ABCA12 gene

ATP binding cassette subfamily A member 12

Normal Function

The *ABCA12* gene provides instructions for making a protein known as an ATP-binding cassette (ABC) transporter. ABC transporter proteins carry many types of molecules across cell membranes. In particular, the ABCA12 protein plays a major role in transporting fats (lipids) and enzymes in cells that make up the outermost layer of skin (the epidermis). This transport of molecules is needed to maintain the layers of lipids within the epidermis that are necessary to prevent water loss (dehydration) and for normal development of the skin.

Health Conditions Related to Genetic Changes

Harlequin ichthyosis

Many variants (also known as mutations) in the *ABCA12* gene have been identified in people with harlequin ichthyosis. This skin condition is characterized by hard, thick scales that are present at birth; excessive dehydration; and increased risk of infections.

Most *ABCA12* gene variants result in the production of an abnormally short protein that cannot transport lipids properly. Individuals with *ABCA12* gene variants that result in a complete absense of ABCA12 protein often have the most severe skin problems and tend not to survive past infancy. In people with abnormal or nonfunctional ABCA12 protein, a lack of lipid transport causes numerous problems with the development of the epidermis before and after birth. Specifically, it prevents the skin from forming an effective barrier, and leads to the skin problems characteristic of harlequin ichthyosis.

Nonbullous congenital ichthyosiform erythroderma

Variants in the *ABCA12* 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 dehydration. These skin abnormalities tend to be less severe than those in harlequin ichthyosis (described above).

Most of the variants that cause NBCIE change single protein building blocks (amino

acids) in the ABCA12 protein. These variants likely lead to the production of a protein with reduced function, which impairs lipid transport and the formation of the lipid layers within the epidermis. Problems with this protective barrier underlie the skin abnormalities and other features of NBCIE.

Lamellar ichthyosis

MedlinePlus Genetics provides information about Lamellar ichthyosis

Other Names for This Gene

- ABCAC_HUMAN
- ATP-binding cassette 12
- ATP-binding cassette transporter 12
- ATP-binding cassette, sub-family A (ABC1), member 12
- ATP-binding cassette, sub-family A, member 12
- ICR2B

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ABCA12 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=26154[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28ABCA12%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ATP-BINDING CASSETTE, SUBFAMILY A, MEMBER 12; ABCA12 (https://omim.org/entry/607800)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/26154)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ABCA12[gene])

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Genomic Location

The *ABCA12* gene is found on chromosome 2 (https://medlineplus.gov/genetics/chromosome/2/).

Last updated January 7, 2022