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.

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

Tests Listed in the Genetic Testing Registry

- Tests of GJB4 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=127534[geneid])

Scientific Articles on PubMed

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

Catalog of Genes and Diseases from OMIM

- GAP JUNCTION PROTEIN, BETA-4; GJB4 (https://omim.org/entry/605425)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/127534)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=GJB4[gene])

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Genomic Location

The *GJB4* gene is found on chromosome 1 (https://medlineplus.gov/genetics/chromosome/1/).

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