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) in cells that make up the outermost layer of skin (the epidermis). This lipid transport 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. The ABCA12 protein is also found in several other tissues, including the testes, placenta, lungs, stomach, fetal brain, and liver.

Health Conditions Related to Genetic Changes

Harlequin ichthyosis

More than 65 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 of the mutations lead to an absence of ABCA12 protein or the production of an extremely small version of the protein that cannot transport lipids properly. 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 against dehydration, and leads to the skin problems characteristic of harlequin ichthyosis.

Nonbullous congenital ichthyosiform erythroderma

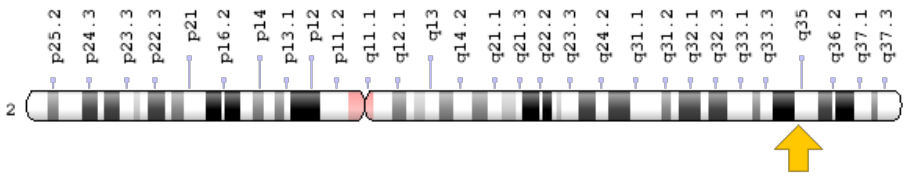
At least 20 mutations in the *ABCA12* gene have been found to cause nonbullous congenital ichthyosiform erythroderma (NBCIE). This condition affects the skin and causes redness; the development of fine, white scales; an increased risk of infections; and dehydration. These skin abnormalities tend to be less severe than those in harlequin ichthyosis (described above). Most of the mutations that cause NBCIE change single amino acids in the ABCA12 protein. These mutations 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

Chromosomal Location

Cytogenetic Location: 2q35, which is the long (q) arm of chromosome 2 at position 35

Molecular Location: base pairs 214,931,542 to 215,138,626 on chromosome 2 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

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

Educational Resources

- The Human ATP-Binding Cassette (ABC) Transporter Superfamily: ABCA Genes https://www.ncbi.nlm.nih.gov/books/NBK3/#A166

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=%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
  http://omim.org/entry/607800

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_ABCA12.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ABCA12%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:14637
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:26154
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/26154
- UniProt
  https://www.uniprot.org/uniprot/Q86UK0

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