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.
**Chromosomal Location**

Cytogenetic Location: 16p11.2, which is the short (p) arm of chromosome 16 at position 11.2

Molecular Location: base pairs 28,878,488 to 28,904,511 on chromosome 16 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

**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**

**Educational Resources**
- Washington University, St. Louis: Neuromuscular Disease Center: Brody's Disease
  https://neuromuscular.wustl.edu/mother/activity.html#brody

**Scientific Articles on PubMed**
- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28ATP2A1%5BTIAB%5D%29+OR+%28SERCA1%5BTIAB%5D%29+AND+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29+AND+English%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2880+days%22+AND+5Bdp%5D
Catalog of Genes and Diseases from OMIM

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

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology

- ClinVar

- HGNC Gene Symbol Report

- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:487

- NCBI Gene

- UniProt
  https://www.uniprot.org/uniprot/O14983

Sources for This Summary

- OMIM: ATPase, Ca(2+)-TRANSPORTING, FAST-TWITCH 1
  http://omim.org/entry/108730

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/8841193

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12763865

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20142766

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