OXCT1 gene
3-oxoacid CoA-transferase 1

Normal Function
The OXCT1 gene provides instruction for making an enzyme called succinyl-CoA:3-ketoacid CoA transferase, often abbreviated as SCOT. The SCOT enzyme is made in the energy-producing centers of cells (mitochondria). The enzyme plays a role in the breakdown of ketones, which are molecules produced by the liver during the breakdown of fats. Ketones are an important source of energy during prolonged periods without food (fasting) or when energy demands are increased, such as during illness or when exercising. In the processing of ketones, the SCOT enzyme converts the molecule acetoacetate to acetoacetyl-CoA.

Health Conditions Related to Genetic Changes
Succinyl-CoA:3-ketoacid CoA transferase deficiency
At least 20 mutations in the OXCT1 gene have been found to cause SCOT deficiency, a condition characterized by episodes of extreme tiredness, appetite loss, and seizures, known as ketoacidotic attacks. Most OXCT1 gene mutations lead to changes in single protein building blocks (amino acids) in the SCOT enzyme and result in an enzyme with little or no function. A reduction in the amount of functional enzyme leads to an inability to break down ketones, often resulting in decreased energy production and an elevated level of ketones in the blood. If these signs become severe, a ketoacidotic attack can occur.
Chromosomal Location

Cytogenetic Location: 5p13.1, which is the short (p) arm of chromosome 5 at position 13.1

Molecular Location: base pairs 41,730,065 to 41,870,535 on chromosome 5 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

• 3-oxoacid CoA transferase 1
• 3-oxoacid-CoA transferase 1
• OXCT
• SCOT
• SCOT1_HUMAN
• somatic-type succinyl CoA:3-oxoacid CoA-transferase
• somatic-type succinyl-CoA:3-oxoacid-CoA-transferase
• succinyl-CoA:3-ketoacid-CoA transferase
• succinyl-CoA:3-ketoacid-coenzyme A transferase 1, mitochondrial
• succinyl CoA:3-oxoacid CoA transferase

Additional Information & Resources

Educational Resources

• Biochemistry (fifth edition, 2002): Ketone Bodies Are a Major Fuel in Some Tissues
  https://www.ncbi.nlm.nih.gov/books/NBK22387/#A3077
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28OXCT1%5BTIAB%5D%29+OR+%28%28OXCT%5BTIAB%5D%29+OR+%28SCOT%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

- 3-OXOACID CoA TRANSFERASE 1
  http://omim.org/entry/601424

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_OXCT1.html

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=OXCT1%5Bgene%5D

- HGNC Gene Symbol Report

- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:5019

- NCBI Gene

- UniProt
  https://www.uniprot.org/uniprot/P55809

Sources for This Summary

- OMIM: 3-OXOACID CoA TRANSFERASE 1
  http://omim.org/entry/601424

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11757586

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20652411
  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/10964512

  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/21296660

  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/15496607

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https://ghr.nlm.nih.gov/gene/OXCT1

Reviewed: December 2011
Published: August 20, 2019

Lister Hill National Center for Biomedical Communications
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