Completed Research Projects With CDC-EHDI Funding

Epidemiology and Etiology of Hearing Loss in Newborns and Young Children - Utah Department of Health, Hawai'i Department of Health, Rhode Island Department of Health

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Background: Hearing loss can occur late in life, or be present at birth, as is the case for 1-3 out of every 1,000 newborns. Formerly, the majority of these children were not diagnosed until they were 2 to 3 years of age, resulting in communication, social, and cognitive delays. Technological advances and increased public health attention have resulted in the development of Early Hearing Detection and Intervention (EHDI) programs. The goals of EHDI programs are to screen newborns for hearing loss by 1 month of age, provide diagnostic evaluations by 3 months of age to those with positive screening results, and provide appropriate intervention services before 6 months of age to those with diagnosed hearing loss.

Previous studies on hearing loss suggest that genetic causes account for at least 50% of childhood-onset hearing loss. As a consequence, genetic evaluations are often included as part of the follow-up services. About 70% of this genetic hearing loss in young children is non syndromic. Autosomal recessive inheritance represents the most common etiology. Determination of the etiology of prelingual hearing loss does not routinely occur in most hearing screening programs. Moreover, the clinical infrastructure to evaluate newborns and infants identified through such detection programs has not been established.

Purpose: The purpose of this study was to describe the epidemiology of hearing loss in infants and young children with a permanent hearing loss of any type. The specific aim of this study was to document the etiology of hearing loss for all children identified before their third birthday.

Methods: Children with hearing loss who took part in this study were examined by a medical doctor who was an expert in clinical genetics. The parent or guardian of each child was asked about the child's family history. Using the child's examination and family history, the physician attempted to determine the cause of the hearing loss. The parents or guardians were given the opportunity to have their child tested for several genes that can cause hearing loss. A genetic counselor explained the results of the tests to the parents or guardians. Testing was voluntary and free.

Summary of Results: This is the first study to determine the causes of hearing loss in a population ascertained through newborn hearing screening. Currently, more than 170 infants and family members are enrolled and have been evaluated. Approximately 100 infants have been found to have nonsyndromic, sensorineural hearing loss. Test results of a mutation in the GJB2 gene (accounting for up to 50% of nonsyndromic autosomal recessive deafness in some populations), the GJB6 gene, and mutations of two mitochondrial genes are pending on most of these children. Final study results are being prepared. (*Updated 10/2006*)