

## "It Seems that Everyone in My Family Loses Their Hearing!" Results of a Study of Hereditary Factors in Adult-Onset Hearing Loss.

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### **Abstract:**

Gallaudet University, the University of Maryland School of Medicine, and SHHH are completing a study to determine the extent to which genetic factors are involved in hearing loss that occurs in adulthood. This study, the first of its kind, is funded by the National Institute on Deafness and Other Communication Disorders, National Institutes of Health. The results of the first phase of this study are described in this article.

### **Article:**

*"Both sides of my family have hearing loss, and my brother's son has a hearing loss too. I can see it just going on and on. What can we do?"*

*"Thank you so much for doing research into genetic hearing loss. My daughter is the fourth straight generation in my family with hearing loss."*

*"My husband lost his hearing gradually over the last 20 years and he is now profoundly deaf. His sister lost her hearing within a period of five years. My husband's father, his paternal aunt and their father were also deaf. As time passes I find nieces and nephews showing signs of hearing loss, and I feel that there must be some genetic factor involved."*

— Excerpts from letters of participants in the Genetic Factors in Adult-Onset Hearing Loss Study, 1992-93.

### ***The Study Design and Participants***

In September 1991, the Genetic Services Center at Gallaudet University received a grant from the National Institute on Deafness and Other Communication Disorders, National Institutes of Health, to perform a study of genetic factors involved in causing adult-onset hearing loss. Self Help for Hard of Hearing People, Inc. (SHHH) and the Division of Human Genetics at the University of Maryland School of Medicine are subcontractors on the grant. This is the first study of this type performed on a large group of people with adult-onset hearing loss.

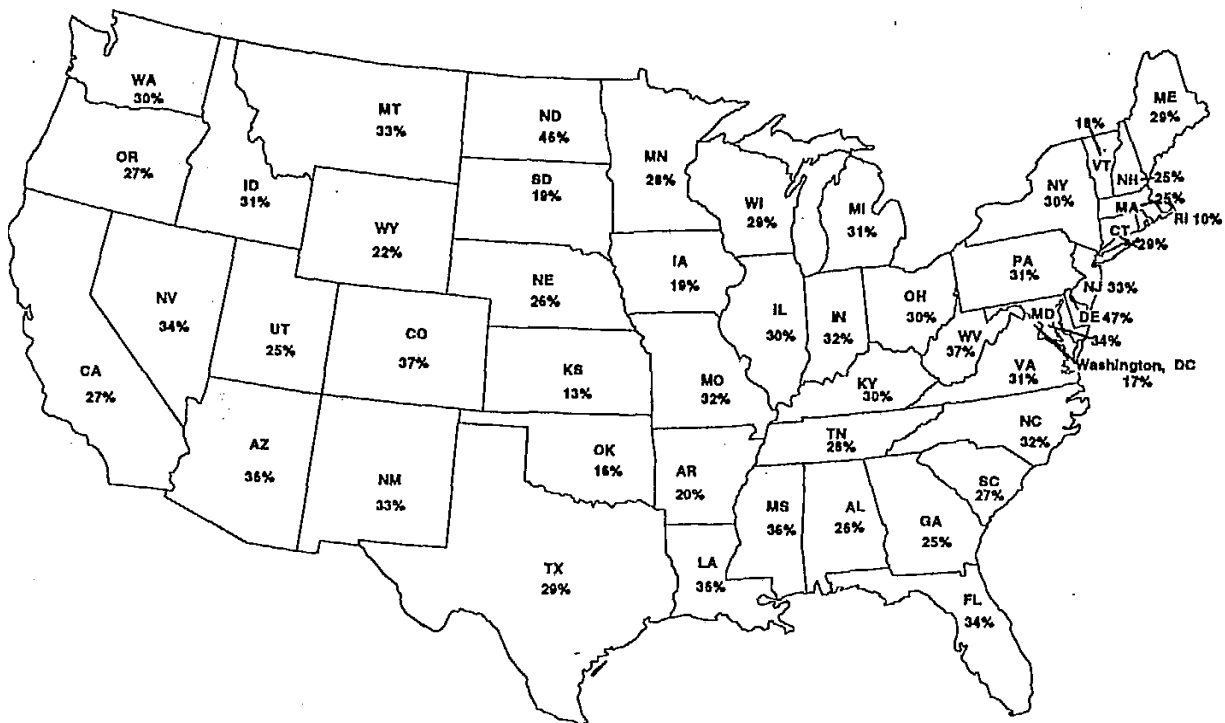
Although we know that 50.60 percent of children who are deaf at birth or at an early age have genetic causes of deafness, we do not know what percent of individuals who develop hearing

loss in adulthood (after the age of 20) have genetic causes of hearing loss. Genetic (hereditary) hearing loss is inherited in families through one of several mechanisms. Often a person with hereditary hearing loss has close relatives who also have hearing loss, but sometimes they do not.

