

GENETIC HEARING LOSS – From Diagnosis to Gene Therapy

Monday October 19, 2020
10:00am-5:30pm - VIA ZOOM

6th Annual Auditory & Vestibular Translational Research Day

Organized by the Center for Comparative and Evolutional Biology of Hearing, of the University of Maryland

Time	Speaker	Title
10:00-10:15	<p>Ronna Hertzano, MD PhD Department of Otolaryngology Head and Neck Surgery & Institute for Genome Sciences University of Maryland School of Medicine, Baltimore Faculty page: Hertzanolab.org</p> <p>Catherine Carr, PhD Distinguished University Professor Department of Biology University of Maryland, College Park Faculty page: http://terpconnect.umd.edu/~cecarr/</p>	Introduction to the Day & Opening remarks
10:15-11:00	<p>Thomas B. Friedman, PhD Acting Scientific Director NIDCD Chief, Laboratory of Molecular Genetics, NIDCD, NIH</p> <p>Faculty page: https://www.nidcd.nih.gov/about/staff/thomas-b-friedman-phd</p>	<p>The Genetic Landscape of Human Hearing Loss Understanding the genetics of human deafness and a discussion of criteria for causality and the challenges of identifying the real mutation associated with deafness.</p> <ul style="list-style-type: none"> Obtaining knowledge of human inherited deafness Differentiating between pathogenic and benign mutations Distinguishing feeble from definitive data of mutations associated with human deafness Recognizing noncoding variants associated with human deafness
11:00-11:15	Short Break	
11:15-11:45	<p>Hela Azaiez, PhD Molecular Otolaryngology and Renal Research Labs (MORL), Department of Otolaryngology, University of Iowa, USA</p> <p>Faculty page: https://morl.lab.uiowa.edu/people/hela-azaiez-phd</p>	<p>Genetic Diagnosis for Deafness: Approaches, Challenges, Opportunities and Perspectives Implementing comprehensive genetic testing and deafness-specific knowledge is key to empower clinical decision-making and enhance our understanding of Deafness biology.</p> <ul style="list-style-type: none"> Recognize the need for comprehensive genetic testing for individuals with hearing loss Appreciate the challenges inherent in genetic variant classification and the essential role for an expert panel in interpreting disease-specific phenotypic-and-genotypic data to generate accurate clinical reports Appreciate how a refined understanding of variant effect improves our understanding of disease biology
11:45-12:15	<p>Carmen C. Brewer, AuD PhD Chief, Audiology Unit, Division of Intramural Research, NIDCD</p> <p>Faculty page: https://www.nidcd.nih.gov/about/staff/carmen-c-brewer-phd</p>	<p>Clinical Correlates to Genetic Hearing Loss – Applying Molecular Genotyping to Patient Care This talk will discuss the application of genetic diagnoses of hearing loss and the corresponding phenotypes to patient care, including patient presentation, variation and counseling.</p> <ul style="list-style-type: none"> Will be able to provide an example of the impact of a genetic diagnosis on patient management Will be able to describe phenotypic variability within a single genetic diagnosis Will be able to describe how knowledge of the phenotype of hearing loss syndrome may lead to genetic diagnosis
12:15-1:30	Lunch Break	
1:30-2:10	<p>Rick A. Friedman, MD PhD Vice Chief, Division of Otolaryngology, UCSD Professor of Surgery Director of UCSD Acoustic Neuroma Center UCSD Health/School of Medicine</p> <p>Faculty page: https://providers.ucsd.edu/details/32904/ent-head-and-neck-(otolaryngology)-surgery</p>	<p>Genetic Association Studies in Human and Mouse – a Unique Opportunity to Understanding Age- and Noise-induced Hearing Loss This presentation will provide an overview of complex traits and genome-wide association studies as they pertain to hearing loss.</p> <ul style="list-style-type: none"> Understand complex traits and their analyses. Understand the fundamentals of GWAS. From GWAS to candidate genes, eQTLs and model organisms.
2:10-2:25	Short Break	
2:25-3:30	<p>Larry Lustig, MD Howard W. Smith Professor and Chair Department of Otolaryngology-Head & Neck Surgery Columbia University Vagelos College of Physicians and Surgeons New York-Presbyterian/Columbia University Irving Medical Center</p> <p>Faculty page: https://www.entcolumbia.org/profile/lrlustig</p>	<p>Cochlear Gene Therapy for Genetic Deafness This talk will summarize the advances in Cochlear gene therapy for genetic deafness, focusing on several forms including deafness caused by mutations in VGLUT3, Otoferlin and Clarin-1.</p> <ul style="list-style-type: none"> To understand how mutations in genes can lead to deafness To understand how gene therapy has been used to treat some forms of Usher syndrome To understand how gene therapy can be adapted to larger genes using dual vector approaches
3:30-3:45	Short Break	
3:45-4:40	<p>Panel Discussion & Quiz Karen Avraham, PhD; Hela Azalez, PhD; Carmen Brewer, AuD, PhD; Wade Chien, MD Thomas B. Friedman, PhD; Larry Lustig, MD and Ronna Hertzano, MD, PhD</p>	<p>After an Interactive Quiz With Prizes, We Will Brainstorm Hot Topics in Genetic Hearing Loss Stay with us for this exciting and interactive session with active viewer participation via KAHOOT!</p>
4:40-4:50	<p>Sandra Gordon-Salant, PhD Professor and Director of the Doctoral Program in Clinical Audiology in the Department of Hearing and Speech Sciences University of Maryland, College Park Faculty page: http://www.umdhearinglab.com/</p>	Closing Remarks
4:50-5:00	Short Break	
5:00-5:30	<p>Genetic Tools Workshop</p> <p>Ronna Hertzano, MD PhD Hela Azaiez, PhD</p>	A walk-through of available genetic, genomic and transcriptomic resources for scientists, clinicians and genetic counselors who care for persons with hearing loss.