

CDB SEMINAR

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Wednesday, November 12, 2014 16:00-17:00 Auditorium C1F

The Genomics of Deafness: Exploring Gene Function and Regulation

Summary

The field of the genetics of deafness has undergone a remarkable transition in recent years. Genomic tools enable deep sequencing of all DNA and RNA molecules in a cell and are being applied to studying the auditory and vestibular systems. In the context of DNA, deep sequencing is being used to discover new genes for deafness, as well as rapidly identify variants associated with this sensory disorder. Approaches combining targeted genomic capture of known deafness genes and deep sequencing, as well as whole exome sequencing, has doubled the number of genes and mutations for deafness. The speed and low cost has made this approach feasible for incorporating into clinical diagnosis protocols. In the context of RNA, deep sequencing is being used to discover and characterize microRNAs and lincRNAs, key regulators in multiple cellular processes. Additional layers of function and regulation are available by dissecting the full repertoire of the transcriptome, including open chromatin structure, histone modifications and DNA methylation of cells derived from the inner ear. Together, these tools facilitate functional genomic studies towards a comprehensive understanding of auditory and vestibular function and the pathology associated with deafness.

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