In November 1991, a short questionnaire was sent to 11,200 SHHH members. This questionnaire asked about the characteristics of the hearing loss (degree, progression and type) and about the cause of the hearing loss. Questions about other family members with hearing loss were also included. About 36 percent (4,039 SHHH members) returned the questionnaire.

The geographic distribution of the respondents (those people who returned the questionnaire) from the SHHH membership is shown with the map of the United States in Figure 1. Most of the respondents were between 41.80 years of age and about 60 percent were female. Analysis of the data from these questionnaires has given us information about audiometric characteristics and some important clues about the importance of genetic factors as a cause of adult-onset hearing loss. The full results of this study have been submitted for publication in a journal for genetics professionals.

**Figure 1**  
**State-Specific Response Rates to the Hearing Loss Survey**  
(Number of questionnaires returned from state/number of questionnaires sent to state SHHH members.)



#### *Audiometric Characteristics*

The study participants were asked several questions about their hearing loss, including the age at which they first noticed the hearing loss, whether the level of the hearing loss is equal (symmetric) in both ears, the type of hearing loss (conductive [bone] or sensorineural [nerve]), and the frequencies affected by the hearing loss. This information was important so that we could relate these findings to the reported cause of the hearing loss and the family history information.

Most of the respondents were up-to-date with their audiologic testing. In fact, about 80 percent reported having an audiogram within the past three years.

Of the responding members of SHHH, 87 Percent reported having "nerve" loss, or a sensorineural hearing loss. This means that the problem lies either in the cochlea of the inner ear, or in the nerve beyond the cochlea. Conductive loss ("bone") refers to a loss which occurs due to changes in the middle ear; only 2.5 percent of responding SHHH members reported "bone" as the only type of hearing loss present. A mixed loss includes both sensorineural and conductive components and was reported by 14 percent of the individuals responding.

Among those members with sensorineural losses, high frequency losses outnumbered low frequency losses by six to one. Forty-five percent of the people reporting conductive losses had more impairment in the low frequencies or had equal loss across frequencies, while 37 percent reported worse hearing in the high frequencies. Almost all of the SHHH respondents have used or are using a hearing aid. Of these, the majority felt that their hearing aids were beneficial, while fewer than 5 percent did not feel they were useful.

The age at which the hearing loss was noticed is often different from when the hearing loss actually began (the age of onset). If the hearing loss began and progressed gradually, it may not have been "noticed" until many years after the actual age of onset. The same is true for a hearing loss present at birth, which may not be diagnosed for months or even years. It appears from our data that as age increases, more men begin noticing a hearing loss.

Another question pertaining to audiologic status of the respondents was whether the hearing loss was symmetric (same in both ears) or asymmetric (different in each ear). Since about 250 SHHH members sent their audiograms along with the questionnaires, we were able to judge the accuracy of the responses to that question. It appears that about 67 percent (about two-thirds) of the respondents accurately reported symmetry or asymmetry of their hearing losses. Out of the entire group, 43 percent reported a symmetric hearing loss, and 57 percent reported an asymmetric loss.

We then compared the reported cause of the hearing loss to the symmetry of the loss. A greater number of asymmetric losses were reported along with the following causes of hearing loss: injury, cancer, ear infection, childhood disease, Meniere's disease, surgery, otosclerosis, and autoimmune disorders. The majority of those reporting heredity or ototoxicity (hearing loss due to medications) as a cause, reported symmetric hearing losses.

### *Cause of the Hearing Loss*

It is not uncommon for the cause of a hearing loss to be unknown or uncertain. For this reason, a cause that is reported on a questionnaire in the absence of a medical diagnosis, must be carefully considered. In most cases, the cause of the hearing loss occurs at the same time that the hearing loss is noticed. For instance, a hearing loss that is not noticed until the age of 33 is not likely to have been caused by childhood ear infections or childhood disease.

A notable exception to this is certain medications known to be toxic to the ear (such as dihydrostreptomycin and neomycin), which may cause a hearing loss that does not begin immediately or continues for months after the medication is discontinued. It is possible that an

early, mild loss, or one of very limited frequency involvement, may not be noticed until combined with presbycusis (hearing loss associated with aging).

It is often difficult to recognize that the cause of hearing loss may be hereditary (genetic). Hereditary hearing loss is passed down through the family in one of several patterns. Genetic factors can cause conductive, sensorineural or mixed hearing losses, and are associated with all frequencies of hearing loss, from low to high.

Knowledge about hereditary hearing loss is not widespread among medical professionals; in fact, they may not recognize that a hearing loss is hereditary even when several other family members are noted to have hearing loss. Many times, a person can have a hereditary hearing loss and be the first person with recognized hearing loss in the family. This makes the cause of the hearing loss even more difficult to identify.

