

LORICRIN gene

loricrin cornified envelope precursor protein

Normal Function

The *LORICRIN* gene is part of a cluster of genes on chromosome 1 called the epidermal differentiation complex. These genes are involved in the formation and maintenance of the outer layer of skin (the epidermis), particularly its tough outer surface (the stratum corneum). The stratum corneum, which is formed in a process known as cornification, provides a sturdy barrier between the body and its environment. Each cell of the stratum corneum, called a corneocyte, is surrounded by a protein shell called a cornified envelope.

The *LORICRIN* gene provides instructions for making a protein called loricrin, which is a major component of the cornified envelope. Links between loricrin and other components of the envelopes hold the corneocytes together and help give the stratum corneum its strength.

Health Conditions Related to Genetic Changes

Vohwinkel syndrome

At least two mutations in the *LORICRIN* gene have been identified in people with the variant form of Vohwinkel syndrome, sometimes called loricrin keratoderma. This disorder is characterized by skin abnormalities including widespread dry, scaly skin (ichthyosis), especially on the limbs. The mutations that cause the variant form of Vohwinkel syndrome change the structure of the loricrin protein; the altered protein is trapped inside the cell and cannot reach the cornified envelope. While other proteins can partially compensate for the missing loricrin, the envelope of some of the corneocytes is thinner than normal, resulting in the dry, scaly skin (ichthyosis) and other skin abnormalities associated with the variant form of Vohwinkel syndrome.

Other Names for This Gene

- LOR
- LORI_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of LORICRIN ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4014\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4014[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28LOR%5BTIAB%5D%29+OR+%28loricrin%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+1800+days%22%5Bdp%5D%29%29%29>)

Catalog of Genes and Diseases from OMIM

- LORICRIN; LOR (<https://omim.org/entry/152445>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/4014>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=LORICRIN\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=LORICRIN[gene]))

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Genomic Location

The *LORICRIN* gene is found on chromosome 1 (<https://medlineplus.gov/genetics/chromosome/1/>).

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