



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

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

# PROGRAMME OVERVIEW

# DAY 1

Thursday, 14 November 2024

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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?

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

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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

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

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# DAY 1

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12:45 - 12:55    Group photo

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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

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

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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

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

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18:00 - 18:05    Closing Day 1

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# DAY 2

Friday, 15 November 2024

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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*

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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*

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12:30 - 13:30      Poster Session

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# DAY 2

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13:30 - 14:30 Lunch

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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

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

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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

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

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

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20:30 Closing Dinner

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Organised by:



**SJD** Sant Joan de Déu  
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