

CDB SEMINAR

Thomas Friedman

NIH/NIDCD

Monday, February 15, 2010 16:00~17:00 C1F CDB Auditorium

Targeted capture and next-generation sequencing identifies TPRN, encoding TAPERIN, as the mutated gene in nonsyndromic human deafness DFNB79

Summary

Hereditary hearing loss is a common genetically and phenotypically heterogeneous human disorder. My laboratory at the National Institute on Deafness and Other Communication Disorders at the National Institutes of Health is focused on a systematic and comprehensive genetic approach to the identification of genes necessary for hearing and a determination of their functions in the inner ear. To accomplish this goal we first ascertain families segregating hearing loss, and map and positionally clone the causative genes. Since the phenotype directs us to the gene, our classical genetic strategy is inherently unbiased by preconceptions of the components necessary for hearing. The seminar will be a discussion of a novel strategy for positionally cloning a "deafness gene" at a locus on chromosome 9 designated *DFNB79*. Recent unpublished data on the *DFNB79* gene and its possible function will be discussed.

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