"News and Impressions from the 12th International Meeting in Lausanne"

Organizer´s summary: Conference highlights (abstracts to appear in tremorjournal.org)
Fabrizio Vacca, Jules Gonin Eye Hospital, University of Lausanne, Lausanne, Switzerland

Summary from the Neuroacanthocytosis Advocacies
Link to lay-language report:
https://naadvocacy.org/wp-content/uploads/2025/11/REPORT-12th-Symposium-Lausanne-12-14-Sep-2025.pdf

Glenn Irvine Prize lecture essentials
Marianna Leonzino, Humanitas Research Hospital, Milan, Italy

Summary by the Cohen Syndrome Research Foundation
Ashley Waterman, Los Angeles, CA, USA

"If I get to Korea, I'll tell you"
José Miguel Figueiredo & Mauro Corage, Teatro do Zero, Vila Franca de Xira, Portugal

23rd VPS13 Forum November 2025

Thank you

To the organisers and hosts:

Dr. Kevin Peikert
Professor Ruth Walker
Professor Dr. Adrian Danek

To all the speakers!

To all attendees and everyone reading this report and helping to share the knowledge.

TABLE OF CONTENTS

INTRODUCTION

The Forum was moderated by Professor Dr. Adrian Danek, Ludwig Maximilian University, Munich, Germany.

The focus was on summarising the news and impressions from the 12th International Meeting on Neuroacanthocytosis Syndromes, Cohen Syndrome and other VPS13-relarted disorders which took place in Lausanne, Switzerland, between 12 and 14 September 2025.

The presentations were:

- **Organiser's Summary: Conference Highlights**
  Fabrizio Vacca, Jules-Gonin Eye Hospital, University of Lausanne, Switzerland
- **Summary from the Neuroacanthocytosis Advocacies**
  Ginger Irvine, Joy Willard-Williford, Despina Dinca – UK & USA based
- **Glenn Irvine Prize Lecture Essentials**
  Marianna Leonzino, Humanitas Research Hospital, Milan, Italy
- **Summary by the Cohen Syndrome Research Foundation**
  Ashley Waterman, Los Angeles, USA
- **"If I Get to Korea, I'll Tell You"**
  José Migue Figueiredi & Mauro Corage, Teatro do Zero, Portugal.

CONFERENCE HIGHLIGHTS

Fabrizio opened the Forum with a summary of the recent Lausanne meeting, which brought together around 60 participants, including families, clinicians, and scientists. He reflected on the warm, open atmosphere that allowed for relaxed discussion and genuine connection.

Key scientific themes

Fabrizio highlighted several major areas of progress:

- **Bridge-like lipid transport proteins:**
  Structural and mechanistic insights into VPS13 and XK have advanced rapidly in

the past 4-5 years, revealing how these proteins move lipids between organelles and how their dysfunction contributes to disease.

- **Model organisms:**
  Mouse models continue to be powerful tools for understanding both Cohen syndrome and NA. Newer systems, such as zebrafish and brain organoids, offer scalable platforms for translational work, even if their relationship to human pathology is still being refined.
- **Translational readiness:**
  Biomarker identification and early translational studies are progressing, helping prepare the field for future clinical trials once therapeutic candidates emerge.
- **Clinical perspectives:**
  Sessions on clinical care underscored the importance of nuanced, individualised approaches for both Cohen syndrome and VPS13A/XK-related disorders.

Fabrizio noted that the meeting abstracts have been submitted for publication, with reviewers currently completing their work.

A brief discussion followed on the role of computational modelling. Participants emphasised that while simulations must be interpreted cautiously, they generate valuable hypotheses and complement structural and cellular studies.

UPDATES FROM THE NA ADVOCACIES

The updates from Ginger, Joy and Despina focused on the growing alignment between scientific progress and community needs.

