SCARB2 gene

scavenger receptor class B member 2

Normal Function

The *SCARB2* gene provides instructions for making a protein called lysosomal integral membrane protein-2 (LIMP-2). As its name suggests, this protein is primarily found in the membrane of cellular structures called lysosomes, which are specialized compartments that digest and recycle materials. Before moving to the lysosome, the LIMP-2 protein is processed in a cellular structure called the endoplasmic reticulum. There, LIMP-2 attaches to an enzyme called beta-glucocerebrosidase and transports it to the lysosome. In lysosomes, beta-glucocerebrosidase breaks down a fatty substance called glucocerebroside. The LIMP-2 protein remains in the lysosomal membrane after transporting beta-glucocerebrosidase and is important for the stability of these structures.

The LIMP-2 protein has additional functions outside the lysosome. In the heart, the protein is found in regions known as intercalated discs, which connect individual heart muscle cells together to form strong fibers. The LIMP-2 protein appears to play a role when the heart muscle is abnormally enlarged and has to work harder than normal, although its exact function is not clear.

The LIMP-2 protein is sometimes found in the outer membrane that surrounds the cell. Certain viruses can attach to LIMP-2, which allows them to enter and infect the cell. In particular, enterovirus 71 and certain strains of coxsackievirus (A7, A14, and A16), which cause a viral infection known as hand, foot, and mouth disease, use the LIMP-2 protein.

Health Conditions Related to Genetic Changes

Action myoclonus–renal failure syndrome

At least 20 mutations in the *SCARB2* gene have been associated with action myoclonus–renal failure (AMRF) syndrome. This rare condition causes episodes of involuntary muscle jerking or twitching, particularly when trying to make intentional movements (action myoclonus). Another common feature of AMRF syndrome is kidney (renal) disease; despite being referenced in the condition name, kidney function is not affected in every person with the condition.

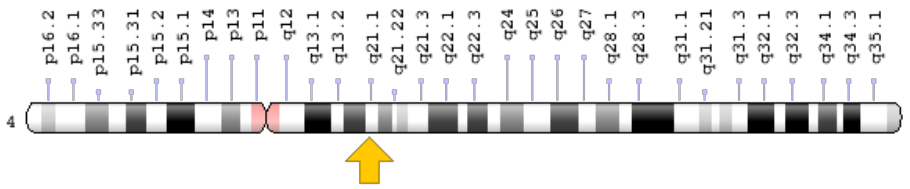
SCARB2 gene mutations associated with AMRF syndrome lead to production of an altered LIMP-2 protein that is stuck in the endoplasmic reticulum and cannot get to the lysosome. As a result, the movement of beta-glucocerebrosidase to lysosomes is impaired. It is thought that a shortage of beta-glucocerebrosidase in these structures contributes to the signs and symptoms of AMRF syndrome, although the mechanism

is unclear. Researchers are working to understand why some people with *SCARB2* gene mutations have kidney problems and others do not.

Chromosomal Location

Cytogenetic Location: 4q21.1, which is the long (q) arm of chromosome 4 at position 21.1

Molecular Location: base pairs 76,158,737 to 76,213,824 on chromosome 4 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- 85 kDa lysosomal membrane sialoglycoprotein
- 85 kDa lysosomal sialoglycoprotein scavenger receptor class B, member 2
- AMRF
- CD36 antigen (collagen type I receptor, thrombospondin receptor)-like 2 (lysosomal integral membrane protein II)
- CD36 antigen-like 2
- CD36L2
- EPM4
- HLGP85
- LGP85
- LIMP-2
- LIMP II
- LIMPII
- lysosome membrane protein 2 isoform 1 precursor
- lysosome membrane protein 2 isoform 2 precursor
- lysosome membrane protein II

- scavenger receptor class B, member 2
- SR-BII

Additional Information & Resources

Clinical Information from GeneReviews

- Action Myoclonus - Renal Failure Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK333437

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28SCARB2%5BTIAB%5D%29+OR+%28scavenger+receptor+class+B+member+2%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+1800+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- SCAVENGER RECEPTOR CLASS B, MEMBER 2
  http://omim.org/entry/602257

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_SCARB2.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SCARB2%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:1665
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:950
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/950
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https://ghr.nlm.nih.gov/gene/SCARB2

Reviewed: June 2016
Published: November 12, 2019

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