PHYH gene
phytanoyl-CoA 2-hydroxylase

Normal Function

The *PHYH* gene provides instructions for making an enzyme called phytanoyl-CoA hydroxylase. This enzyme is critical for the normal function of cell structures called peroxisomes. These sac-like compartments contain enzymes needed to break down many different substances, including fatty acids and certain toxic compounds.

One substance that is broken down in peroxisomes is phytanic acid, a type of fatty acid obtained from the diet (particularly from beef and dairy products). Phytanoyl-CoA hydroxylase is responsible for one of the first steps in breaking down phytanic acid as part of a process known as alpha-oxidation. In subsequent steps, additional enzymes in peroxisomes and other parts of the cell further process this compound into smaller molecules that the body can use for energy.

Researchers suspect that phytanoyl-CoA hydroxylase may have other functions in addition to its role in breaking down phytanic acid. For example, this enzyme appears to help determine the number of peroxisomes within cells and is involved in regulating their activity.

Health Conditions Related to Genetic Changes

Refsum disease

Mutations in the *PHYH* gene have been found to cause more than 90 percent of all cases of Refsum disease. About 30 mutations in this gene have been identified. These mutations alter the structure or production of phytanoyl-CoA hydroxylase, which reduces the enzyme's activity. A shortage of this enzyme disrupts the breakdown of phytanic acid in peroxisomes. As a result, phytanic acid and related compounds build up in the body's tissues. The accumulation of phytanic acid is toxic to cells, although it is unclear how an excess of this substance affects vision and smell and causes the other specific features of Refsum disease.
Chromosomal Location

Cytogenetic Location: 10p13, which is the short (p) arm of chromosome 10 at position 13

Molecular Location: base pairs 13,277,796 to 13,300,130 on chromosome 10 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)

Other Names for This Gene

- LN1
- LNAP1
- PAHX
- PAHX_HUMAN
- PHYH1
- phytanic acid oxidase
- phytanoyl-CoA alpha hydroxylase
- phytanoyl-CoA 2 oxoglutarate dioxygenase
- phytanoyl-CoA alpha-hydroxylase
- phytanoyl-CoA dioxygenase, peroxisomal

Additional Information & Resources

Educational Resources

- Basic Neurochemistry (sixth edition, 1999): Peroxisomal Disease
  https://www.ncbi.nlm.nih.gov/books/NBK28022/
- Madame Curie Bioscience Database: The Biogenesis and Cell Biology of Peroxisomes in Human Health and Disease
  https://www.ncbi.nlm.nih.gov/books/NBK6339/
Clinical Information from GeneReviews

- Refsum Disease
  https://www.ncbi.nlm.nih.gov/books/NBK1353

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28PHYH%5BTIAB%5D%29+OR+%28phytanoyl-CoA+2-hydroxylase%5BTIAB%5D%29%29+OR+%28PAHX%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29%29+AND+human%5Bmh%5D+AND+english%5Bla%5D+AND+%22last+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- PHYTANOYL-CoA HYDROXYLASE
  http://omim.org/entry/602026

Research Resources

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

- HGNC Gene Symbol Report

- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:5264

- NCBI Gene

- UniProt
  https://www.uniprot.org/uniprot/O14832

Sources for This Summary


