SERAC1 gene

serine active site containing 1

Normal Function

The *SERAC1* gene provides instructions for making a protein whose function is not completely understood. Studies suggest that the SERAC1 protein is involved in altering (remodeling) certain fats called phospholipids, particularly a phospholipid called phosphatidylglycerol.

Another phospholipid called cardiolipin is made from phosphatidylglycerol. Cardiolipin is a component of the membrane that surrounds cellular structures called mitochondria, which convert the energy from food into a form that cells can use, and is important for the proper functioning of these structures.

Researchers believe that the SERAC1 protein is also involved in the movement of a waxy, fat-like substance called cholesterol within cells. Cholesterol is a structural component of cell membranes and plays a role in the production of certain hormones and digestive acids. It has important functions both before and after birth.

Health Conditions Related to Genetic Changes

MEGDEL syndrome

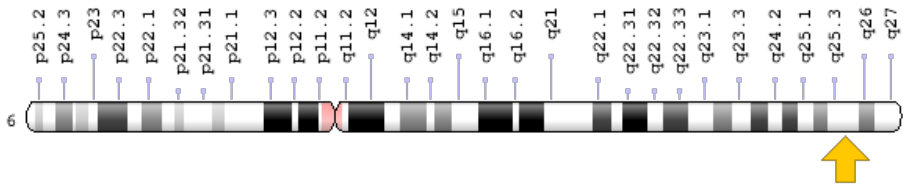
At least 16 mutations in the *SERAC1* gene have been found to cause MEGDEL syndrome. This condition is characterized by hearing loss, neurological problems, certain changes in the brain described as Leigh-like disease, and abnormally high amounts of an acid called 3-methylglutaconic acid in the urine. The *SERAC1* gene mutations that cause this condition reduce the amount of SERAC1 protein that is produced or lead to production of a protein with little or no function. As a result, phosphatidylglycerol remodeling is impaired, which likely alters the composition of cardiolipin. Researchers speculate that the abnormal cardiolipin affects mitochondrial function, reducing cellular energy production and leading to the neurological and hearing problems characteristic of MEGDEL syndrome. It is unclear how *SERAC1* gene mutations lead to abnormal release of 3-methylglutaconic acid in the urine.

Leigh syndrome

Chromosomal Location

Cytogenetic Location: 6q25.3, which is the long (q) arm of chromosome 6 at position 25.3

Molecular Location: base pairs 158,109,504 to 158,168,280 on chromosome 6 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- FLJ14917
- protein SERAC1
- serine active site-containing protein 1

Additional Information & Resources

Educational Resources

- Molecular Cell Biology (fourth edition, 2000): Phospholipids Are the Main Lipid Constituents of Most Biomembranes
  https://www.ncbi.nlm.nih.gov/books/NBK21583/#_A1147_

Clinical Information from GeneReviews

- MEGDEL Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK195853

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28SERAC1%29+OR+%28MEGDEL%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

Catalog of Genes and Diseases from OMIM

- SERINE ACTIVE SITE-CONTAINING PROTEIN 1
  http://omim.org/entry/614725

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SERAC1%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:21061
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:84947
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/84947
- UniProt
  https://www.uniprot.org/uniprot/Q96JX3

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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/22683713

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