



# Fact Sheet

## Keratitis-Ichthyosis-Deafness Syndrome (KID) Syndrome

Information retrieved from: <http://rarediseases.info.nih.gov/qard/3113/kid-syndrome/more-about-this-disease>; <http://disorders.eyes.arizona.edu/disorders/kid-syndrome>; and [http://firstskinfoundation.org/content.cfm/Ichthyosis/KID-Syndrome-Fact-Sheet/page\\_id/899](http://firstskinfoundation.org/content.cfm/Ichthyosis/KID-Syndrome-Fact-Sheet/page_id/899)

**What is KID Syndrome?** KID syndrome is a rare genetic multi-system disorder. In the time since it was first described in 1915, only about one hundred cases have been published. Based on a review of many of these published cases, it appears that all occurrences of KID syndrome have skin abnormalities of the palms and soles and severe hearing impairment (sensorineural deafness).

**What Causes KID Syndrome?** Since KID syndrome is genetic, it can be transmitted from parent to child in an autosomal dominant way (each individual affected with the disease would have one abnormal and one normal copy of the disease gene). When, by chance, the abnormal gene copy is passed on to the offspring, the child will be affected. When the normal gene copy is transmitted, the child will be unaffected. The risk for an individual with KID syndrome to have an affected child is 50% for each pregnancy. Nevertheless, nine out of ten patients carry a new, spontaneously occurring mutation that is not present in either biological parent.

Researchers at the Department of Dermatology and Cutaneous Biology, Thomas Jefferson University in Philadelphia discovered the gene whose mutations cause KID syndrome. It is called gap junction protein beta 2 (GJB2) and is located on the long arm of human chromosome 13. This gene encodes the structural protein 'connexin-26' (Cx26), which forms gap junction channels that connect neighboring cells and permit the exchange of small molecules and ions. About 80% of KID patients carry a common mutation replacing an aspartic acid residue at position 50 of Cx26 with an asparagine. The remainder of patients usually harbors unique mutations. It is thought that the protein made from the abnormal gene copy interferes with the assembly of gap junctions and the function of normal Cx26 in a 'dominant negative' manner. Therefore, the direct cell-cell communication in the skin and other tissues, such as cornea and inner ear, can be impaired.

**What are the Signs and Symptoms of KID Syndrome?** KID syndrome is present at birth. This syndrome belongs to a group of skin disorders known as the ichthyoses due to the palms of the hands and soles of the feet typically having thickened, hardened skin. In most cases, more skin-surface areas than that of the palms and soles are affected. Symptoms include red, rough, thickened patches that are sometimes scaling. A large number of individuals develop eye problems, such as cornea defects (keratitis) causing the eyes to be extremely sensitive to light. Keratitis is an inflammation of the cornea, which is the clear dome-shaped window in front of the eye. The keratitis can be superficial (involving only the top of surface of the cornea) or involve deeper layers of the cornea. It may involve one or both eyes. Other abnormalities that may occur involve the eyes, such as neovascularization, in which small blood vessels grow from the iris over the cornea, and progressive vision loss. Also, a small number of patients may have recurrent or chronic conjunctivitis (inflammation or infection of transparent membrane that lines the eyelid and covers the sclera or the whites of the eyes).

The hearing loss associated with KID Syndrome is present at birth and is a non-progressive sensorineural loss. The severity of the hearing loss and need for amplification will need to be determined by an audiologist.

Sparse hair growth or areas of baldness (alopecia) is relatively common; but a complete lack of hair is rare. Less common abnormalities are absent or abnormal nails, recurrent infections, abnormal teeth, reduced sweating, and mental or growth delay. Some of the rare features include an increased risk for developing skin or mucus-membrane cell carcinoma or malignant scalp tumors, which may lead to early death, and life-threatening infections during the newborn period. The symptoms vary greatly between affected individuals and can change over time.

**What is the Treatment for KID Syndrome?** Because several organ systems are involved, and there is increased potential impairment of hearing, speech, and sight, patients usually require multidisciplinary treatment. The use of a lubricant in the eyes may provide significant relief from symptoms but scarring may eventually require corneal transplant surgery (penetrating keratoplasty). The threat of skin cancers and fatal liver (hepatic) failure requires monitoring throughout life. Nevertheless, the exact pathomechanisms leading to KID syndrome are still not fully understood and continue to be the subject of current and future research.

**What are the Medical and Educational Implications of KID Syndrome?** A child with KID syndrome should be followed by medical practitioners to determine his or her health needs, including eye and/or audiological care needs. The child may or may not qualify as a child with a disability based on special education eligibility criteria. Vision and/or hearing loss will need to be quantified to determine a child's need for specialized instruction and the possibility of related services.

## **Resources**

First—Foundation for Ichthyosis and Related Skin Types

[http://firstskinfoundation.org/content.cfm/Ichthyosis/About-FIRST/page\\_id/735](http://firstskinfoundation.org/content.cfm/Ichthyosis/About-FIRST/page_id/735)

## **For more information about the CO Services for Children and Youth with Combined Vision and Hearing Loss**

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*Fact Sheets from the Colorado Services to Children and Youth with Combined Vision and Hearing Loss Project are to be used by both families and professionals serving individuals with vision and hearing loss. The information applies to children, birth through 21 years of age. The purpose of the Fact Sheet is to give general information on a specific topic. More specific information for an individual student can be provided through personalized technical assistance available from the project. For more information call (303) 866-6681 or (303) 866-6605. Updated: 8/10*