Reports of the cause of hearing loss revealed some interesting information, which is displayed in Figure 2. A large proportion of respondents felt their hearing loss had a genetic origin. The auditory diseases category included Meniere's disease and otosclerosis, which typically occur in adulthood. In fact, otosclerosis is known to be hereditary; and of the respondents who reported having otosclerosis, it was more likely to be found in their male family members than in their female family members. Only a small number of the respondents reported old age as the cause of their hearing loss; most respondents first noticed their hearing loss before the age of 60.

**Figure 2**  
**Self-Reported Causes of Hearing Loss in SHHH Members**  
**Early- Versus Late-Onset Hearing Loss**

	Age Onset Greater Than 20 (1,472 People)	Age Onset Less Than or Equal to 20 (821 People)
<b>Genetic</b>	29.4%	27.0%
<b>Specific Auditory Diseases</b> (e.g., Meniere's Disease, Otosclerosis, ear infection)	21.7%	15.2%
<b>Exposure to Extreme Noise</b>	15.9%	2.4%
<b>Environmental Insult</b> (e.g., head injury, surgery, radiation, birth injury or trauma, prematurity, ototoxicity)	10.3%	13.6%
<b>Other</b>	7.9%	5.8%
<b>Systemic Infections/Diseases</b> (e.g., autoimmune disease, neoplasms, childhood diseases, other systemic illnesses, in utero exposures)	7.3%	26.0%
<b>Presbycusis</b>	5.3%	1.0%
<b>Congenital</b>	2.1%	8.6%

#### *Family History of Hearing Loss*

One way to determine that a person has hereditary hearing loss is to examine the number of deaf or hard of hearing relatives the person has and their exact blood relationship to that person. Of

those respondents who reported childhood onset of their hearing loss, about half had at least one parent with a hearing loss. Surprisingly, 62 percent of those with adult-onset hearing loss had at least one parent with hearing loss. This may indicate that many of the people with adult-onset hearing loss who answered the survey have hereditary types of hearing loss inherited from a parent. However, this is difficult to prove because we do not know the exact type of hearing loss present in the parents of these individuals.

About 45 percent of early- and late-onset groups had a sibling (brother or sister) with a hearing loss. This finding surprised us and may be another indicator that many people with adult-onset hearing loss actually do have a genetic cause of hearing loss. Those individuals who had other deaf or hard of hearing relatives tended to have developed or noticed their own hearing loss at a comparatively younger age. As the age of onset of hearing loss in the respondents increased, so did the age of onset in parents. Of those reporting a genetic cause for their hearing loss, about the same number of mothers as fathers were also reported to have hearing loss. More than 74 percent of the respondents reported having a relative other than a parent or sibling who also had a hearing loss.

Although we could not estimate the percentage of hearing loss that was actually due to genetic factors or the exact modes of inheritance responsible, we collected large amounts of evidence to document that genetic factors do indeed have a significant role in causing adult-onset hearing loss. This is a new finding and shows that further investigation is needed. As our knowledge of genetics increases we are learning that some forms of hearing loss are caused directly by different genes, while other types of hearing loss are caused indirectly by genes which may make a person more susceptible to losing their hearing because of exposure to strong medications, noise, or other factors.

### *Where Do We Go From Here?*

Our findings indicate that further studies are needed to learn more about the exact genetic mechanisms that are involved in causing adult-onset hearing loss. The next phase of our study is already underway. In January 1993, we mailed a detailed nine-page questionnaire to 1,000 individuals who responded to the first questionnaire. We knew that such a long questionnaire would be difficult to fill out; we asked the respondents to complete detailed information on five generations of their family.

However, we were delighted and overwhelmed with the response to this follow-up questionnaire; more than 600 people (62 percent) responded! The information from this survey will be helpful to us in analyzing inheritance patterns of hearing loss in several families and perhaps even in identifying which genes may be involved. The results of this phase of our study will appear in a future issue of the SHHH Journal. We have applied for additional funding from the National Institutes of Health to perform more detailed studies on some families — including DNA analysis to search for the genes which cause hereditary adult-onset hearing loss. We hope that our exciting work will continue to expand. We are very grateful to the many SHHH members who took the time to help us with this study.

*Susan L. Phillips is a Ph.D. candidate in audiology at the University of Maryland, College Park. Melissa J. Stick is a Ph.D. candidate, and Anne M. Sill, Valerie L. Prenger, Ph.D., and Joann A. Boughman, Ph.D., are genetics researchers in the Division of*

*Human Genetics at the University of Maryland School of Medicine. Kathleen S. Amos, Ph.D., is the director of the Genetic Services Center at Gallaudet University. For further information on the study described in this article, please write to Dr. Amos at the Genetic Services Center, Gallaudet University, 800 Florida Avenue, N.E., Washington, D.C. 20002.*

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