

ALOX12B gene

arachidonate 12-lipoxygenase, 12R type

Normal Function

The *ALOX12B* gene provides instructions for making an enzyme called 12R-LOX. This enzyme is part of a family of enzymes called arachidonate lipoxygenases. Most of these enzymes help add an oxygen molecule to a particular fatty acid called arachidonic acid. Arachidonate lipoxygenases add oxygen molecules at different locations on the arachidonic acid molecule, producing a variety of substances called fatty acid hydroperoxides. The fatty acid hydroperoxides are then processed into molecules that play an important role in chemical signaling within cells.

Specifically, the 12R-LOX enzyme helps add an oxygen molecule to arachidonic acid to make a substance called 12R-hydroperoxyeicosatetraenoic acid (12R-HPETE). 12R-HPETE is later converted to a signaling molecule that is 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

More than 55 mutations in the *ALOX12B* 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 the mutations change single protein building blocks (amino acids) in the 12R-LOX enzyme. Many *ALOX12B* gene mutations lead to the production of a nonfunctional 12R-LOX enzyme, which disrupts the processing of the molecules involved in the formation of the lipid layer within the epidermis. Problems with this protective barrier underlie the skin abnormalities and other features of NBCIE.

Other disorders

ALOX12B gene mutations have been found to cause another form of ichthyosis called self-healing collodion baby. Individuals with this condition are born with a tight, clear sheath covering their skin called a collodion membrane, which is usually shed during the first few weeks of life. While babies with NBCIE (described above) may also be born with a collodion membrane, infants with self-healing collodion baby often show normal

or near normal skin within a few months.

Only a few people with self-healing collodion baby have been found to have *ALOX12B* gene mutations; the majority of cases are caused by mutations in other genes.

Other Names for This Gene

- 12R-lipoxygenase
- 12R-LOX
- arachidonate 12-lipoxygenase, 12R-type
- epidermis-type lipoxygenase 12
- LX12B_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ALOX12B ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=242\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=242[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28ALOX12B%5BTIAB%5D%29+OR+%2812R-LOX%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+2880+days%22%5Bdp%5D%29>)

Catalog of Genes and Diseases from OMIM

- ARACHIDONATE 12-LIPOXYGENASE, R TYPE; ALOX12B (<https://omim.org/entry/603741>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/242>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ALOX12B\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ALOX12B[gene]))

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Genomic Location

The *ALOX12B* gene is found on chromosome 17 (<https://medlineplus.gov/genetics/chromosome/17/>).

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