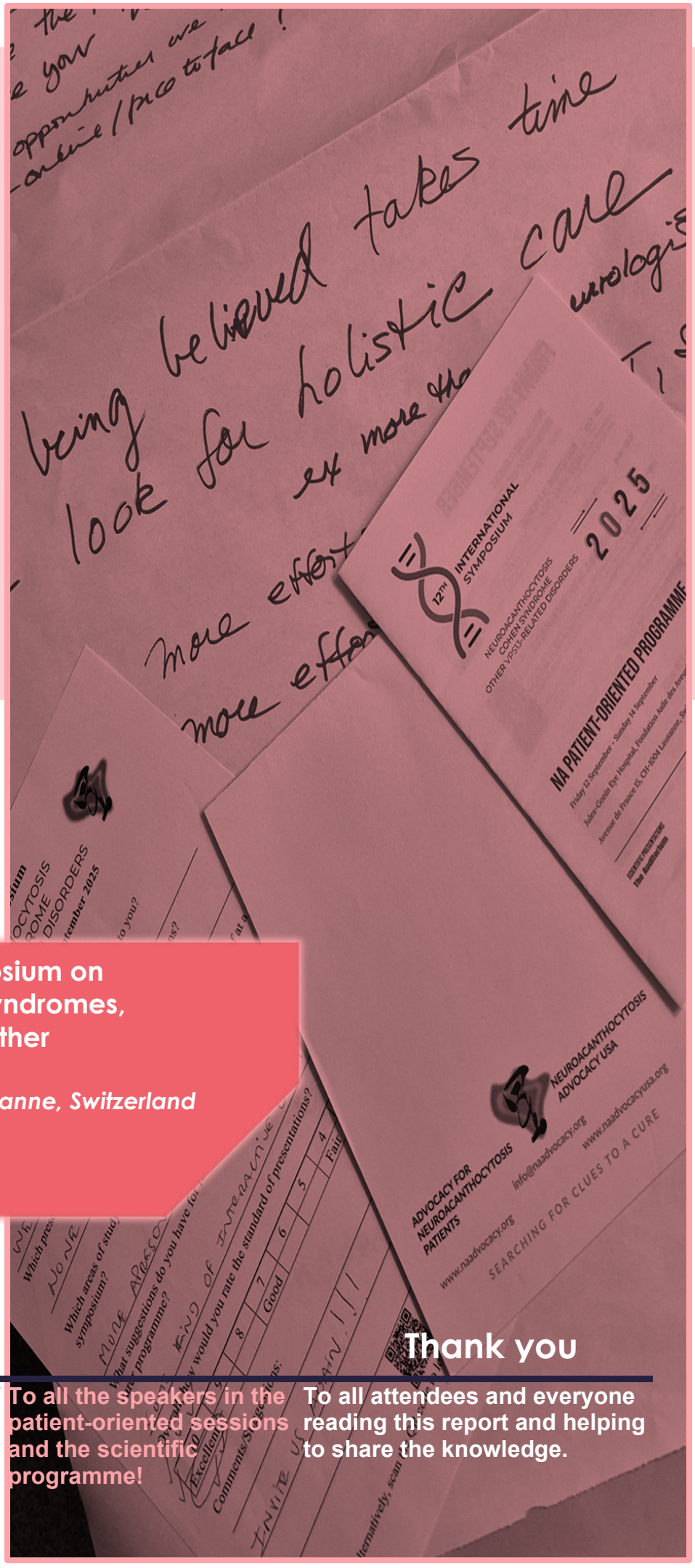

12th International Symposium on Neuroacanthocytosis Syndromes, Cohen Syndrome and other VPS13-related disorders

12-14 September 2025, Lausanne, Switzerland

REPORT

Thank you

To the organisers and hosts of the 12th International Meeting on Neuroacanthocytosis, Cohen Syndrome and other VPS13-related disorders

To all the speakers in the patient-oriented sessions and the scientific programme!

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

INTRODUCTION

Organisers:
Collaboration between the NA community and Cohen syndrome community

Location:
Jules-Gonin Eye Hospital, Lausanne, Switzerland

The Symposium aims:

- to provide a platform for knowledge exchange, focusing on the latest advancements in understanding and managing VPS13-associated diseases
- to advance multidisciplinary solutions and accelerate the development of cutting-edge therapies.

