GJB4 gene

gap junction protein beta 4

Normal Function

The *GJB4* gene provides instructions for making a protein called gap junction beta 4, more commonly known as connexin 30.3. This protein is part of the connexin family, a group of proteins that form channels called gap junctions on the surface of cells. Gap junctions open and close to regulate the flow of nutrients, charged atoms (ions), and other signaling molecules from one cell to another. They are essential for direct communication between neighboring cells.

Connexin 30.3 is found in several different tissues, including the outermost layer of the skin (the epidermis). This protein appears to play a role in the growth and maturation of epidermal cells.

Health Conditions Related to Genetic Changes

**Erythrokeratodermia variabilis et progressiva**

At least eight *GJB4* gene mutations have been identified in people with erythrokeratodermia variabilis et progressiva (EKVP), a skin disorder characterized by areas of hyperkeratosis, which is abnormally thickened skin, and temporarily reddened patches called erythematous areas. Each of these mutations changes a single protein building block (amino acid) in connexin 30.3. Studies suggest that the abnormal protein can build up in a cell structure called the endoplasmic reticulum (ER), triggering a harmful process known as ER stress. Researchers suspect that ER stress damages cells in the epidermis and leads to their premature death. The mechanisms by which epidermal damage and cell death contribute to hyperkeratosis at erythematous areas are poorly understood.
Chromosomal Location

Cytogenetic Location: 1p34.3, which is the short (p) arm of chromosome 1 at position 34.3

Molecular Location: base pairs 34,759,741 to 34,763,724 on chromosome 1 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- connexin 30.3
- connexin-30.3
- CX30.3
- CXB4_HUMAN
- EKV
- gap junction beta-4 protein
- gap junction protein, beta 4, 30.3kDa

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): Gap Junctions Allow Ions and Small Molecules to Flow between Communicating Cells
  https://www.ncbi.nlm.nih.gov/books/NBK22492/

- Madame Curie Bioscience Database: Gap Junctions: Cell-Cell Channels in Animals
  https://www.ncbi.nlm.nih.gov/books/NBK6455/

  https://www.ncbi.nlm.nih.gov/books/NBK26857/#A3494
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28GJB4%5BTIAB%5D%29+OR+%28connexin+30.3%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

Catalog of Genes and Diseases from OMIM

- GAP JUNCTION PROTEIN, BETA-4
  http://omim.org/entry/605425

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=GJB4%5Bgene%5D

- HGNC Gene Family: Gap junction proteins
  https://www.genenames.org/cgi-bin/genefamilies/set/314

- HGNC Gene Symbol Report
  https://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=4286

- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:127534

- NCBI Gene

- UniProt
  https://www.uniprot.org/uniprot/Q9NTQ9

Sources for This Summary

- OMIM: GAP JUNCTION PROTEIN, BETA-4
  http://omim.org/entry/605425


  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/23037955

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/21188847

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/26826093

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19291775

Reprinted from Genetics Home Reference: 

Reviewed: October 2018
Published: November 7, 2018

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services