



CureARS



CHOC

The Mitochondrial ARS (mtARS) Genes Annual Scientific Symposium

February 7, 2025

Time (EST)	Duration	Presenter	Topic
10:00 - 10:05 AM	5 Mins	Jose Abdenur, MD CHOC, part of Rady Children's Health	<i>Welcome</i>
10:05 - 10:10 AM	5 Mins	Ashley Rowland & Desiree Magee Co-Founders of CureARS	<i>CureARS Introduction</i>
Drug Repurposing			
10:10 - 10:40 AM	30 Mins	Cristina Remes, PhD Children's Hospital of Philadelphia	<i>Mechanistic and Therapeutic Modeling in ARS2 Class of Primary Mitochondrial Disease in C. elegans and zebrafish animal models</i>
10:40 - 11:10 AM	30 Mins	Ethan Perlstein, PhD Founder/CEO of Perlara & Curetopia	<i>Yeast-powered drug repurposing for inherited metabolic diseases</i>
mtARS Prevalence			
11:10 - 11:25AM	15 Mins	Renkui Bai, MD, PhD, FACMG GeneDx	<i>How common Mitochondrial ARS (Mt-aaRS) Genes-related disease are in patients tested by clinical exome sequencing at GeneDx</i>
Data & Project Set-up for Clinical Studies			
11:25 - 11:45 AM	20 Mins	Lois Sayrs, PhD, Director, Biostatistics CHOC, part of Rady Children's Health	<i>Informatics and Statistics Research Activation in Clinical Studies in Rare</i>
11:45 AM - 12:05 PM	20 Mins	Catarina Olimpico, MD University of Cambridge, Horvath Laboratory	<i>Cognate amino acid supplementation in mitochondrial aminoacyl-tRNA synthetase diseases - developing an experimental medicine study</i>
LUNCH 12:05PM-12:50PM (45 Minutes)			
12:50 - 1:00 PM	10 Mins	Sundeep Dugar, PhD, Co-CEO Blue Oak Nutraceuticals	<i>Mitokatlyst - E</i>
1:00 - 1:20 PM	20 Mins	Rebecca Ganetzky, MD Children's Hospital of Philadelphia Ya-Ming Hou, PhD, Professor Thomas Jefferson University	<i>A kinetic model for compound heterozygous pathogenic variants in YARS2-associated neonatal phenotype</i>
Natural History			
1:20 - 1:30 PM	10 Mins	Neda Zadeh, MD CHOC, part of Rady Children's Health	<i>Thoughts on the Future of Genetic Testing</i>
1:30 - 1:40 PM	10 Mins	Amy Goldstein, MD Clinical Director Children's Hospital of Philadelphia	<i>MTARS patient clinical cohort at CHOP</i>
1:40 - 1:50 PM	10 Mins	Amena Smith-Fine, MD Center for Leukodystrophies, Kennedy Krieger Institute	<i>LBSL Natural History</i>
RNA Biology			
1:50 - 2:10 PM	20 Mins	Jay Hesselberth, PhD University of Colorado School of Medicine	<i>Integrated analysis of tRNA sequence, modification, and aminoacylation by nanopore sequencing</i>
Gene Therapy & ASO			
2:10 - 2:30 PM	20 Mins	Christina Nemeth Mertz, PhD Kennedy Krieger Institute	<i>AAV9 and ASO therapy in LBSL - UPDATE</i>



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2:30 - 3:00 PM	30 Mins	Dan Wang, PhD, Assistant Professor Department of Genetic and Cellular Medicine and RNA Therapeutics Institute, UMass Chan Medical School	<i>Developing a potential AAV gene therapy for AARS2-related disease</i>
BREAK 3:00-3:10PM			
Clinical Trials / Treatment			
3:10 - 3:20 PM	10 Mins	Laura Dennis MacMullen, CCRC Clinical Research Program Manager Children's Hospital of Philadelphia	<i>Elamipretide treatment in mtARS patients</i>
Research Readiness / Strategic Planning			
3:20 - 3:30 PM	10 Mins	Karmen Trzupsek, Senior Director of Scientific Programs Global Genes	<i>Global Genes Research Readiness & Strategic Planning</i>
Panel: Amino Acid Supplementation in mtARS Patients			
3:30 - 4:20 PM	50 Mins	Marni Falk, MD Children's Hospital of Philadelphia Rita Horvath, PhD, MD, Professor of Neurogenetics University of Cambridge Johan Van Hove, MD Children's Hospital of Colorado Jennifer Yang, MD Rady Children's Hospital San Diego Karmen Trzupsek, Senior Director of Scientific Programs Global Genes	<i>Panel Title: Amino Acid Supplementation in mtARS Patients Moderator: Marni Falk</i>
Biomarker Research / Diagnostics			
4:20 - 4:40 PM	20 Mins	Johan Van Hove, PhD, MD Children's Hospital of Colorado	<i>A systematic study of the functional diagnosis of ARS2 disorders</i>
4:40 - 4:50 PM	10 Mins	David Stroud, PhD University of Melbourne in Australia	<i>Proteomics enables ultra-rapid diagnosis of mitochondrial disease</i>
Closing			
4:50 - 5:00 PM	10 Mins	Jennifer Yang, MD Assistant Clinical Professor of Neurosciences in the Division of Pediatric Neurology at UC San Diego and Rady Children's Hospital San Diego	Patient engagement in mitochondrial disease research