Patient & carer sessions in Lausanne

The advocacies hosted a dedicated programme for patients and carers, featuring:

- **Occupational therapy (Joanna Valente):**
  Practical strategies to preserve independence, dignity, and daily function through adaptive tools and personalised approaches.
- **Speech and language therapy (Elina Tripoliti):**
  Techniques to support communication, swallowing, and connection, reminding everyone that even when speech changes, the person remains.
- **Mental health (Matt Bolts Johnson):**
  A powerful reminder that *mental health is not a luxury, it is a foundation*, especially in rare disease communities where life becomes complex for both patients and carers.

Community reflections

Participants valued:

- Mental health support
- Eating, drinking, and communication strategies
- The chance to share lived experiences across VPS13A and XK disease
- More opportunities for direct interaction with researchers and clinicians

Suggestions received:

- A physiotherapy session at the next symposium
- More disease-specific information for XK families
- A carer-focused meeting or online session

Scientific takeaways from the patient perspective

- Growing recognition that NA syndromes form a shared family of disorders
- No one-size-fits-all clinical approach, but earlier diagnosis and better care are emerging
- Simple organisms teach complex lessons about lipid transport
- Poster sessions reflected collaboration and curiosity
- Model organisms and patient-derived neurons are shaping future research
- Better awareness and patient-centred care can improve diagnosis and support.

A gentle call to action

Adrian emphasised the importance of biological samples, including blood, skin biopsies, and, when families feel able to consider it, postmortem tissue, for advancing research.

While it's recognised to be a generally difficult topic and acknowledging that the decisions around tissue donations are deeply personal, it is also noted that such considerate contributions can meaningfully accelerate scientific understanding.

GLENN IRVINE PRIZE LECTURE ESSENTIALS

Marianna, the 2025 Glenn Irvine Prize winner, presented an overview of her postdoctoral work on the VPS13 protein family.

Research highlights:

- **VPS13 proteins at membrane contact sites:**
  Her work helped establish that VPS13A, C, and D localise to specific membrane contact sites and contain a continuous hydrophobic channel enabling bulk lipid transfer.

- **Focus on VPS13D:**
  VPS13D is essential in humans and animal models, with mutations causing severe movement disorders. Marianna's research explores:
  - how VPS13D depletion affects mitochondria, Golgi structure, and lipid composition
  - how patient-derived mutations alter neuronal development, mitochondrial morphology, and synaptic activity
  - why different mutations lead to different levels of protein stability and disease severity.

Her presentation demonstrated how fundamental cell biology can illuminate disease mechanisms and guide future therapeutic strategies.

UPDATES FROM COHEN SYNDROME RESEARCH FOUNDATION

The scheduled presentation from the Cohen Syndrome Research Foundation could not be delivered due to the speaker's technical difficulties and will be rescheduled in a next

meeting. The Forum acknowledged the importance of continued collaboration across VPS13-related conditions and looks forward to future updates.

THEATRE PLAY

“If I get to Korea, I’ll tell you”

Presented by & Mauro Corage, Teatro do Zero

The Forum concluded with a moving theatrical performance exploring identity, illness, and resilience. The play offered a powerful artistic lens on living with a rare condition, honest, courageous, and deeply human.

Written and performed by José Miguel Figueiredo, a patient living with VPS13A disease, the performance was made possible through Teatro do Zero, Portugal and its director, Mauro Corage.

The theatre play was recorded at the 12th Symposium, and it was warmly received by the scientific and patient communities alike on the day and in the Forum. The creators kindly asked that the recording remain unlisted and shared only with trusted individuals.

NEXT VPS13 FORUMS

Dates for your diary:

- **26 January 2026**
- **27 April 2026**
- **27 July 2026**
- **26 October 2026.**

The exact times and topics will be announced nearer the time in the email invitation you will receive from Dr Kevin Peikert and also on all our social media channels.

Thank you!

ADVOCACY FOR NEUROACANTHOCYTOSIS PATIENTS

NA ADVOCACY USA
NEUROACANTHOCYTOSIS ADVOCACY USA, INC.

info@naadvocacy.org

www.naadvocacy.org www.naadvocacyusa.org