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### NONSYNDROMIC DEAFNESS (7/13)

Nonsyndromic deafness is hearing loss that is not associated with other signs and symptoms. In contrast, syndromic deafness involves hearing loss that occurs with abnormalities in other parts of the body. Different types of nonsyndromic deafness are named according to their inheritance patterns.

Most forms of nonsyndromic deafness are associated with permanent hearing loss caused by damage to structures in the inner ear. The inner ear consists of three parts: a snail-shaped structure called the cochlea that helps process sound, nerves that send information from the cochlea to the brain, and structures involved with balance. Loss of hearing caused by changes in the inner ear is called sensorineural deafness.

The severity of hearing loss varies and can change over time. It can affect one ear (unilateral) or both ears (bilateral). Degrees of hearing loss range from mild (difficulty understanding soft speech) to profound (inability to hear even very loud noises). The loss may be stable, or it may progress, as a person gets older. Particular types of nonsyndromic deafness often show distinctive patterns of hearing loss. For example, the loss may be more pronounced at high, middle, or low tones. Nonsyndromic deafness can occur at any age. Hearing loss that is present before a child learns to speak is classified as prelingual or congenital. Hearing loss that occurs after the development of speech is classified as postlingual.

About 1 in 1,000 children in the United States is born with profound deafness, and another 2 to 3 per 1,000 children are born with partial hearing loss. More than half of these cases are caused by genetic factors. Most cases of genetic deafness (70 percent to 80 percent) are nonsyndromic; specific genetic syndromes cause the remaining cases. It is estimated that half of all sensorineural hearing loss occurs on a genetic basis. However, most children who are born with significant hearing loss have two parents with normal hearing and no brothers or sisters with hearing loss. Until recently, there were few tests available to determine if a child in this situation had a genetic hearing loss. Genetic hearing loss was usually identified only if another sibling had a similar hearing loss, if either or both parents had hearing loss, or if the affected child had a known genetic syndrome in which hearing loss occurs in association with other findings. However, only minorities of children with hearing loss have immediate family members with hearing loss, or an identifiable syndrome. Therefore, the presence of genetic hearing loss could not be confirmed for most families. As a result, parents seeking to understand the cause of their child's hearing loss had to be told that a genetic cause remained a possibility but could not be proven.

In adults, the chance of developing hearing loss increases with age; hearing loss occurs in half of all people older than 80 years. Overall, 1 in 10 people in the United States—more than 28 million—are currently affected by hearing loss, and this number continues to increase as the population ages.

At the present time, there is much ongoing research to determine which genes have the potential to cause hearing loss. Changes in a gene known as Connexin 26 (Cx 26) have been found to be responsible for half of all hearing loss that occurs on a genetic basis. Cx 26 is a gene that makes an important protein needed for function of certain populations of cells inside the inner ear. Changes in the building blocks that make up the Cx 26 gene may result in hearing loss, especially if inherited from both parents.

The majority of individuals with hearing loss secondary to Cx 26 have hearing loss in the severe-to-profound range. Therefore, Cx 26 is a major cause of deafness. Individuals with hearing loss secondary to Cx 26 do not have other clinical findings associated with their hearing loss. For example, they have normal X-rays of the inner ear and normal vestibular (balance) function. They do not have syndromes in which the hearing loss is associated with other disorders such as abnormalities of the kidney, heart or eye. (For example, individuals with Usher's syndrome lose their vision.)

Molecular genetic testing for hearing loss secondary to Cx 26 and other genetic causes is now available (although not paid for by insurance companies) through the Molecular Otolaryngology and Renal Research Laboratories (MORL) at the University of Iowa (<http://www.healthcare.uiowa.edu/labs/morl/otoscope/info.html>). The test called OtoSCOPE® involves taking a blood sample, that examines the individual's genes in detail and looks for changes known to cause hearing loss. Identification of a hearing loss gene as the cause of a young child's hearing loss can significantly reduce the number of tests a physician might request in order to look for other causes. As a result of the availability of this testing, many families are now having their question answered as to the reason for their child's hearing loss.