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Genomic Analysis of Hereditary Hearing Loss

Positions

Professor, Sackler Faculty of Medicine

Vice Dean for Preclinical Affairs, Sackler Faculty of Medicine

Scientific Board Member, I-CORE for Gene Regulation in Complex Human Disease

President, Federation of the Israel Societies for Experimental Biology (ILANIT)

President, Israel Society of Auditory Research

Associate Editor, *European Journal of Human Genetics*

Research

Our primary interest is the genetic basis of hereditary hearing loss or deafness. Our group is working towards the identification, characterization and regulation of genes associated with hereditary hearing loss. For gene discovery, we focus on the Israeli Jewish and Palestinian Arab populations in the Middle East. Our studies have encompassed the prevalence of connexin 26 mutations in these populations, the most common form of deafness, to the identification of mutations in over 30 genes, since this is a genetically heterogeneous disease. We are employing deep sequencing, also known as massively parallel sequencing, to identify mutations using the latest genomic technology. Our work has provided the link between gene discovery and clinical diagnosis in genetic clinics in medical centers throughout Israel. In addition, we have studied the auditory and vestibular systems of a dozen mouse mutants, focusing on mutation identification, morphological and functional analysis of the organ of Corti and its cells, and behavioral analysis of hearing and balance disorders. This has allowed us to define the pathways leading to deafness in mouse models for human deafness. Most recently, we have demonstrated that microRNAs are essential for development and function of inner ear hair cells in vertebrates through microRNA expression, mouse mutants and target identification.

Publications

Manuscripts

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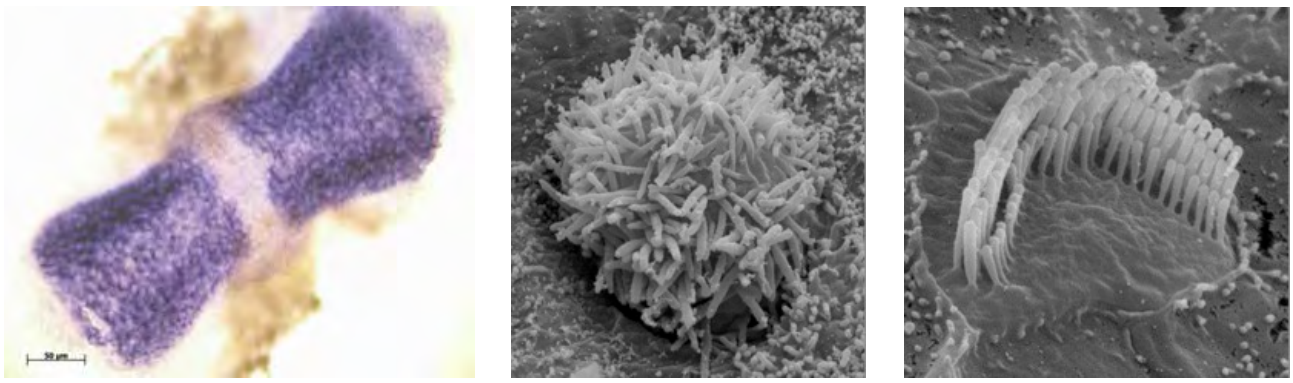

Wild type and mutant hair cell bundles in the PCKO mouse, lacking microRNAs in the inner ear, demonstrated by scanning electron microscopy (2 left panels). *In situ* hybridization reveals expression of the microRNA-182 in the inner ear crista (right).

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Grants

2011 – 2015 Gene Expression and microRNA Regulation in Hair and Supporting Cells of Mouse, Israel Science Foundation

2011 – 2016 Gene Discovery for Hearing Loss in Middle East by Massively Parallel Sequencing, National Institutes of Health, Co-PI: Moien Kanaan

2012 – 2015 Morphodynamics of Mammalian Planar Cell Polarity - a Quantitative Approach, Human Frontier Science Program, Co-PIs: Ping Chen, David Sprinzak, Fumio Matsuzaki

2014 – 2017 Epigenetic Regulation in the Mammalian Inner Ear. Binational Science Foundation. Co-PI: R. David Hawkins.