

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.

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

Tests Listed in the Genetic Testing Registry

- Tests of ATP6V1B1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=525\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=525[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?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%29>)

Catalog of Genes and Diseases from OMIM

- ATPase, H⁺ TRANSPORTING, LYSOSOMAL, 56/58-KD, V1 SUBUNIT B, ISOFORM 1; ATP6V1B1 (<https://omim.org/entry/192132>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/525>)

- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ATP6V1B1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ATP6V1B1[gene]))

References

- Alper SL. Familial renal tubular acidosis. *J Nephrol.* 2010 Nov-Dec;23 Suppl16:S57-76. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/21170890>)
- Andreucci E, Bianchi B, Carboni I, Lavoratti G, Mortilla M, Fonda C, Bigozzi M, Genuardi M, Giglio S, Pela I. Inner ear abnormalities in four patients with dRTA and SNHL: clinical and genetic heterogeneity. *Pediatr Nephrol.* 2009 Nov;24(11):2147-53. doi: 10.1007/s00467-009-1261-3. Epub 2009 Jul 29. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/19639346>)
- Batlle D, Haque SK. Genetic causes and mechanisms of distal renal tubular acidosis. *Nephrol Dial Transplant.* 2012 Oct;27(10):3691-704. doi:10.1093/ndt/gfs442. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/23114896>)
- Gil H, Santos F, Garcia E, Alvarez MV, Ordonez FA, Malaga S, Coto E. Distal RTA with nerve deafness: clinical spectrum and mutational analysis in five children. *Pediatr Nephrol.* 2007 Jun;22(6):825-8. doi: 10.1007/s00467-006-0417-7. Epub 2007 Jan 11. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/17216496>)
- Mohebbi N, Vargas-Poussou R, Hegemann SC, Schuknecht B, Kistler AD, Wuthrich RP, Wagner CA. Homozygous and compound heterozygous mutations in the ATP6V1B1 gene in patients with renal tubular acidosis and sensorineural hearing loss. *Clin Genet.* 2013 Mar;83(3):274-8. doi: 10.1111/j.1399-0004.2012.01891.x. Epub 2012 May 11. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/22509993>)
- Nikali K, Vanegas JJ, Burley MW, Martinez J, Lopez LM, Bedoya G, Wrong OM, Povey S, Unwin RJ, Ruiz-Linares A. Extensive founder effect for distal renal tubular acidosis (dRTA) with sensorineural deafness in an isolated South American population. *Am J Med Genet A.* 2008 Oct 15;146A(20):2709-12. doi:10.1002/ajmg.a.32495. No abstract available. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/18798332>)
- Sethi SK, Singh N, Gil H, Bagga A. Genetic studies in a family with distal renal tubular acidosis and sensorineural deafness. *Indian Pediatr.* 2009 May;46(5):425-7. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/19478356>)
- Stover EH, Borthwick KJ, Bavalia C, Eady N, Fritz DM, Rungroj N, Giersch AB, Morton CC, Axon PR, Akil I, Al-Sabban EA, Baguley DM, Bianca S, Bakkaloglu A, Bircan Z, Chauveau D, Clermont MJ, Guala A, Hulton SA, Kroes H, Li Volti G, Mir S, Mocan H, Nayir A, Ozen S, Rodriguez Soriano J, Sanjad SA, Tasic V, Taylor CM, Topaloglu R, Smith AN, Karet FE. Novel ATP6V1B1 and ATP6V0A4 mutations in autosomal recessive distal renal tubular acidosis with new evidence for hearing loss. *J Med Genet.* 2002 Nov;39(11):796-803. doi: 10.1136/jmg.39.11.796. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/12414817>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1735017/>)

Genomic Location

The *ATP6V1B1* gene is found on chromosome 2 (<https://medlineplus.gov/genetics/chromosome/2/>).

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