Attendees:

- Over 50 scientists, researchers and clinicians
- 15 patients (both VPS13A and XK), family members and carers.

Programme:

- Scientific track
  - Two keynote lectures
  - Eight sessions, including poster presentations and viewings

- Patient-oriented track:
  - Three presentations on occupational therapy, mental health tools and speech & swallowing therapy. Read on for the separate detailed reports of these sessions. These will be particularly beneficial to our patients, families and carers.
  - Theatre play – Read on more about it and see how you can watch it.
  - Reiki sessions – We offered our patients, families and carers taster sessions and you can read more about it.

Day	Scientific	Patient-oriented	Scientific
1	Keynote Lecture 1 – Clinical Perspectives	1 – Occupational Therapy Reiki sessions	2 – Lipid Transport Proteins 3 – Poster Presentations
2	Keynote Lecture 4 – Model Organisms	2 – Mental Health 3 – Theatre Play 4 – Speech and Swallowing Therapies	5 – Advances in VPS13A Research 6 – Poster viewing
3	7 – Cohen Syndrome	Reiki sessions	8 – Clinical New Directions Glenn Irvine Prize Lecture

DAY 1 - 12 SEPTEMBER 2025

The 12th International Meeting on Neuroacanthocytosis, Cohen Syndrome, and other VPS13-related disorders commenced with the welcome from Dr Muhammad Ansar and Dr Thomas Wolfensberger, Director of Jules Gonin Eye Hospital, University of Lausanne, Switzerland setting the tone for a weekend of deep scientific exchange and community connection.

Dr Wolfensberger acknowledged the unique convergence of clinicians, researchers, patients, and advocates gathered to explore the complexities of VPS13-associated diseases. His address emphasized the importance of multidisciplinary collaboration in tackling rare neurogenetic conditions. He highlighted the hospital's role not only as a host but as a symbol of the kind of integrated care and research the symposium aims to promote. The setting itself, with its tiered lecture hall and portraits of pioneering figures, reinforced the legacy and ambition behind the event.

Dr Wolfensberger and Dr Ansar also paid tribute to the enduring efforts of advocacy organisations and families whose lived experiences continue to shape the research agenda. They highlighted the symposium's commitment to putting the patient voices at the heart of it, fostering translational research, and accelerating therapeutic innovation.

Keynote Lecture

Disorders of Bulk Lipid Transfer, An Emerging Disease Category

Professor Dr Adrian Danek

Professor Danek opened the scientific programme with a thought-provoking keynote that invited everyone to rethink how we understand certain rare diseases. His talk focused on the group of conditions linked to changes in the VPS13 genes, which are involved in moving lipids (fats) between different parts of our cells. These lipids are essential for keeping cells healthy and working properly, especially in the brain and nervous system.

Rather than looking at each condition in isolation, Professor Danek suggested that we might be seeing a bigger picture: a new category of diseases caused by problems in this lipid transport system. He explained that the VPS13 gene family includes VPS13A (linked

to chorea-acanthocytosis), VPS13B (Cohen syndrome), VPS13C (a form of Parkinson's), and VPS13D (associated with movement and coordination difficulties).

These genes help shuttle lipids between places like the mitochondria (the cell's energy centre), the endoplasmic reticulum (a kind of cell factory), and other important structures.

Using clear visuals (and even a bit of humour), he showed how scientists are working to group these conditions based on how the proteins look and behave, rather than just the symptoms they cause. This approach could help researchers find shared treatments and better understand why these diseases affect so many different parts of the body.

Professor Danek also acknowledged the challenges: these conditions don't always fit neatly into current medical categories, and symptoms can vary widely. But he emphasized that by studying the biology behind them, we may be able to identify new markers, like blood tests or imaging signs, that could help with earlier diagnosis and more targeted care.

Throughout his talk, he celebrated the power of collaboration. He showed photos of past symposiums and reminded everyone that progress happens when scientists, researchers and clinicians, and patients, their families and carers work together. By joining forces and embracing new ways of thinking, we can move closer to treatments that make a real difference in people's lives.

