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 \textit{ALOX12B} gene mutations; the majority of cases are caused by mutations in other genes.

\textbf{Chromosomal Location}

Cytogenetic Location: 17p13.1, which is the short (p) arm of chromosome 17 at position 13.1

Molecular Location: base pairs 8,072,636 to 8,087,716 on chromosome 17 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)

\textbf{Other Names for This Gene}

- 12R-lipoxygenase
- 12R-LOX
- arachidonate 12-lipoxygenase, 12R-type
- epidermis-type lipoxygenase 12
- LX12B\_HUMAN

\textbf{Additional Information & Resources}

\textbf{Educational Resources}

- The Endothelium (2011): Lipoxygenases
  https://www.ncbi.nlm.nih.gov/books/NBK57147/#s4.3.3

\textbf{Clinical Information from GeneReviews}

- Autosomal Recessive Congenital Ichthyosis
  https://www.ncbi.nlm.nih.gov/books/NBK1420
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28ALOX12B%5BTIAB%5D%29+OR+%2812R-LOX%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29%29+AND+human%5Bmh%5D+AND+%22last+2880+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- ARACHIDONATE 12-LIPOXYGENASE, R TYPE
  http://omim.org/entry/603741

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_ALOX12B.html
- ClinVar
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:242
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/O75342

Sources for This Summary

- OMIM: ARACHIDONATE 12-LIPOXYGENASE, R TYPE
  http://omim.org/entry/603741
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11773004

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/23954555

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/26298204  
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4556770/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15629692

Reprinted from Genetics Home Reference:  

Reviewed: July 2017  
Published: July 16, 2019

Lister Hill National Center for Biomedical Communications  
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