

## ACOX1 gene

acyl-CoA oxidase 1

### Normal Function

The *ACOX1* gene provides instructions for making an enzyme called peroxisomal straight-chain acyl-CoA oxidase. This enzyme is found in sac-like cell structures (organelles) called peroxisomes, which contain a variety of enzymes that break down many different substances. The peroxisomal straight-chain acyl-CoA oxidase enzyme plays a role in the breakdown of certain fat molecules called very long-chain fatty acids (VLCFAs). Specifically, it is involved in the first step of a process called the peroxisomal fatty acid beta-oxidation pathway. This process shortens the VLCFA molecules by two carbon atoms at a time until the VLCFAs are converted to a molecule called acetyl-CoA, which is transported out of the peroxisomes for reuse by the cell.

### Health Conditions Related to Genetic Changes

#### Peroxisomal acyl-CoA oxidase deficiency

More than 20 *ACOX1* gene mutations have been identified in people with peroxisomal acyl-CoA oxidase deficiency. These mutations prevent the peroxisomal straight-chain acyl-CoA oxidase enzyme from breaking down VLCFAs efficiently. As a result, these fatty acids accumulate in the body. It is unclear exactly how VLCFA accumulation leads to the specific features of peroxisomal acyl-CoA oxidase deficiency. However, researchers suggest that the abnormal fatty acid accumulation triggers inflammation in the nervous system that leads to the breakdown of myelin, which is the covering that protects nerves and promotes the efficient transmission of nerve impulses. Destruction of myelin leads to a loss of myelin-containing tissue (white matter) in the brain and spinal cord; loss of white matter is described as leukodystrophy. Leukodystrophy is likely involved in the development of the neurological abnormalities that occur in peroxisomal acyl-CoA oxidase deficiency.

### Other Names for This Gene

- ACOX
- acyl-CoA oxidase 1, palmitoyl
- acyl-CoA oxidase, straight-chain
- acyl-Coenzyme A oxidase 1, palmitoyl

- AOX
- PALMCOX
- palmitoyl-CoA oxidase
- peroxisomal acyl-coenzyme A oxidase 1
- peroxisomal fatty acyl-CoA oxidase
- SCOX
- straight-chain acyl-CoA oxidase

## **Additional Information & Resources**

### Tests Listed in the Genetic Testing Registry

- Tests of ACOX1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=51\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=51[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28ACOX1%5BTIAB%5D%29+OR+%28%28ACOX%5BTIAB%5D%29+OR+%28palmitoyl-CoA+oxidase%5BTIAB%5D%29+OR+%28peroxisomal+fatty+acyl-CoA+oxidase%5BTIAB%5D%29+OR+%28SCOX%5BTIAB%5D%29+OR+%28straight-chain+acyl-CoA+oxidase%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+1440+days%22%5Bdp%5D%29>)

### Catalog of Genes and Diseases from OMIM

- ACYL-CoA OXIDASE 1, PALMITOYL; ACOX1 (<https://omim.org/entry/609751>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/51>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ACOX1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ACOX1[gene]))

## **References**

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## Genomic Location

The *ACOX1* gene is found on chromosome 17 (<https://medlineplus.gov/genetics/chromosome/17/>).

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