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.

Other Names for This Gene

- AAPA3037
- C-alpha-formylglycine-generating enzyme 1
- FGE

- FGly-generating enzyme
- sulfatase-modifying factor 1
- UNQ3037

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SUMF1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=285362[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28SUMF1%5BTIAB%5D%29+OR+%28sulfatase+modifying+factor+1%5BTIAB%5D%29%29+OR+%28%28FGE%5BTIAB%5D%29+OR+%28FGly-generating+enzyme%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+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- SULFATASE-MODIFYING FACTOR 1; SUMF1 (https://omim.org/entry/607939)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/285362)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SUMF1[gene])

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Genomic Location

The *SUMF1* gene is found on chromosome 3 (https://medlineplus.gov/genetics/chromosome/3/).

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