



12th SYMPOSIUM ON ATP1A3 IN DISEASE

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

PROGRAMME OVERVIEW

DAY 1

Thursday, 14 November 2024

8:00 - 8:20 Welcome and opening remarks

DECODING ATP1A3

Insights into Rare Disease Mechanisms

- 8:20 - 8:45 **Arn van Maagdenberg**
Leiden University Medical Centre
Cortical spreading depolarization in mouse models in AHC
- 8:45 - 9:10 **Mads Eskesen Christensen**
Aarhus University
Novel insights into Na,K-ATPase structures
- 9:10 - 9:35 **Mads Andersen**
Aarhus University
The Na,K-ATPase in spreading depolarization: What can we learn from insect models?

9:35 - 9:40 *Lived Experience: family testimonials*

9:45 - 10:15 Coffee break

BRIDGING DISCOVERIES

From research to therapeutic applications

- 10:20 - 10:45 **Markus Terrey**
The Jackson Laboratory
Gene delivery and editing in mouse models of AHC
- 10:45 - 11:10 **Alex Sousa**
Broad Institute of MIT and Harvard
Progress in gene editing for AHC
- 11:10 - 11:35 **Al George**
Northwestern University
Antisense-oligonucleotide (ASO) therapy development for AHC

CLINICAL APPROACHES

Shaping future treatments

- 11:35 - 12:00 **Esther Lasheras, Jaume Campistol**
Hospital Sant Joan de Deu
Transition from paediatric to adulthood care
- 12:00 - 12:25 **Elisa de Grandis**
University of Genoa
Development of AHC-specific severity scale

12:25 - 12:40 *Poster Blitz: rapid-fire poster presentations*

DAY 1

12:45 - 12:55 Group photo

13:00 - 14:30 Lunch - Poster viewing - Networking

GLOBAL ADVOCACY

Empowering collaboration and support

14:35 - 15:00 **Nina Frost**
Hope for Annabel Foundation
Centering Patient Priorities in Research for AHC

15:00 - 15:25 **Siggi Johannesson**
AHC Iceland, AHC Europe
AHC organisations in Europe

15:25 - 15:35 *Short talk selected from abstracts*

15:40 - 15:55 Coffee break

CLINICAL APPROACHES

Shaping future treatments

16:00 - 16:25 **Eleni Panagiotakaki**
University Hospital of Lyon
Overview on ATP1A3 non-AHC phenotypes (RECA, DYT12, CAPOS...)

16:25 - 16:50 **Mohamad Mikati**
Duke University School of Medicine
Long-term outcomes from the AHC natural history study

16:50 - 17:15 **Laia Villalta**
Hospital Sant Joan de Deu
Behavioural problems in neurodevelopmental disorders

17:15 - 17:40 **Andrew Landstrom**
Duke University School of Medicine
Cardiac manifestations of ATP1A3-associated diseases

17:40 - 17:45 *Lived Experience: family testimonials*

18:00 - 18:05 Closing Day 1

DAY 2

Friday, 15 November 2024

8:00 - 8:10 Opening Day 2

DECODING ATP1A3

Insights into Rare Disease Mechanisms

- 8:10 - 8:35 **Kathleen Sweadner**
Massachusetts General Hospital
Cell biological insights in ATP1A3 disorders
- 8:35 - 9:00 **Pablo Artigas**
Texas Tech University Health Sciences Center
Progress in understanding the functional impact of disease mutations on Na, K-ATPase function
- 9:00 - 9:25 **Erin Heinzen**
University of North Carolina School of Medicine
Genetic landscape of AHC
- 9:25 - 9:50 **Anne Hart**
Brown University
Modeling ATP1A3 dysfunction in *C.elegans*
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9:50 - 10:00 *Short talk selected from abstracts*

10:05 - 10:35 Coffee break

BRIDGING DISCOVERIES

From research to therapeutic applications

- 10:40 - 11:05 **Jacqueline Alves**
Federal University of Goiás
Molecular activators of ATP1A3
- 11:05 - 11:30 **Victoria de Felipe**
UMC Utrecht
N-of-one strategy in rare disorders
- 11:30 - 11:55 **Danilo Tizziano**
Catholic University of the Sacred Heart
Drug repurposing for AHC
- 11:55 - 12:20 **Quentin Welniarz**
Pitié Salpêtrière Hospital
Oxygen trial in AHC
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12:20 - 12:25 *Lived Experience: family testimonials*

12:30 - 13:30 Poster Session

DAY 2

13:30 - 14:30 Lunch

14:35 - 15:00 **Simon Frost**
Cure AHC
Keynote address

CLINICAL APPROACHES

Shaping future treatments

15:00 - 15:25 **Simona Balestrini**
UCL Queen Square Institute of Neurology
Electrophysiology biomarkers in AHC

15:25 - 15:50 **Dario Ortigoza**
Hospital Sant Joan de Deu
Movement disorders and treatment management in ATP1A3-related disorders

15:50 - 16:00 *Short talk selected from abstracts*

16:05 - 16:20 Coffee break

GLOBAL ADVOCACY

Empowering collaboration and support

16:25 - 16:50 **Alexis Arzimanoglou**
Hospital Sant Joan de Deu
European Reference Networks: an essential asset at the service of rare and complex diseases

16:50 - 17:15 **Manuel del Castillo**
Hospital Sant Joan de Deu
UNICAS Network - a paradigmatic project to improve care of paediatric patients with rare disorders

CLINICAL APPROACHES

Shaping future treatments

17:15 - 17:40 **Tsuneya Ikezu**
Mayo Clinic
ATP1A3 as a target for isolating neuron-specific extracellular vesicles from human brain and biofluids

17:40 - 18:05 **Baptiste Balanca**
University of Lyon
Non-invasive recording, analysis, and interpretation of spreading depolarizations in neurointensive care

18:05 - 18:30 **Joan Gibert**
Hospital Sant Joan de Deu
Transforming healthcare. The role of artificial intelligence in clinical practice

18:30 - 18:35 *Lived Experience: family testimonials*

18:35 - 18:45 Closing words from the Standing Committee and Announcement of the 13th Symposium on ATP1A3 in Disease

20:30 Closing Dinner

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