ATP2A1 gene

ATPase sarcoplasmic/endoplasmic reticulum Ca2+ transporting 1

Normal Function

The *ATP2A1* gene provides instructions for making an enzyme called sarco(endo) plasmic reticulum calcium-ATPase 1 (SERCA1). This enzyme belongs to a family of ATPase enzymes that help control the level of positively charged calcium atoms (calcium ions) inside cells. The SERCA1 enzyme is found in skeletal muscle cells. (Skeletal muscles are the muscles used for movement.) Within muscle cells, the SERCA1 enzyme is located in the membrane of a structure called the sarcoplasmic reticulum. This structure plays a major role in muscle contraction and relaxation by storing and releasing calcium ions. When calcium ions are transported out of the sarcoplasmic reticulum, muscles contract; when calcium ions are transported into the sarcoplasmic reticulum, muscles relax. The SERCA1 enzyme transports calcium ions from the cell into the sarcoplasmic reticulum, triggering muscle relaxation.

Health Conditions Related to Genetic Changes

Brody myopathy

At least 10 mutations in the *ATP2A1* gene have been found to cause Brody myopathy, a muscle disorder characterized by muscle cramping after exercise. Most *ATP2A1* gene mutations lead to a premature stop signal in the instructions for making the SERCA1 enzyme, resulting in a nonfunctional enzyme. Other mutations lead to the production of a SERCA1 enzyme with decreased function. As a result, calcium ions are slow to enter the sarcoplasmic reticulum and muscle relaxation is delayed. After exercise or other strenuous activity, during which the muscles rapidly contract and relax, people with Brody myopathy develop muscle cramps because their muscles cannot fully relax. Scientists believe that other proteins or other pathways may function in the absence of a fully functional SERCA1 enzyme to transport calcium ions into the sarcoplasmic reticulum and help with muscle relaxation.

Other Names for This Gene

- AT2A1_HUMAN
- ATP2A
- ATPase, Ca++ transporting, cardiac muscle, fast twitch 1

- calcium-transporting ATPase sarcoplasmic reticulum type, fast twitch skeletal muscle isoform 1
- endoplasmic reticulum class 1 Ca2+ ATPase
- sarcoplasmic/endoplasmic reticulum calcium ATPase 1
- SERCA1
- SR Ca2+ ATPase 1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ATP2A1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=487[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28ATP2A1%5BTIAB%5D%29+OR+%28SERCA1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2880+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ATPase, Ca(2+)-TRANSPORTING, FAST-TWITCH 1; ATP2A1 (https://omim.org/entry/108730)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/487)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ATP2A1[gene])

References

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Genomic Location

The *ATP2A1* gene is found on chromosome 16 (https://medlineplus.gov/genetics/chromosome/16/).

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