

ATP2A2 gene

ATPase sarcoplasmic/endoplasmic reticulum Ca²⁺ transporting 2

Normal Function

The *ATP2A2* gene provides instructions for making an enzyme called sarco(endo)plasmic reticulum calcium-ATPase 2 (SERCA2). This enzyme belongs to a family of ATPase enzymes that helps control the level of positively charged calcium atoms (calcium ions) inside cells. SERCA2 is found in the endoplasmic reticulum inside the cell and in a related structure called the sarcoplasmic reticulum inside muscle cells. The endoplasmic reticulum is a structure that is involved in protein processing and transport. The sarcoplasmic reticulum assists with muscle contraction and relaxation by releasing and storing calcium ions. Calcium ions act as signals for a large number of activities that are important for the normal development and function of cells. SERCA2 allows calcium ions to pass into and out of the cell in response to cell signals.

Health Conditions Related to Genetic Changes

Darier disease

Over 100 variants (also called mutations) in the *ATP2A2* gene have been found to cause Darier disease. This disorder is a skin condition characterized by wart-like blemishes on the body. Most of these variants change a single protein building block (amino acid) in the SERCA2 enzyme. The variants that cause Darier disease prevent cells from producing enough SERCA2, or they cause cells to produce nonfunctional versions of SERCA2. Cells with only one functional copy of the *ATP2A2* gene produce half the normal amount of SERCA2 protein. It is thought that insufficient amounts of SERCA2 combined with outside factors such as heat and minor injuries cause the signs and symptoms of Darier disease.

Other Names for This Gene

- AT2A2_HUMAN
- ATP2B
- ATPase, Ca⁺⁺ dependent, slow-twitch, cardiac muscle-2
- ATPase, Ca⁺⁺ transporting, cardiac muscle, slow twitch 2
- calcium-transporting ATPase sarcoplasmic reticulum type, slow twitch skeletal

muscle isoform

- sarcoplasmic reticulum Ca(2+)-ATPase 2
- sarcoplasmic/endoplasmic reticulum calcium ATPase 2
- SERCA2
- SR Ca(2+)-ATPase 2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ATP2A2 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=488\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=488[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28ATP2A2%5BTIAB%5D%29+OR+%28SERCA2%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+1800+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- ATPase, Ca(2+)-TRANSPORTING, SLOW-TWITCH; ATP2A2 (<https://omim.org/entry/108740>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/488>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ATP2A2\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ATP2A2[gene]))

References

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Genomic Location

The *ATP2A2* gene is found on chromosome 12 (<https://medlineplus.gov/genetics/chromosome/12/>).

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