Ongoing Research Projects With CDC-EHDI Funding

Genetic Services for Congenital Hearing Loss - University of North Carolina* at *Chapel Hill

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Background: Nationwide, approximately 90% of newborns in the United States are now screened for hearing loss shortly after birth, with approximately 1 to 3 out of every 1,000 births diagnosed with a permanent congenital hearing loss. Studies on hearing loss suggest that genetic causes account for at least 50% of childhood-onset hearing loss. The American College of Medical Genetics recommends that all children with a congenital hearing loss be offered a comprehensive genetic evaluation.

As universal newborn hearing screening and genetic testing for hearing loss become more widely practiced, it is important to identify factors that limit and facilitate the access of patients and families to genetic services, how families understand and use genetic information, and what factors might add to parental anxiety. Identifying these factors can lead to the development of tools to overcome them, such as improvement in accessibility of genetic services, education of primary care and specialty health care providers, and informational resources for families.

Purpose: The foci of this project are to (1) determine the number of infants with congenital hearing loss identified through the North Carolina Newborn Hearing Screening Program who receive a comprehensive genetic evaluation; (2) determine what factors limit or facilitate access to genetic services for infants with congenital hearing loss; and (3) assess families' experiences with and attitudes toward genetic services and their understanding of genetic information they receive.

Methods: This project is being accomplished in two phases. Phase I of the study consists of obtaining population data on how many infants identified with congenital hearing loss through the state newborn hearing screening program have had a genetic evaluation. In this phase, the researchers will also gather data on the factors that determine access to genetic services. During phase II of the study, the researchers will use semi structured interviews to obtain additional information from parents whose children have had a genetics evaluation, genetic testing for hearing loss, or both. In this phase, researchers will study parents' understanding of genetic information provided, and parental attitudes regarding this information.

Current Status: In-depth interviews have been completed with 19 families, ascertained through the original surveys that were completed and through a genetics clinic. Many of the original surveys were returned due to incorrect addresses, which contributed to a lower than expected response. As a result, researchers have partnered with the North Carolina based Beginnings for Parents of Children Who Are Deaf or Hard of Hearing, Inc. (i.e., Beginnings), which is a nonprofit organization that was established to provide emotional support and access to information for families of children with hearing loss from birth through 21 years of age. This organization maintains a database with current addresses of families. Through this collaboration, Beginnings will mail out surveys that were prepared by the researchers to families served by the agency (*Updated 10/2006*).