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 110,281,247 to 110,351,093 on chromosome 12 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (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%29+AND+english%5B5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%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

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15149492

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16766529

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15099360

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11168576

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