Take-home message: Even though these conditions are complex and still not fully understood, there is growing momentum in the scientific world to treat them as part of a shared family. That means more opportunities for research, more chances for therapies, and, most importantly, more hope for the future.

Scientific Session #1

Clinical perspectives of VPS13A / XK / Cohen Syndrome

Clinical Management in VPS13A / XK Disease

Professor Ruth Walker

Professor Ruth Walker opened the clinical session with a compassionate and practical overview of how VPS13A disease (also known as chorea-acanthocytosis) and XK disease (also known as McLeod syndrome) are managed in real-world settings. She explained that these conditions often begin with subtle changes, such as mood shifts, obsessive behaviours, or movement difficulties that can be hard to recognise at first. Over time, symptoms may include involuntary movements, speech and swallowing challenges, and changes in thinking or behaviour.

Her talk emphasized that each person's experience is unique, even among family members with the same diagnosis. Because of this, care must be tailored to the individual, often involving a team of specialists - neurologists, psychiatrists, therapists, and social workers. She also discussed treatment options, including medications for movement and psychiatric symptoms, and the potential role of deep brain stimulation in some cases.

Take-home message: You are not alone, and while there's no one-size-fits-all approach, there are many tools and strategies to help manage symptoms and support wellbeing over time.

Building Bridges to the Future: Insights from Two Decades of Comprehensive Medical Care for Over 100 Cohen Syndrome Patients

Dr Heng Wang

Dr Wang shared reflections from over 20 years of caring for more than 100 individuals with Cohen syndrome, a rare condition linked to changes in the VPS13B gene. His talk highlighted the value of long-term, holistic care, addressing not just medical needs but emotional and developmental support across the lifespan.

Take-home message: Long-term, personalized care can transform lives, and the lessons learned from one rare condition often help guide better care for others.

Update on VPS13A Disease

Dr Kevin Peikert

Dr Peikert offered a clear and compassionate update on VPS13A disease. He walked the audience through the wide range of symptoms that can appear, starting with subtle psychiatric changes like obsessive thoughts or mood shifts, and progressing to movement difficulties, speech and swallowing problems, and even seizures.

He emphasized that the disease can look very different from person to person, and even within the same family.

Dr Peikert also shared new insights into how the disease affects the brain and body, including findings from imaging and blood tests. He highlighted the importance of early recognition and the need for tailored care plans that address both physical and emotional challenges. His presentation included real-life examples and data from recent studies, helping families better understand what to expect and how research is moving forward.

Take-home message: VPS13A disease is complex, but growing knowledge is helping clinicians offer better care. With continued research and support, there is hope for earlier diagnosis, and improved symptom management.

Patient-oriented Session #1

How Occupational Therapy Supports People Living With NA Syndromes
Joana Valente

Occupational therapist Joana Valente, from ParkinsonNet, Luxembourg, led a session exploring how OT can help people affected by NA. Her talk offered practical strategies, real-life examples, and a compassionate look at how OT preserves independence and dignity.

Click to read the comprehensive report on this session, which is available on our website.

The feedback received from the participants recommends it to anyone affected by either of the NA syndromes.

Reiki Sessions
Sally Cowan

Reiki is a Japanese form of energy healing based on the concept of Ki – the universal life force that flows through all living things. It's a gentle, non-intrusive technique that channels the Reiki energy to balance the chakras (energy points), promote deep relaxation and instil a sense of well-being.

While Reiki does not replace conventional medical care, it may support healing processes, help to balance the immune system and encourage personal and spiritual awareness amidst busy lives.

Patients were the most interested in experiencing the 20-minute sessions provided by Sally during the first and last day (with some return customers!). The experience was different for each person and can be subtle, especially for a first-time recipient.

Scientific Session #2

Lipid Transport Proteins

Partner Proteins of Yeast VPS13

Professor Aaron M. Neiman

Professor Neiman opened the lipid transport session by taking the audience into the microscopic world of yeast cells, a model organism that continues to offer powerful clues about human biology. His talk focused on how the VPS13 protein in yeast interacts with other proteins to move lipids between different parts of the cell. These interactions are essential for maintaining healthy cell membranes and supporting vital processes like growth and division.

By studying yeast, researchers can uncover how similar proteins work in humans, including those linked to NA and other VPS13-related conditions.

