Hystrix-like ichthyosis with deafness

Description

Hystrix-like ichthyosis with deafness (HID) is a disorder characterized by dry, scaly skin (ichthyosis) and hearing loss that is usually profound. Hystrix-like means resembling a porcupine; in this type of ichthyosis, the scales may be thick and spiky, giving the appearance of porcupine quills.

Newborns with HID typically develop reddened skin. The skin abnormalities worsen over time, and the ichthyosis eventually covers most of the body, although the palms of the hands and soles of the feet are usually only mildly affected. Breaks in the skin may occur and in severe cases can lead to life-threatening infections. Affected individuals have an increased risk of developing a type of skin cancer called squamous cell carcinoma, which can also affect mucous membranes such as the inner lining of the mouth. People with HID may also have patchy hair loss caused by scarring on particular areas of skin.

Frequency

HID is a rare disorder. Its prevalence is unknown.

Causes

HID is caused by mutations in the *GJB2* gene. This gene provides instructions for making a protein called gap junction beta 2, more commonly known as connexin 26. Connexin 26 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 neighboring cells that are in contact with each other. Gap junctions made with connexin 26 transport potassium ions and certain small molecules.

Connexin 26 is found in cells throughout the body, including the inner ear and the skin. In the inner ear, channels made from connexin 26 are found in a snail-shaped structure called the cochlea. These channels may help to maintain the proper level of potassium ions required for the conversion of sound waves to electrical nerve impulses. This conversion is essential for normal hearing. In addition, connexin 26 may be involved in the maturation of certain cells in the cochlea. Connexin 26 also plays a role in the growth and maturation of the outermost layer of skin (the epidermis).

At least one *GJB2* gene mutation has been identified in people with HID. This mutation changes a single protein building block (amino acid) in connexin 26. The mutation is thought to result in channels that constantly leak ions, which impairs the health of the cells and increases cell death. Death of cells in the skin and the inner ear may underlie the signs and symptoms of HID.

Because the *GJB2* gene mutation identified in people with HID also occurs in keratitis-ichthyosis-deafness syndrome (KID syndrome), a disorder with similar features and the addition of eye abnormalities, many researchers categorize KID syndrome and HID as a single disorder, which they call KID/HID. It is not known why some people with this mutation have eye problems while others do not.

Learn more about the gene associated with Hystrix-like ichthyosis with deafness

- GJB2

Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

In some cases, an affected person inherits the mutation from one affected parent. Other cases result from new mutations in the gene and occur in people with no history of the disorder in their family.

Other Names for This Condition

- HID syndrome
- Ichthyosis, hystrix-like, with deafness

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Ichthyosis, hystrix-like, with hearing loss (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1865234/)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Catalog of Genes and Diseases from OMIM

- ICHTHYOSIS, HYSTRIX-LIKE, WITH DEAFNESS (https://omim.org/entry/602540)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28hystrix+ichthyosis%29+AND+%28connexin+26%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

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