481/

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28ATP6V1B1%5BTIAB%5D%29+OR+%28%28VATB%5BTIAB%5D%29+OR+%28VMA2%5BTIAB%5D%29+OR+%28ATP6B1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- ATPase, H+ TRANSPORTING, LYSOSOMAL, 56/58-KD, V1 SUBUNIT B, ISOFORM 1
  http://omim.org/entry/192132

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ATP6V1B1%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:853
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:525
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/525
- UniProt
  https://www.uniprot.org/uniprot/P15313

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