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Genetic deafness: From molecular level to patient care

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We have established the Miami Ontogenetic Program combining the research and the clinical component. The program has provided a unique platform to carry out translational research on delivering genetic services to deafness patient care. The extreme heterogeneity of non-syndromic sensorineural hearing loss (NSHL) makes serial sequencing approaches unfavorable in terms of efficiency and cost. However, the discovery that genes at only 2-3 loci account for a major component of human deafness suggested that the sequential screening of DNA samples from probands in multiplex sibships for mutations would be a cost effective strategy. We aim to identify the genetic cause of NSHL through an integrated paradigm combining microarray, copy number variations (CNVs) analysis, whole genome sequencing (WES) and a hearing-centric database. We are using a DNA microarray panel (Miami-CapitalBio) as the initial screening to simultaneously detect the most common deafness-causative mutations from four genes. We then perform a custom capture/next-generation sequencing gene panel (MiamiOtoGenes) composed of 180 known deafness genes (Agilent SureSelect DNA Design). Patients for whom the two panels do not provide a meaningful result, WES is performed to achieve a comprehensive interrogation of the full spectrum of variants to detect single-nucleotide variants (SNVs), insertion/deletions (Indels) and CNVs. If variants are not found in the genes included on Miami-CapitalBio and MiamiOtoGenes, WES should be considered in small multiplex families. The multidisciplinary team approach is an effective way to bring the sequencing data to clinical practice for the clinical diagnosis and management of deaf and hard-of-hearing families.

Biography

Xuezhong Liu an internationally renowned Surgeon-Scientist is the Leonard M. Miller Professor and Vice Chair of Otolaryngology. He has had a career long interest in genetic deafness and has made many significant contributions. He is the author of more than 200 scientific papers in top journals. His exemplary translational research on hereditary hearing loss from basic sciences to clinical application (bench to bedside) for the past three years, ranking in the top 1 percent of NIH-funded physician-scientists in the auditory field.

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