



ATP1A3 IN DISEASE SYMPOSIUM

Session I. Disease Natural History

Chair: Alfred George Jr., Northwestern University

Eleni Panagiotakaki

University Hospital of Lyon

"Overview of the IAHCRC Consortium and IAHCRC-CLOUD Platform"

Mohamad Mikati

Duke University School of Medicine

"OBSERV-AHC study Design and Methods of Analysis"

Lyndsey Prange

Duke University School of Medicine

"OBSERV-AHC Methods of Spell Documentation: Video Library and Paper Diary"

Shital Patel

Duke University School of Medicine

"OBSERV-AHC Preliminary Reports on Efficacy study on CBD"

Andrew Landstrom

Duke University School of Medicine

"Not just the brain: The role of ATP1A3 in cardiac arrhythmogenesis"

Carmen Fons

Sant Joan de Déu Children's Hospital OBSERV-AHC

"Efficacy study on the Ketogenic Diet"

Session III. Quantifying Phenotypes and Clinical Trial Readiness

Chair: Eleni Panagiotakaki, University of Lyon

Terry Jo Bichell

Vanderbilt University, Combined Brain

"Lessons learned from Angelman syndrome and other rare genetic neurodevelopmental disorders"

Allison Brashear

University of Buffalo

"Quantifying phenotypes in ATP1A3-related disorders"

Maria Papadopoulou

University Hospital of Lyon

"OBSERV-AHC Electronic Diary App"

FRIDAY - SATURDAY, October 27-28, 2023

Northwestern University, Feinberg School of Medicine

303 E. SUPERIOR ST. CHICAGO, IL, USA 60611

Session II. Basic Mechanisms / Neurophysiology

Chair: Kathleen Sweadner, Harvard Medical School

Sho Yano

University of Chicago

"Cation leak through mutant ATP1A3 pumps"

Hanne Poulsen

Aarhus University

"Structural insights into ATP1A3 dysfunction"

Arn van den Maagdenberg

University of Leiden

"Investigating spreading depolarization in mouse models"

Hendrik Rosewich

University of Tübingen

"Modeling alternating hemiplegia of childhood in bioengineered neuronal organoids"

Anne Hart

Brown University

"Modeling alternating hemiplegia of childhood in *C. elegans*"

Nina Frost

Hope for Anabel Foundation

"Aligning Priorities and Research Strategic Plan"

Session IV. Therapeutic Advances

Chair: Arn van den Maagdenberg

Anna Mingorance

LouLou Foundation

"Paving the therapeutic road ahead"

Alex Sousa

Broad Institute and MIT

"Prime editing for alternating hemiplegia of childhood"

Markus Terrey

Jackson Laboratories

"Update on AAV gene therapy for alternating hemiplegia of childhood"

Alfred George Jr.

Northwestern University

"Antisense oligonucleotide therapeutics for alternating hemiplegia of childhood"