

The Mitochondrial ARS (Mt-aaRS) Genes Annual Scientific Symposium 2023

February 10, 2023

Time Scheduled	Presenter Information
10:00 am - 10:08 am	Welcome & Introduction <i>Brent Dethlefs, Executive Director, CHOC Research Institute</i>
10:08 am - 10:15 am	CureARS Introduction Desiree Magee & Ashley Rowland <i>Founders of CureARS</i>
10:15 am – 11:00 am	Panel #1: Natural History Studies in Rare Diseases Ali Fatemi, MD <i>Kennedy Krieger Institute</i> Amy Goldstein, MD <i>Children's Hospital of Philadelphia</i> Catarina Olimpio, MD <i>University of Cambridge, Horvath Laboratory</i>
11:00 am - 11:15 am	A Diagnostic Odyssey: Pathway from symptoms to diagnosis Neda Zadeh, MD <i>Children's Hospital of Orange County</i>
11:15 am – 11:35 am	Mechanistic and Therapeutic Modeling in ARS2 Class of Primary Mitochondrial Disease in <i>C. elegans</i> and zebrafish animal models Christina Remes, PhD <i>Children's Hospital of Philadelphia</i> Neal Mathew, PhD <i>Children's Hospital of Philadelphia</i>
11:35 am - 11:55 am	The clinical spectrum of ARS disorders: A case study Jennifer H. Yang, MD <i>University of California San Diego (UCSD) and Rady Children's Hospital San Diego</i>
11:55 am - 12:05 pm	How common Mitochondrial ARS (Mt-aaRS) Genes-related disease are in patients tested by clinical exome sequencing at GeneDx Renkui Bai, MD, PhD, FACMG <i>GeneDx</i>
12:05 pm - 12:15 pm	MitoSHARE Registry Phil Yeske <i>United Mitochondrial Disease Foundation (UMDF)</i>
12:15 pm - 1:00 pm	Lunch Break (patient video/slides)
1:00 pm - 1:30 pm	Multi-gene Research Modeling mitochondrial aminoacyl-tRNA synthetase defects in vitro in neurons Oliver Podmanicky, PhD Student <i>University of Cambridge, Horvath Laboratory</i>

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1:30 pm – 1:45 pm	<p>AARS2 & SARS2 Research Henna Tynismaa, PhD <i>University of Helsinki, Finland</i></p>
1:45 pm - 2:15 pm	<p>YARS2 [Tyrosyl-tRNA synthetase] Project & Modeling Rebecca Ganetzky, MD <i>Children's Hospital of Philadelphia</i> Ya-Ming Hou, PhD <i>Thomas Jefferson University</i></p>
2:15 pm - 2:45 pm	<p>Panel #2: Mito Cocktail as a Therapeutic</p> <p>Rebecca Ganetzky, MD <i>Children's Hospital of Philadelphia</i> Melissa A. Walker M.D., PhD <i>Massachusetts General Hospital</i> Brian Kirmse, MD <i>University of Mississippi Medical Center</i> Divakar S. Mithal MD, PhD <i>Lurie Children's Hospital and Northwestern University in Chicago</i></p>
2:45 pm - 3:00 pm	Break
3:00 pm – 3:30 pm	<p>Enhanced Energy Metabolism in Aminolevulinatase/Iron-Exposed DARS2 deficient Fibroblasts Jose Abdenur, MD <i>Children's Hospital of Orange County</i></p>
3:30 pm – 4:00 pm	<p>AAV9 and ASO therapy in LBSL Christina Nemeth Mertz, PhD <i>Kennedy Krieger Institute</i></p>
4:00 pm – 4:30 pm	<p>Research Opportunities and Advances for the ARS Community at the NIH Eliza Gordon-Lipkin, MD <i>National Institute of Health</i></p>
4:30 pm – 5:00 pm	<p>Diagnosing ARS2 defects in fibroblasts: Functional studies and the role of protein-based diagnostics Johan Van Hove, MD <i>Children's Hospital of Colorado</i> David Stroud, PhD <i>University of Melbourne in Australia</i></p>
5:00 pm	Closing Remarks