ABCB7 gene

ATP binding cassette subfamily B member 7

Normal Function

The *ABCB7* gene provides instructions for making a protein known as an ATP-binding cassette (ABC) transporter. ABC transporter proteins carry many types of molecules across membranes in cells.

The ABCB7 protein is located in the inner membrane of cell structures called mitochondria. Mitochondria are involved in a wide variety of cellular activities, including energy production, chemical signaling, and regulation of cell growth and division. In the mitochondria of developing red blood cells (erythroblasts), the ABCB7 protein plays a critical role in the production of heme. Heme contains iron and is a component of hemoglobin, the protein that carries oxygen in the blood.

The ABCB7 protein is also involved in the formation of certain proteins containing clusters of iron and sulfur atoms (Fe-S clusters). Researchers suspect that the ABCB7 protein transports Fe-S clusters from mitochondria, where they are formed, to the surrounding cellular fluid (cytosol), where they can be incorporated into proteins. Overall, researchers believe that the ABCB7 protein helps maintain an appropriate balance of iron (iron homeostasis) in developing red blood cells.

Health Conditions Related to Genetic Changes

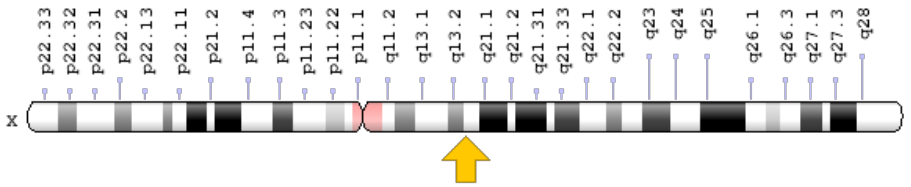
X-linked sideroblastic anemia and ataxia

At least three mutations in the *ABCB7* gene have been identified in people with X-linked sideroblastic anemia with ataxia. Each of these mutations changes a single protein building block (amino acid) in the ABCB7 protein, slightly altering its structure. These changes disrupt the protein's usual role in heme production and iron homeostasis. Anemia results when heme cannot be produced normally, and therefore not enough hemoglobin is made. It is unclear how changes in the *ABCB7* gene lead to problems with coordination and balance (ataxia) and other movement abnormalities.

Chromosomal Location

Cytogenetic Location: Xq13.3, which is the long (q) arm of the X chromosome at position 13.3

Molecular Location: base pairs 75,051,048 to 75,156,283 on the X chromosome (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ABC transporter 7 protein
- ABCB7_HUMAN
- ASAT
- Atm1p
- ATP-binding cassette 7
- ATP-binding cassette sub-family B member 7, mitochondrial
- ATP-binding cassette, sub-family B (MDR/TAP), member 7
- ATP-binding cassette, sub-family B, member 7
- EST140535

Additional Information & Resources

Educational Resources

- The Human ATP-Binding Cassette (ABC) Transporter Superfamily: ABCB Genes https://www.ncbi.nlm.nih.gov/books/NBK3/#A179

Clinical Information from GeneReviews

- X-Linked Sideroblastic Anemia and Ataxia https://www.ncbi.nlm.nih.gov/books/NBK1321

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28ABCB7%5BTIAB%5D%29+OR+%28%28ABC7%5BTIAB%5D%29+OR+%28Atm1p%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+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- ATP-BINDING CASSETTE, SUBFAMILY B, MEMBER 7
  http://omim.org/entry/300135

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ABCB7%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:48
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:22
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/22
- UniProt
  https://www.uniprot.org/uniprot/O75027

Sources for This Summary

- Allikmets R, Raskind WH, Hutchinson A, Schueck ND, Dean M, Koeller DM. Mutation of a putative mitochondrial iron transporter gene (ABC7) in X-linked sideroblastic anemia and ataxia (XLSA/A). Hum Mol Genet. 1999 May;8(5):743-9.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10196363
- Bekri S, Kispal G, Lange H, Fitzsimons E, Tolmie J, Lill R, Bishop DF. Human ABC7 transporter: gene structure and mutation causing X-linked sideroblastic anemia with ataxia with disruption of cytosolic iron-sulfur protein maturation. Blood. 2000 Nov 1;96(9):3256-64.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11050011
- Cavadini P, Biasiotto G, Poli M, Levi S, Verardi R, Zanella I, Derosas M, Ingrassia R, Corrado M, Arosio P. RNA silencing of the mitochondrial ABCB7 transporter in HeLa cells causes an iron-deficient phenotype with mitochondrial iron overload. Blood. 2007 Apr 15;109(8):3552-9. Epub 2006 Dec 27.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17192393
- Maguire A, Hellier K, Hammans S, May A. X-linked cerebellar ataxia and sideroblastic anaemia associated with a missense mutation in the ABC7 gene predicting V411L. Br J Haematol. 2001 Dec; 115(4):910-7.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11843825

- Pondarre C, Campagna DR, Antiochos B, Sikorski L, Mulhern H, Fleming MD. Abcb7, the gene responsible for X-linked sideroblastic anemia with ataxia, is essential for hematopoiesis. Blood. 2007 Apr 15;109(8):3567-9. Epub 2006 Dec 27.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17192398
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1852240/
- Pondarré C, Antiochos BB, Campagna DR, Clarke SL, Greer EL, Deck KM, McDonald A, Han AP, Medlock A, Kutok JL, Anderson SA, Eisenstein RS, Fleming MD. The mitochondrial ATP-binding cassette transporter Abcb7 is essential in mice and participates in cytosolic iron-sulfur cluster biogenesis. Hum Mol Genet. 2006 Mar 15;15(6):953-64. Epub 2006 Feb 8.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16467350
- Shimada Y, Okuno S, Kawai A, Shinomiya H, Saito A, Suzuki M, Omori Y, Nishino N, Kanemoto N, Fujiwara T, Horie M, Takahashi E. Cloning and chromosomal mapping of a novel ABC transporter gene (hABC7), a candidate for X-linked sideroblastic anemia with spinocerebellar ataxia. J Hum Genet. 1998;43(2):115-22.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/9621516
- Taketani S, Kakimoto K, Ueta H, Masaki R, Furukawa T. Involvement of ABC7 in the biosynthesis of heme in erythroid cells: interaction of ABC7 with ferrochelatase. Blood. 2003 Apr 15;101(8): 3274-80. Epub 2002 Dec 12.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12480705

Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/gene/ABCB7

Reviewed: April 2009
Published: September 10, 2019

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services