

Diagnostic of Pediatric Hearing Impairment – Deafness Gene Chip

TECHNICAL FIELD

Diagnostic: Deafness Gene Chip (2002-0412)

BACKGROUND

Congenital hearing loss represents one of the most common birth defects in the United States and the prevalence of permanent congenital hearing loss is approximately 1.2 per 1000 live births. Conductive hearing loss involves transmission of vibrations to the inner ear, and sensorineural hearing loss occurs when there is damage to the inner ear or to the nerve pathways from the inner ear to the brain. Nearly 80% of infant hearing loss cases are due to sensorineural hearing loss and half of these cases are genetic in origin.

While hearing loss in infants can go undetected for months after birth, early detection and intervention can alleviate most of the developmental and behavioral difficulties found in hearing impaired children. The earlier the intervention occurs, the greater the enhancement of speech and language skills. With special education cost estimates for such late-identified hearing impaired children ranging from \$38,000 to \$220,000 over a K-12 education, total societal costs (special education and lost job productivity) for such an individual approaches \$1 million.

Early, accurate, and specific genetic testing will enable accurate and rapid diagnoses and identification of children for whom early intervention will provide critical benefits and potentially optimize treatment options.



TECHNOLOGY

It is estimated that in the U.S., approximately 40,000 children are born with sensorineural hearing loss and that 80% of these children are candidates for the microarray screening. It is believed that twenty-five percent of these U.S. cases (8,000) may be captured through diagnostic screens. Dr. John Greinwald of the Cincinnati Children's Research Foundation has demonstrated that diagnostic gene chip arrays can be used in pediatric screening for hearing loss. The gene chip uses microarray technology developed within the past decade to provide a rapid and accurate means for analyzing nucleic acid samples.

Microarray technology provides a means to test for the genetic causes of current and potential future hearing loss in infants. The resulting diagnostic test can be used to allow early intervention of hearing loss in infants.

APPLICATIONS

1. Gene-based test for diagnosis of pediatric hearing loss
2. Research tool

ADVANTAGES

- Rapid, accurate diagnostic
- Standardized diagnostic tool
- Early intervention and better disease management
- Large potential savings to health care system

INVESTIGATOR

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STATUS

Patent applications pending.

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THE INVENTOR

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BACKGROUND

BS: Wofford College, Spartanburg, SC (1983)

MD: Medical University of South Carolina, Charleston, SC (1987)

Internship: Surgical Intern, Naval Medical Center, Portsmouth, VA (1988)

Residency: Otolaryngology, Naval Medical Center, Portsmouth, VA (1995)

Fellowship: Pediatric Otolaryngology, University of Iowa, Iowa City, IA (1998)

Most Recent Publications

Wang Q, Li R, Zhao H, Peters JL, Liu Q, Yang L, Han D, Greinwald JH, Young, WY and Guan MX. **Clinical and molecular characterization of a Chinese patient with auditory neuropathy associated mitochondrial 12S rRNA T1095C mutation.** *Am. J. Med. Genet*; 133A:27-30, 2005.

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Preciado D, Lim L, Cohen A, Madden C, Myer D, Ngo C, Bradshaw J, Lawson L, Choo D, Greinwald J. **A diagnostic paradigm for childhood idiopathic sensorineural hearing loss.** *Otolaryngology Head and Neck Surgery*; 131:804-809, 2004.

Salamone F, Bobbitt B, Myer C, Rutter M, Greinwald J. **Bacterial tracheitis reexamined: Is there a less severe manifestation?** *Otolaryngology Head and Neck Surgery*; 131:871-876, 2004.

Guo Y, Pilipenko V, Lim LH, Dou H, Johnson L, Srisailapathy CR, Ramesh A, Choo DI, Smith RJ, Greinwald JH. **Refining the DFNB17 interval in consanguineous Indian families.** *Molecular Biology Report*; 31:97-105, 2004.

Li, R, Greinwald, JH, Yang, L, Choo, DI, Wenstrup, RJ and Guan, MX. **Molecular analysis of mitochondrial 12S rRNA and tRNAser(UCN) genes in sporadic pediatric subjects with nonsyndromic hearing loss.** *Journal of Medical Genetics*; 41:615-20, 2004.