

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.

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

Tests Listed in the Genetic Testing Registry

- Tests of ABCB7 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=22\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=22[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?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%29>)

Catalog of Genes and Diseases from OMIM

- ATP-BINDING CASSETTE, SUBFAMILY B, MEMBER 7; ABCB7 (<https://omim.org/entry/300135>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/22>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ABCB7\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ABCB7[gene]))

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Genomic Location

The *ABCB7* gene is found on the X chromosome (<https://medlineplus.gov/genetics/chromosome/x/>).

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