Welcome to the 6th Annual Auditory & Vestibular Translational Research Day!

Organized by:

Ronna Hertzano, MD, PhD
University of Maryland SOM, Baltimore

Catherine Carr, PhD
University of Maryland College Park

Sandra Gordon-Salant, PhD
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Hela Azaiez, PhD
University of Iowa

Special thanks to our UMSOM students:

Kathleen Gwilliam

Webinar/ Zoom coordination

Kevin Rose

Workshop & Audio/Vis Construction

Erika Lipford, MSc

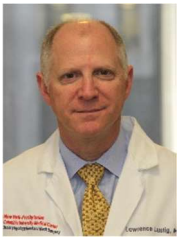
KAHOOT construction

Benjamin Shuster

Soundtracks & KAHOOT

Schedule:

Time	Speaker	Title
10:00-10:15  	**Ronna Hertzano, MD, PhD** *Department of Otolaryngology Head and Neck Surgery* *Department of Anatomy & Neurobiology* *Institute for Genome Sciences* *University of Maryland School of Medicine, Baltimore* *Faculty page: Hertzanolab.org* **Catherine Carr, PhD** *Distinguished University Professor* *Department of Biology* *University of Maryland, College Park* *Faculty page: http://terpconnect.umd.edu/~cecarr/*	**Introduction to the Day & Opening remarks**
10:15-11:00 	**Thomas B. Friedman, PhD** *Acting Scientific Director NIDCD* *Chief, Laboratory of Molecular Genetics, NIDCD, NIH* *Faculty page: https://www.nidcd.nih.gov/about/staff/thomas-b-friedman-phd*	**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.* *Learning objectives*: • *Obtaining knowledge of human inherited deafness* • *Differentiating between pathogenic and benign mutations associated with human deafness* • *Recognizing noncoding variants associated with human deafness*
11:00-11:15	**Short Break**	
11:15-11:45 	**Hela Azaiez, PhD** *Molecular Otolaryngology and Renal Research Labs (MORL), Department of Otolaryngology, University of Iowa, USA* *Faculty page: https://morl.lab.uiowa.edu/people/hela-azaiez-phd*	**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.* *Learning objectives:* • *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 	**Carmen C. Brewer, AuD PhD** *Chief, Audiology Unit, Division of Intramural Research, NIDCD* *Faculty page: https://www.nidcd.nih.gov/about/staff/carmen-c-brewer-phd*	**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.* *Learning objectives:* • *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*

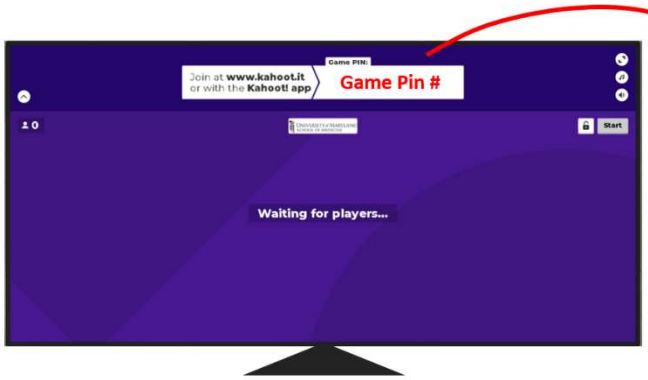
12:15-1:30	**Lunch Break**	
1:30-2:10 	**Rick A. Friedman, MD, PhD** *Vice Chief, Division of Otolaryngology, UCSD* *Professor of Surgery* *Director of UCSD Acoustic Neuroma Center* *UCSD Health/School of Medicine* *Faculty page:* *https://providers.ucsd.edu/details/32904/ent-head-and-neck-(otolaryngology)-surgery*	**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.* *Learning objectives:* • *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**	
2:25-3:15 	**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* *Faculty page:* *https://www.entcolumbia.org/profile/lrlustig*	**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.* *Learning objective:* • *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*
3:15-3:30	**Short Break**	
3:30-4:00	**Interactive Quiz with Prizes!** ***Hosted by:Ben Shuster and Erika Lipford***	*Join in for an interactive and fun quiz/trivia on hearing loss and its treatments via KAHOOT!*
4:00-4:30	**Panel Discussion** ***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*** **(moderator)**	**We will Brainstorm Hot Topics in Genetic Hearing Loss**       
4:30-4:45 	**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/*	**Closing Remarks**
4:45-5:00	**Short Break**	
5:00-5:45	**Genetic Tools Workshop** **Ronna Hertzano, MD, PhD** **Hela Azaiez, PhD**	*A walk through of available genetic, genomic and transcriptomic resources for scientists, clinicians and genetic counselors who care for persons with hearing loss.*

Join us for an interactive quiz from 3:30-4:00pm!

1. **How are we participating in this fun interactive quiz/trivia?**
   We are using a program called Kahoot!

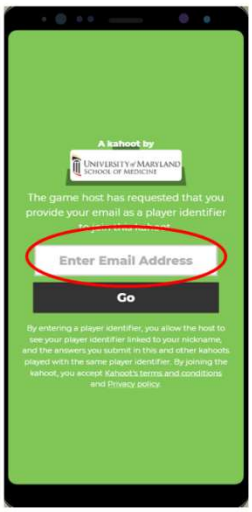
2. **How do I use Kahoot?**
   Kahoot is very simple to use. Please follow the below steps (we'll also walk through this when starting the interactive quiz).

