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Genomic foundation of sensorineural hearing loss

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**Abstract (approximately 250-300 words limit)**

Sensorineural hearing loss (SNHL) is a common sensory disorder with a significant Mendelian genetic component. Despite comprehensive genetic testing like exome sequencing, about 50% of SNHL cases remain genetically unresolved. The role of whole-genome sequencing (WGS) in diagnosing these unresolved cases is not well established. This study aims to assess the diagnostic and clinical value of WGS in SNHL.

We investigated the genetic basis of SNHL in 394 families and 746 individuals, including probands (183, 46.2% male; median age 13, range 0-76) and their relatives. Initially, we achieved a complete genetic diagnosis in 220 families (55.86%) using standard genetic tests. For cases still undiagnosed, WGS was performed on 120 patients (both syndromic and a subset of nonsyndromic SNHL) selected through a stratified sampling approach.

WGS identified causal variants in 24 of the 120 families (20.0%). Notably, 14 of these (11.7%) included deep intronic variants, small structural variants, and complex genomic rearrangements that were not detectable with previous methods. WGS proved most beneficial for patients with specific features such as early onset, syndromic characteristics, and those analyzed through trio-based WGS.

This study demonstrates that WGS can enhance the diagnostic yield for SNHL by approximately 12% beyond what is achieved with exome sequencing and other techniques. It confirms the clinical utility of WGS in diagnosing SNHL and highlights its potential in advancing precision medicine by providing a detailed genome-phenome landscape and uncovering genotype-phenotype correlations.

**Keywords**

Sensorineural hearing loss; Whole-genome sequencing; Genetic diagnosis; Deep intronic variants; Structural variants; Precision medicine; Genotype-phenotype correlation

**Recent Publications:** Minimum 3 publications to be included (Not mandatory)

**Biography** **(150 words limit)**

**Myeongsin Kang, M.D., is an Assistant Clinical Professor in the Department of Otorhinolaryngology at Seoul National University Hospital, specializing in otology. With a deep commitment to advancing medical science, Dr. Kang actively engages in research to improve diagnostic and therapeutic approaches in the field of hearing and balance disorders. As a junior faculty member, Dr. Kang is dedicated to contributing to the progress of otology through clinical practice and academic endeavors.**

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