GJB3 Gene

Subjects: Genetics & Heredity
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Gap junction protein beta 3

genes

1. Normal Function

The *GJB3* gene provides instructions for making a protein called gap junction beta 3, more commonly known as connexin 31. 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 31 is found in several different parts of the body, including the outermost layer of the skin (the epidermis) and structures of the inner ear. Connexin 31 plays a role in the growth and maturation of cells in the epidermis. The exact role of this protein in the inner ear is less clear, although it appears to be involved in hearing.

2. Health Conditions Related to Genetic Changes

2.1 Erythrokeratodermia Variabilis Et Progressiva

At least 15 *GJB3* 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 31. Studies suggest that the abnormal protein produced from certain *GJB3* gene mutations 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. Other *GJB3* gene mutations result in the production of abnormal proteins that may form channels that do not function properly, which may also lead to premature cell death in the epidermis. In addition, abnormal connexin 31 proteins may interact with other connexin proteins, preventing the formation or function of other types of gap junctions. The mechanisms by which epidermal damage and cell death contribute to hyperkeratosis and erythematous areas are poorly understood.

2.2 Nonsyndromic Hearing Loss

3. Other Names for This Gene

- connexin 31
- CX31
- CXB3_HUMAN
- DFNA2
- gap junction protein, beta 3, 31kDa
- PNHI

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