ALOXE3 gene

arachidonate epidermal lipoxygenase 3

Normal Function

The *ALOXE3* gene provides instructions for making an enzyme called eLOX3. This enzyme is part of a family of enzymes called arachidonate lipoxygenases. Most enzymes in this family help add an oxygen molecule to certain fatty acids to produce substances called fatty acid hydroperoxides.

Unlike other lipoxygenases, the eLOX3 enzyme does not act directly on fatty acids. Instead, it is involved in the step following the creation of fatty acid hydroperoxides. The eLOX3 enzyme processes fatty acid hydroperoxides, which are later converted to signaling molecules that are involved in the formation of the layers of fats (lipids) within the outermost layer of the skin (the epidermis). The lipid layers are necessary to prevent water loss (dehydration) through the skin.

Health Conditions Related to Genetic Changes

Nonbullous congenital ichthyosiform erythroderma

At least 20 mutations in the *ALOXE3* gene have been found to cause nonbullous congenital ichthyosiform erythroderma (NBCIE). This condition affects the skin and causes redness; the development of fine, white scales; an increased risk of infections; and excessive dehydration. Most of these mutations change single protein building blocks (amino acids) in the eLOX3 enzyme. Many *ALOXE3* gene mutations lead to the production of a nonfunctional eLOX3 enzyme, which disrupts the processing of the molecules involved in the formation of the lipid layers within the epidermis. Problems with this protective barrier underlie the skin abnormalities and other features of NBCIE.

Other Names for This Gene

- E-LOX
- e-LOX-3
- eLOX3
- epidermal lipoxygenase
- LOXE3_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ALOXE3 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=59344[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28ALOXE3%5BTIAB%5D%29+OR+%28eLOX3%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+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ARACHIDONATE LIPOXYGENASE 3; ALOXE3 (https://omim.org/entry/607206)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/59344)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ALOXE3[gene])

References

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Genomic Location

The *ALOXE3* gene is found on chromosome 17 (https://medlineplus.gov/genetics/chromosome/17/).

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