



12th SYMPOSIUM ON ATP1A3 IN DISEASE

14 – 15 November 2024
Sant Pau Recinto Modernista
Barcelona, Spain

PROGRAMME OVERVIEW

BASIC SCIENCE

Progress understanding ATP1A3 structure and function

Pablo Artigas

Progress in understanding the functional impact of disease mutations on Na, K-ATPase function

Mads Eskesen Christensen

Novel insights into Na,K-ATPase structures

Kathleen Sweadner

Cell biological insights in ATP1A3 disorders

Arn van Maagdenberg

Spreading depolarization in mouse models in AHC

Erin Heinzen

Genetic landscape of AHC

Anne Hart

Modeling ATP1A3 dysfunction in *C.elegans*

Mads Andersen

The NA,K-ATPase in spreading depolarization: What can we learn from insect models?

TRANSLATIONAL SCIENCE

Gene based treatments

Alex Sousa

Progress in gene editing for AHC

Markus Terrey

Gene delivery and editing in mouse models of AHC

TBD

Aplice-switching oligonucleotide (ASO) therapy development

Pharmacological and functional treatments

Jackeline Alves

Molecules activators of ATP1A3

Danilo Tizziano

Drug repurposing

Quentin Welniarz

Oxygen trial in AHC

Clinical trials models for rare diseases

Floor Jansen

N-of-one strategy in rare disorders

CLINICAL SCIENCE

Natural history and phenotype in ATP1A3

Mohamad Mikati

Results of long-term outcomes (natural history study in AHC)

Elisa de Grandis

Development of specific severity AHC scale

Eleni Panagiotakaki

Overview on ATP1A3-non AHC phenotypes (RECA, DYT12, CAPOS...)

Esther Lasheras, Jaume Campistol

Transition from paediatric to adulthood care - A10 programme HSJD

TBC

Behaviour problems in patients with ID

Biomarkers in ATP1A3 disorders

Simona Balestrini

Electrophysiology biomarkers in AHC

Juan Dario Ortigozo

Movement disorders and treatment management in ATP1A3-related disorders

T. Ikezu

ATP1A3 as a target for isolating neuron-specific extracellular vesicles from human brain and biofluids

Baptiste Balança

Recording, analysis, and interpretation of spreading depolarizations in neurointensive care

Joan Gibert

Transforming healthcare. The role of artificial intelligence in clinical practice

NETWORKS AND ADVOCACY

Alexis Arzimanoglou

European Reference Networks: an essential asset at the service of rare and complex diseases

Siggi Johannesson

AHC Europe

Nina Frost

What matters to families

Jackeline Alves

UNICAS Network - a paradigmatic project to improve care of paediatric patients with rare disorders
