SAR1B gene

secretion associated Ras related GTPase 1B

Normal Function

The *SAR1B* gene provides instructions for making a protein that is produced in a variety of tissues. Most research involving the SAR1B protein has studied its role in the digestive tract.

The SAR1B protein is needed for the transport of molecules called chylomicrons. During digestion, chylomicrons are formed within cells called enterocytes that line the small intestine and absorb nutrients. Chylomicrons are needed to absorb fat-soluble vitamins (vitamins K, E, and D) and carry fats and cholesterol from the small intestine into the bloodstream.

Within enterocytes, SAR1B proteins help transport immature chylomicrons from a cell structure called the endoplasmic reticulum to another cell structure called the Golgi apparatus. Immature chylomicrons are processed within the Golgi apparatus, resulting in mature chylomicrons. These mature chylomicrons are then released from enterocytes into the bloodstream so the body can use the fats and fat-soluble vitamins they carry. Sufficient levels of fats, cholesterol, and vitamins are necessary for normal growth and development.

In other tissues, such as the heart and other muscles, the SAR1B protein is likely involved in transporting calcium within cells.

Health Conditions Related to Genetic Changes

Chylomicron retention disease

More than 20 mutations in the *SAR1B* gene have been found to cause chylomicron retention disease. This is an inherited disorder that impairs the normal absorption of fats, cholesterol, and fat-soluble vitamins from food. Most of the mutations change one protein building block (amino acid) in the SAR1B protein. Other mutations lead to the production of an abnormally small version of the protein that cannot function properly.

SAR1B gene mutations disrupt the SAR1B protein's ability to transport immature chylomicrons from the endoplasmic reticulum to the Golgi apparatus. As a result, mature chylomicrons are not released into the bloodstream. Impaired chylomicron transport causes severely decreased absorption (malabsorption) of dietary fats and fat-

soluble vitamins, leading to nutritional and developmental problems in people with chylomicron retention disease.

Other Names for This Gene

- GTP-binding protein Sara
- SAR1 gene homolog B (S. cerevisiae)
- SAR1 homolog B (S. cerevisiae)
- SAR1a gene homolog 2
- SAR1B_HUMAN
- SARA2
- secretion associated, Ras related GTPase 1B

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SAR1B (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=51128[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28SAR1B%5BTIAB%5D%29+OR+%28SARA2%5BTIAB%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

- SECRETION-ASSOCIATED RAS-RELATED GTPase 1B; SAR1B (https://omim.org/entry/607690)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/51128)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SAR1B[gene])

References

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Genomic Location

The *SAR1B* gene is found on chromosome 5 (https://medlineplus.gov/genetics/chromosome/5/).

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