

# 12<sup>th</sup> SYMPOSIUM ON ATP1A3 IN DISEASE

14 – 15 November  
2024

Espai Modernista Sant Pau  
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
--------------	-------------------------------------

# 12<sup>th</sup> SYMPOSIUM ON ATP1A3 IN DISEASE

14 – 15 November  
2024

Espai Modernista Sant Pau  
Barcelona  
Spain

## Programme Overview

### 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
Manuel del Castillo	UNICAS Network - a paradigmatic project to improve care of paediatric patients with rare disorders