GJB6 Gene

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Gap junction protein beta 6

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1. Normal Function

The *GJB6* gene provides instructions for making a protein called gap junction beta 6, more commonly known as connexin 30. Connexin 30 is a member of the connexin protein family. Connexin proteins form channels called gap junctions that permit the transport of nutrients, charged atoms (ions), and signaling molecules between adjoining cells. The size of the gap junction and the types of particles that move through it are determined by the particular connexin proteins that make up the channel. Gap junctions made with connexin 30 transport potassium ions and certain small molecules.

Connexin 30 is found in several different tissues throughout the body, including the brain, inner ear, skin (especially the palms of the hands and soles of the feet), hair follicles, and nail beds. Because of its presence in the inner ear, researchers are interested in this protein's role in hearing. Hearing requires the conversion of sound waves to electrical nerve impulses. This conversion involves many processes, including maintenance of the proper level of potassium ions in the inner ear. Some studies indicate that gap junctions made with connexin 30 help to maintain the correct level of potassium ions.

2. Health Conditions Related to Genetic Changes

2.1 Clouston Syndrome

At least four *GJB6* gene mutations have been identified in people with a skin disorder called Clouston syndrome, which is also known as hidrotic ectodermal dysplasia 2. Characteristics of Clouston syndrome include fingernail abnormalities, hair loss, and thickened skin on the palms of the hands and soles of the feet. The *GJB6* gene mutations that cause Clouston syndrome change single protein building blocks (amino acids) in the connexin 30 protein. Although the effects of these mutations are not fully understood, they lead to abnormalities in the growth, division, and maturation of cells in the hair follicles, nails, and skin.

2.2 Nonsyndromic Hearing Loss

Researchers have identified a few *GJB6* gene mutations in individuals with nonsyndromic hearing loss, which is loss of hearing that is not associated with other signs and symptoms. Mutations in this gene cause a form of nonsyndromic hearing loss called DFNA3. This form of hearing loss can either be present before a child learns to speak (prelingual) or begin after a child learns to speak (postlingual). The hearing loss ranges from mild to profound, becomes more severe over time, and particularly affects the ability to hear high-frequency sounds.

At least two *GJB6* gene mutations have been reported to cause DFNA3. Each of these mutations changes a single amino acid in connexin 30. The mutations are described as "dominant negative" because they lead to an abnormal version of connexin 30 that appears to block the formation of functional gap junctions. A shortage of these channels may alter the level of potassium ions in the inner ear, which would disrupt the conversion of sound waves to nerve impulses.

3. Other Names for This Gene

- CX30
- CXB6_HUMAN
- DFNA3

- ED2
- EDH
- gap junction protein, beta 6
- gap junction protein, beta 6, 30kDa
- HED

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