SUMF1 gene
sulfatase modifying factor 1

Normal Function
The SUMF1 gene provides instructions for making an enzyme called formylglycine-generating enzyme (FGE). This enzyme is found in a cell structure called the endoplasmic reticulum, which is involved in protein processing and transport. The FGE enzyme modifies other enzymes called sulfatases, which aid in breaking down substances that contain chemical groups known as sulfates. These substances include a variety of sugars, fats, and hormones. Specifically, FGE converts a protein building block (amino acid) within sulfatases called cysteine into a molecule called C-alpha-formylglycine.

Health Conditions Related to Genetic Changes

Multiple sulfatase deficiency
At least 35 mutations in the SUMF1 gene have been found to cause multiple sulfatase deficiency. This condition is apparent at birth or early childhood and is characterized by neurological decline, scaly skin (ichthyosis), and skeletal abnormalities. Most SUMF1 gene mutations that cause multiple sulfatase deficiency change single amino acids in the FGE enzyme. These changes severely reduce enzyme function or produce an unstable enzyme that is quickly broken down. The activity of multiple sulfatases is impaired because the FGE enzyme modifies all known sulfatase enzymes. Sulfate-containing molecules that are not broken down build up in cells, often resulting in cell death. The death of cells in particular tissues, specifically the brain, skeleton, and skin, cause many of the signs and symptoms of multiple sulfatase deficiency. Research indicates that mutations that lead to reduced FGE enzyme function are associated with the less severe cases of the condition, whereas mutations that lead to the production an of unstable FGE enzyme tend to be associated with the more severe cases of multiple sulfatase deficiency.
Chromosomal Location

Cytogenetic Location: 3p26.1, which is the short (p) arm of chromosome 3 at position 26.1

Molecular Location: base pairs 4,034,714 to 4,467,282 on chromosome 3 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- AAPA3037
- C-alpha-formylglycine-generating enzyme 1
- FGE
- FGly-generating enzyme
- sulfatase-modifying factor 1
- UNQ3037

Additional Information & Resources

Educational Resources

  https://www.ncbi.nlm.nih.gov/books/NBK1900/

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28SUMF1%5BTIAB%5D%29+OR+%28sulfatase+modifying+factor+1%5BTIAB%5D%29+OR+%28FGE%5BTIAB%5D%29+OR+%28FGly-generating+enzyme%5BTIAB%5D%29+AND+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D
Catalog of Genes and Diseases from OMIM

- SULFATASE-MODIFYING FACTOR 1
  http://omim.org/entry/607939

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SUMF1%5Bgene%5D

- HGNC Gene Symbol Report

- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:285362

- NCBI Gene

- UniProt
  https://www.uniprot.org/uniprot/Q8NBK3

Sources for This Summary

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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17657823

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- OMIM: SULFATASE-MODIFYING FACTOR 1
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  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3062010/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/23321616
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https://ghr.nlm.nih.gov/gene/SUMF1

Reviewed: July 2014
Published: April 2, 2019

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
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Department of Health & Human Services