The Mitochondrial ARS (Mt-aaRS) Genes Annual Scientific Symposium 2022

February 10th, 2022

Agenda	
***Times are in Eastern Standard Time (EST). This agenda is subject to change and is tentative.**	
Time Scheduled	**Presenter Information**
9:00 am	***Welcome & Introduction*** **Ali Fatemi, MD** Chief Medical Officer, Center for Leukodystrophies, Kennedy Krieger Institute
9:10 am	***CureARS Introduction*** **Desiree Magee & Ashley Rowland** Founders/Owners
9:15 am	***Clinical Presentations and molecular mechanisms of mtARS mutations*** **Rita Horvath, MD, PhD** University of Cambridge, Horvath Laboratory
9:45 am	***Challenges of studying ARS2 defect in skin fibroblasts*** **Johan Van Hove, MD** Children's Hospital Colorado
10:15 am	***The Power of Yeast for Functional Analysis of Novel Variants in Mitochondrial Aminoacyl-tRNA Synthetase*** **Cristina Dallabona, Postdoctorate Researcher** University of Parma, Italy
10:30 am	**Live Q&A** All panelists
10:45 am	**Break**
11:00 am	***Investigation of cell type specific effects of AARS2 and SARS2 patient mutations using stem cell derived models*** **Henna Tyynismaa, PhD** University of Helsinki, Finland
11:30 am	***Investigating the role of seryl-tRNA synthetase [SARS2] in mitochondrial biology and recessive disease*** **Christina Del Greco, PhD Student** University of Michigan, Anthony Antonellis Laboratory
12:00 pm	***YARS2 [Tyrosyl-tRNA synthetase] Project & Modeling*** **Rebecca Ganetzky, MD** Children's Hospital of Philadelphia **Ya-Ming Hou, PhD** Thomas Jefferson University
12:10 pm	**Live Q&A** All panelists
12:20 pm	**Lunch Break**
1:15 pm	***Characterization of MARS2 Deficiency*** **Bryn Webb, MD** University of Wisconsin-Madison
1:45 pm	***Fibroblast Studies in DARS2*** **Jose Abdenur, MD**

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	Children's Hospital of Orange County
2:15 pm	***Leukoencephalopathy with Brainstem and Spinal Cord Involvement and Lactate Elevation (LBSL) - A prototype disorder of mitochondrial tRNA Synthetases*** **Amena Smith-Fine, MD & Christina Mertz, PhD** Center for Leukodystrophies, Kennedy Krieger Institute **Marc Engelen, MD, PhD** Department of (Pediatric) Neurology, Amsterdam Leukodystrophy Center, Amsterdam UMC
3:00 pm	**Live Q&A** All panelists
3:15 pm	**Break**
3:25 pm	***Mt-aaRS Patient Registry*** **Alyssa Mendel, Research Project Manager** CoRDS (Coordination of Rare Disease Registry) at Sanford Health
3:30 pm	***Mt-aaRS research proposal overview and demonstration of the types of high-throughput and animal modeling experiments*** **Neal Mathew, PhD** Children's Hospital of Philadelphia
3:40 pm	***Rare Disease Research & Advocacy with Allstripes*** **Richard Elles, PMP** Director of Patient Advocacy & Industry Engagement at Allstripes
3:45 pm	***Panel Discussion: Advances in Therapeutics*** ***Introductory Comments and Quick Overview of Therapies in Development for Mitochondrial Disease*** **Philip Yeske, PhD** United Mitochondrial Disease Foundation (UMDF) ***Precision Mitochondrial Medicine: Using preclinical models to identify therapeutic leads to evaluate in mitochondrial disease patients in the Mitochondrial Medicine Frontier Program*** **Marni Falk, MD** Children's Hospital of Philadelphia ***Vatiquinone for Treatment of -ARS2 Disorders*** **Matthew Klein, MD, MS, FACS** PTC Therapeutics ***Function™: A scalable platform to enable patient-centric drug discovery for rare diseases*** **Christopher Moxham, PhD** Rarebase ***Commercial Laboratory Outreach Programs for Rare Diseases.*** **Paul Kruszka, MD, FACG** GeneDx
5:00 pm	**Closing Remarks**