# SURF1 gene

SURF1 cytochrome c oxidase assembly factor

## Normal Function

The *SURF1* gene provides instructions for making a protein that is important in oxidative phosphorylation, the process by which the energy from food is converted into a form cells can use. Oxidative phosphorylation involves a series of reactions that take place through several different protein complexes. The SURF1 protein aids in the correct assembly of one of the protein complexes, or enzymes, involved in oxidative phosphorylation called complex IV.

Complex IV, also known as cytochrome *c* oxidase or COX, accepts negatively charged particles (electrons) from earlier steps in oxidative phosphorylation. In addition, the enzyme accepts positively charged particles (protons) from inside the mitochondrion. Using the electrons and protons, the COX enzyme performs a chemical reaction that converts oxygen to water. The enzyme also transfers additional protons across the specialized membrane inside the mitochondrion. These processes create energy that is used to generate adenosine triphosphate (ATP), the cell's main energy source.

## Health Conditions Related to Genetic Changes

Charcot-Marie-Tooth disease

MedlinePlus Genetics provides information about Charcot-Marie-Tooth disease

Cytochrome c oxidase deficiency

MedlinePlus Genetics provides information about Cytochrome c oxidase deficiency

Leigh syndrome

More than 80 different *SURF1* gene mutations have been identified in people with Leigh syndrome, a progressive brain disorder that usually appears in infancy or early childhood. Affected children may experience delayed development, muscle weakness, problems with movement, or difficulty breathing.

Approximately 10 to 15 percent of people with Leigh syndrome have a mutation in the *SURF1* gene. Most *SURF1* gene mutations result in an abnormally short protein. Other mutations replace a single protein building block in the SURF1 protein. The mutated

proteins are broken down in the cell, which results in the absence of SURF1 protein. Lack of SURF1 protein hinders the proper formation of the COX complex. As a result, COX enzyme activity is severely reduced, which leads to impaired oxidative phosphorylation.

Although the exact mechanism is unclear, researchers believe that impaired oxidative phosphorylation can lead to cell death because of decreased energy available in the cell. Certain tissues that require large amounts of energy, such as the brain, muscles, and heart, seem especially sensitive to decreases in cellular energy. Cell death in the brain likely causes the characteristic changes in the brain seen in Leigh syndrome, which contribute to the signs and symptoms of the condition. Cell death in other sensitive tissues may also contribute to the features of Leigh syndrome.

## Other Names for This Gene

- SURF-1
- SURF1_HUMAN
- surfeit 1
- surfeit locus protein 1

## Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SURF1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6834[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28SURF1%5BTIAB%5D%29+OR+%28surfeit+1%5BTIAB%5D%29%29+OR+%28surfeit+locus+protein+1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2160+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- SURFEIT 1; SURF1 (https://omim.org/entry/185620)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/6834)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SURF1[gene])

## References


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- Yao J, Shoubridge EA. Expression and functional analysis of SURF1 in Leighsyndrome patients with cytochrome c oxidase deficiency. Hum Mol Genet. 1999Dec;8(13):2541-9. doi: 10.1093/hmg/8.13.2541. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/10556303)


## Genomic Location

The *SURF1* gene is found on chromosome 9 (https://medlineplus.gov/genetics/chromosome/9/).

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