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.

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-oxoacid CoA transferase

- succinyl-CoA:3-ketoacid-CoA transferase
- succinyl-CoA:3-ketoacid-coenzyme A transferase 1, mitochondrial

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of OXCT1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5019[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?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; OXCT1 (https://omim.org/entry/601424)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/5019)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=OXCT1[gene])

References

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Genomic Location

The *OXCT1* gene is found on chromosome 5 (https://medlineplus.gov/genetics/chromosome/5/).

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