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. Within the cell, SERCA2 is found in the endoplasmic reticulum and a related structure in muscle cells called the sarcoplasmic reticulum. The endoplasmic reticulum is a structure inside the cell 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

More than 130 mutations in the ATP2A2 gene have been found to cause Darier disease. Most of these mutations change a single protein building block (amino acid) in the SERCA2 enzyme. All mutations cause the production of a nonfunctional SERCA2 enzyme or cause no SERCA2 to be produced from one copy of the gene. 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 injury cause the signs and symptoms of Darier disease.
Chromosomal Location

Cytogenetic Location: 12q24.11, which is the long (q) arm of chromosome 12 at position 24.11

Molecular Location: base pairs 110,281,227 to 110,351,093 on chromosome 12 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)

Credit: Genome Decoration Page/NCBI

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

Educational Resources

• Biochemistry (fifth edition, 2002): The Sarcoplasmic Reticulum Ca2+ ATPase Is an Integral Membrane Protein
  https://www.ncbi.nlm.nih.gov/books/NBK22464/#A1780

• Molecular Cell Biology (fourth edition, 2000): Muscle Ca2+ ATPase Pumps Ca2+ Ions from the Cytosol into the Sarcoplasmic Reticulum

Scientific Articles on PubMed
• PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?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%5B5D%29+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM
• ATPase, Ca(2+)-TRANSPORTING, SLOW-TWITCH
  http://omim.org/entry/108740

Research Resources
• Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_ATP2A2.html

• ClinVar

• HGNC Gene Symbol Report

• Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:488

• NCBI Gene

• UniProt
  https://www.uniprot.org/uniprot/P16615

Sources for This Summary
• OMIM: ATPase, Ca(2+)-TRANSPORTING, SLOW-TWITCH
  http://omim.org/entry/108740

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17214724

• Basic Neurochemistry (sixth edition, 1999): ATP-Dependent Ca2+ Pumps
  https://www.ncbi.nlm.nih.gov/books/NBK27906/
• Biochemistry (fifth edition, 2002): Mechanism of P-Type ATPase Action
  https://www.ncbi.nlm.nih.gov/books/NBK22464/?rendertype=figure&id=A1782


Reprinted from Genetics Home Reference:

Reviewed: March 2008
Published: January 22, 2019

Lister Hill National Center for Biomedical Communications
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