



## ATP6V0A2 gene

ATPase H<sup>+</sup> transporting V0 subunit a2

### Normal Function

The *ATP6V0A2* gene provides instructions for making one part, the a2 subunit, of a large protein complex (a group of proteins that work together). This protein complex is known as a vacuolar H<sup>+</sup>-ATPase (V-ATPase). A V-ATPase acts as a pump to move positively charged hydrogen atoms (protons) across cell membranes.

V-ATPases are embedded in the membranes surrounding cells, where they transport protons into and out of cells. 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.

Within cells, V-ATPases help regulate the pH of particular cell compartments. These compartments include endosomes and lysosomes, which digest and recycle materials that the cell no longer needs. Studies suggest that V-ATPases are also involved in the movement (trafficking) of small sac-like structures called vesicles. Vesicles transport many types of molecules within cells.

V-ATPases also play a key role in a complex process called glycosylation, in which proteins are modified by adding sugar molecules. Glycosylation is necessary for the normal function of many different kinds of proteins. V-ATPases regulate the pH of a cellular structure called the Golgi apparatus, where glycosylation occurs.

### Health Conditions Related to Genetic Changes

#### Cutis laxa

At least 18 mutations in the *ATP6V0A2* gene have been identified in people with cutis laxa. *ATP6V0A2* mutations cause a form of the disorder called autosomal recessive cutis laxa type II, which is characterized by loose, sagging skin and distinctive facial features. Some affected individuals also have delayed development, intellectual disability, seizures, and problems with movement that can worsen over time.

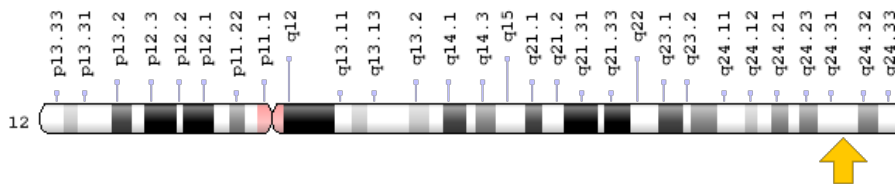
Mutations in the *ATP6V0A2* gene prevent the cell from producing a functional a2 subunit, which disrupts the normal function of V-ATPases. It is unclear how these genetic changes cause the signs and symptoms of cutis laxa. Researchers suspect that changes in V-ATPase function may disrupt the normal glycosylation of proteins, including several that are involved in the assembly and maintenance of elastic fibers. Elastic fibers are slender bundles of proteins that provide strength and flexibility to connective tissue (tissue that supports the body's joints and organs). People with

cutis laxa have a reduced density of elastic fibers, which weakens connective tissue in the skin, lungs, and other organs. These defects in connective tissue underlie many of the major features of the disorder.

### Chromosomal Location

Cytogenetic Location: 12q24.31, which is the long (q) arm of chromosome 12 at position 24.31

Molecular Location: base pairs 123,712,318 to 123,761,755 on chromosome 12 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

### Other Names for This Gene

- A2V-ATPase
- ATP6a2
- ATP6N1D
- ATPase, H<sup>+</sup> transporting, lysosomal V0 subunit a2
- J6B7
- Stv1
- TJ6
- TJ6M
- TJ6s
- Vph1
- VPP2\_HUMAN

## **Additional Information & Resources**

### Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): What Is the Purpose of Glycosylation?  
<https://www.ncbi.nlm.nih.gov/books/NBK26941/#A2354>
- Molecular Cell Biology (fourth edition, 2000): V-Class H<sup>+</sup> ATPases Pump Protons across Lysosomal and Vacuolar Membranes  
<https://www.ncbi.nlm.nih.gov/books/NBK21481/#A4080>

### GeneReviews

- ATP6V0A2-Related Cutis Laxa  
<https://www.ncbi.nlm.nih.gov/books/NBK5200>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28ATP6V0A2%5BTIAB%5D%29+OR+%28%28V-ATPase%5BTIAB%5D%29+AND+%28a2%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

### OMIM

- ATPase, H<sup>+</sup> TRANSPORTING, LYSOSOMAL, V0 SUBUNIT A2  
<http://omim.org/entry/611716>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_ATP6V0A2.html](http://atlasgeneticsoncology.org/Genes/GC_ATP6V0A2.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=ATP6V0A2%5Bgene%5D>
- HGNC Gene Family: V-type ATPases  
<https://www.genenames.org/cgi-bin/genefamilies/set/415>
- HGNC Gene Symbol Report  
[https://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=18481](https://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=18481)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/23545>
- UniProt  
<http://www.uniprot.org/uniprot/Q9Y487>

## Sources for This Summary

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