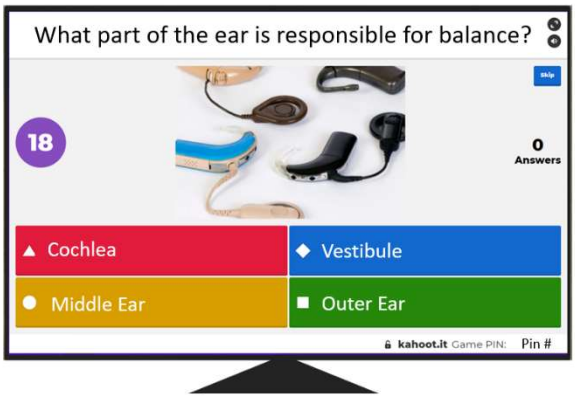
STEP 1: Using your phone or tablet, open a new internet browser and type in 'kahoot.it' in the search bar



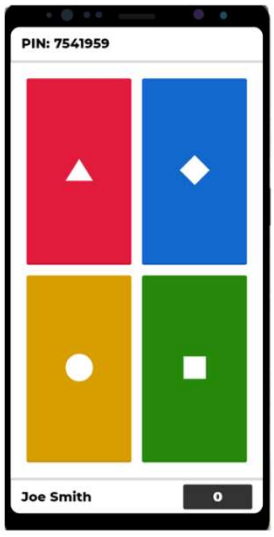
STEP 2: Type the game pin displayed on your computer into the phone and click 'enter'

STEP 3: Type in your email address and name, which will be used to contact you if you win a prize!





STEP 4: Once the game has started, use your phone to click the color and shape corresponding to the answer you believe is correct!

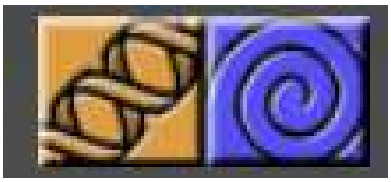


3. You say there are prizes?!?! 😊

Yes! The top 5 winners will receive Amazon gift cards. The winners are determined as the individuals that have answered the most questions correctly and quickly.

Join us for a Genetics Tools Workshop to learn about the following very helpful and useful genetic tools!

(5:00-5:45pm)



The Hereditary Hearing Loss Homepage (https://hereditaryhearingloss.org/) provides an up-to-date overview of the genetics of hereditary hearing impairment. It lists data and links for all mapped loci and identified genes for non-syndromic hearing loss and several prevalent syndromes.

Video contributed by: Kevin Booth, PhD

DEAFNESS VARIATION DATABASE

The Deafness Variation Database (DVD: http://deafnessvariationdatabase.org/) is a comprehensive, open-access resource that integrates all available genetic, genomic and clinical data together with expert curation to generate a single classification for each variant in all known genes implicated in non-syndromic deafness and prevalent syndromic forms.

Video contributed by: Robert Marini

AudioGene

Audiogene (https://audiogene.eng.uiowa.edu/) is a supervised support vector machine learning algorithm that uses audiometric of patients with hearing loss to predict the genetic etiology.

Video contributed by: William Walls

IMPC
International Mouse Phenotyping Consortium

The International Mouse Phenotyping Consortium (IMPC: https://www.mousephenotype.org/) is a global effort to identify the function of every protein-coding gene in the mouse genome. Our growing catalogue of mammalian gene function is freely available for researchers.

Video contributed by: Michael Bowl, PhD

Allen Brain Atlas (https://portal.brain-map.org/) includes in situ hybridization images of sections of the entire mouse embryo or mouse brain at different developmental stages. Here we show how to utilize this resource to look at gene expression in the developing inner ear.

Video contributed by: Thomas Coate, PhD

gEAR (https://umgear.org/) provides a single resource for data deposition, display, analysis, and interrogation, with a heavy focus on allowing users to customize the site and their own data displays. Currently, we have over 90 ear related datasets organized in thematic profiles.

Video contributed by: Ronna Hertzano, MD, PhD

Enrichr (http://amp.pharm.mssm.edu/Enrichr) is an easy to use intuitive enrichment analysis web-based tool providing various types of visualization summaries of collective functions of gene lists. Enrichr is open source and freely available online.

Video contributed by: Beatrice Milon, PhD

Pharos (https://pharos.nih.gov/) is the user interface to the Knowledge Management Center (KMC) for the Illuminating the Druggable Genome (IDG) program funded by the National Institutes of Health (NIH) Common Fund. The goal of KMC is to develop a comprehensive, integrated knowledge-base for the Druggabale Genome (DG) to illuminate the uncharacterized and/or poorly annotated portion of the DG, focusing on G-protein-coupled receptors (GPCRs), ion channels (ICs), and kinases.

Video contributed by: Michael Hoa, MD

SCENIC: single-cell regulatory network inference and clustering (http://scenic.aertslab.org) is a computational method for simultaneous gene regulatory network reconstruction and cell-state identification from single-cell RNA-seq data.

Video contributed by: Mannat Karla

Cell Trails (https://hellerlab.stanford.edu/celltrails/) is an unsupervised algorithm for the *de novo* chronological ordering, visualization and analysis of single-cell expression data. CellTrails enables the reconstruction of branching trajectories and provides an intuitive graphical representation of expression patterns along all branches simultaneously. It allows the user to define and infer the expression dynamics of individual and multiple pathways towards distinct phenotypes.

Video contributed by: Stefan Heller, PhD