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NEWS

Modifier Gene Makes Some Hearing Loss More Severe

BETHESDA, MD (April 14, 2005) Scientists have identified a genetic mutation in humans that affects the severity of hearing loss caused by a mutation of another gene. National Institute on Deafness and Other Communication Disorders (NIDCD) scientists Drs. Julie Schultz and Andrew Griffith, co-authors at NIH and the Mayo Clinic Foundation reported their findings in the April 14 issue of *The New England Journal of Medicine*.

Genetic mutations are estimated to cause at least one half of all cases of congenital or childhood-onset hearing loss. Individual variations in the severity of hearing loss are common and typically attributed to environmental factors and modifier genes — genes that alter the clinical expression of a mutation in another gene.

In the current study, five adult siblings from the same family were found to possess a mutant form of the gene that encodes for the protein cadherin 23, which is required for the development of hair cells in the inner ear. However, the degree of hearing loss among the siblings varied. While three of the five individuals had severe to profound deafness, the other two had hearing loss only in the higher frequencies. This variability suggested the action of a modifier gene.

NIDCD scientists, led by Drs. Thomas Friedman and Konrad Noben-Trauth had previously discovered that mutations of the cadherin 23 gene cause hearing loss in humans and mice. Dr. Noben-Trauth and his co-workers had also shown that alterations of another gene, *ATP2B2*, can affect the severity of hearing loss caused by a cadherin 23 mutant gene in mice.

The mutant form of the human *ATP2B2* gene, called V586M, accounted for the more severe hearing loss in the siblings who were profoundly deaf. The two siblings with better hearing were found to have normal copies of the gene. About 1 in 20 Caucasians are carriers of V568M. deafness genes.

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Advanced Hearing Aid Center provides comprehensive audiological evaluations and hearing aid services. It is located in the Country Village Center at the corner of Long Pond Road and Maiden Lane, Greece.