

## SUCLA2 gene

succinate-CoA ligase ADP-forming subunit beta

### Normal Function

The *SUCLA2* gene provides instructions for making one part (a beta subunit) of an enzyme called succinate-CoA ligase. The body makes two slightly different versions of this enzyme: ADP-forming succinate-CoA ligase (A-SUCL) and GDP-forming succinate-CoA ligase (G-SUCL). Only A-SUCL contains the beta subunit produced from the *SUCLA2* gene. This version of the enzyme is most active in tissues that require a large amount of energy, such as those of the brain and muscles.

Succinate-CoA ligase plays a critical role in mitochondria, which are structures inside cells that convert the energy from food into a form that cells can use. Within mitochondria, this enzyme is involved in a series of chemical reactions known as the citric acid cycle or Krebs cycle. These reactions allow cells to use oxygen and generate energy.

Mitochondria each contain a small amount of DNA, known as mitochondrial DNA or mtDNA. Studies suggest that succinate-CoA ligase interacts with another enzyme, nucleoside diphosphate kinase, to produce and maintain the building blocks of mitochondrial DNA. Having an adequate amount of mitochondrial DNA is essential for normal energy production within cells.

### Health Conditions Related to Genetic Changes

#### Succinate-CoA ligase deficiency

At least four mutations in the *SUCLA2* gene have been identified in people with succinate-CoA ligase deficiency. Each of these mutations alters the structure of A-SUCL, reducing the enzyme's activity. However, *SUCLA2* gene mutations do not affect the other version of succinate-CoA ligase, G-SUCL. Studies suggest that the activity of G-SUCL may be able to compensate for a loss of A-SUCL in some tissues.

A shortage (deficiency) of A-SUCL leads to problems with the production and maintenance of mitochondrial DNA in the brain, muscles, and other tissues that require a large amount of energy. A reduction in the amount of mitochondrial DNA (known as mitochondrial DNA depletion) impairs mitochondrial function and the production of energy within cells. These problems lead to weak muscle tone (hypotonia), delayed

development, and the other characteristic features of succinate-CoA ligase deficiency.

### Leigh syndrome

MedlinePlus Genetics provides information about Leigh syndrome

### **Other Names for This Gene**

- A-BETA
- ATP-specific succinyl-CoA synthetase, beta subunit
- renal carcinoma antigen NY-REN-39
- SCS-betaA
- SUCB1\_HUMAN
- succinate-CoA ligase beta subunit
- succinate-CoA ligase, ADP-forming, beta subunit

### **Additional Information & Resources**

#### Tests Listed in the Genetic Testing Registry

- Tests of SUCLA2 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8803\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8803[geneid]))

#### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28SUCLA2%5BTIAB%5D%29+OR+%28%28succinate-CoA+ligase%5BTIAB%5D%29+AND+%28beta%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

- SUCCINATE-CoA LIGASE, ADP-FORMING, SUBUNIT BETA; SUCLA2 (<https://omim.org/entry/603921>)

#### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/8803>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=SUCLA2\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=SUCLA2[gene]))

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## Genomic Location

The *SUCLA2* gene is found on chromosome 13 (<https://medlineplus.gov/genetics/chromosome/13/>).

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