Professor Neiman highlighted several partner proteins that help guide VPS13 to the right location in the cell, ensuring it can do its job effectively. These findings help build a clearer picture of how lipid transport works, and what might go wrong when these systems are disrupted by genetic changes.

Take-home message: Even the simplest organisms can teach us a lot. By understanding how VPS13 works in yeast, scientists are laying the groundwork for better understanding and eventually treating rare human diseases.

The Alkuraya-Kučinskas Syndrome

Dr Alexandre Reymond

Dr Reymond introduced the audience to Alkuraya-Kučinskas syndrome, a rare neurodevelopmental condition caused by changes in a gene involved in lipid transport. Though not directly related to NA, the talk offered valuable insights into how disruptions in similar cellular pathways can lead to complex symptoms affecting movement, cognition, and development. By studying these overlapping mechanisms, researchers hope to uncover broader patterns that could inform care across rare diseases.

Take-home message: Even when conditions differ, shared biology can bring communities together, and every piece of research helps build a fuller picture of how our cells work and how we might help them heal.

Architecture of a Native Bridge-like Lipid Transport Protein Complex

Dr Sarah A. Clark

Dr Clark's presentation explored the intricate structure of lipid transport proteins, specifically those that act like bridges between different parts of the cell. These proteins help shuttle lipds across membranes, a process that's vital for cell health and communication. Understanding their architecture helps researchers figure out how these proteins work, and what might go wrong in diseases where lipid transport is disrupted.

Take-home message: By mapping the shape and structure of these proteins, scientists are getting closer to understanding how cells stay balanced, and how we might fix things when they don't.

The Molecular Mechanism of Lipid Transport by Bridge-like Lipid Transfer Proteins

Dr Stefano Vanni

Dr Vanni delved into the molecular workings of bridge-like lipid transfer proteins, explaining how they move fats between cell compartments. His talk focused on the step-by-step mechanics of this process, offering insights that could one day help in designing treatments for conditions caused by faulty lipid transport.

Take-home message: The more we understand the fine details of how these proteins move lipids, the better equipped we are to tackle diseases that stem from their malfunction.

Revealing the Regulation of VPS13 Through the Analysis of Physical Interactions

Dr Joanna Kamińska

Dr Kamińska's presentation explored how the VPS13 protein is regulated inside cells, specifically by looking at which other proteins it physically interacts with. Using advanced lab techniques, her team identified several partner proteins that may help guide VPS13 to the right location or control when and how it works. These interactions could be key to understanding why VPS13-related diseases develop and how they affect different parts of the body.

She also discussed how changes in VPS13 might trigger stress responses in cells, and how certain pathways, like those involving calcium or nutrient sensing, could be involved. While the research is still unfolding, it adds another layer to our understanding of how this important protein behaves in health and disease.

Take-home message: By uncovering the network of proteins that interact with VPS13, scientists are getting closer to understanding how it's regulated, and how we might one day intervene when things go wrong.

Scientific Session #3 & #6

Poster Presentations & Viewing

Summary of Friday & Saturday sessions

The poster sessions brought together a vibrant mix of researchers, clinicians, and students, each contributing a unique piece to the puzzle of VPS13-related disorders. First there were short 5-minute presentations in the Auditorium for each poster exhibited just outside, in the networking space. Later the attendees wandered from board to board,

engaging in lively conversations and pausing to absorb the latest findings, from molecular mechanisms to animal models and computational tools.

There were posters that directly addressed VPS13A and XK disease. One explored autopsy findings in XK disease. Another looked at how the loss of VPS13A affects mitochondrial metabolism in neurons, offering new clues about how energy production might be disrupted in VPS13A disease. Another, from the University of Cambridge, presented compelling data on motor function and brain degeneration in VPS13A-deficient mice, reinforcing the importance of animal models in understanding disease progression and testing future therapies.

Other posters expanded the conversation to related conditions and shared pathways. Studies on VPS13D and XK protein levels, in-silico lipid transport modelling, and all in all they all added depth to the collective understanding.

The sessions also featured research on Cohen syndrome, including a zebrafish model that revealed new roles for VPS13B in cilia function, an exciting direction for future exploration.

Take-home message: The poster session was a testament to the power of collaboration and curiosity. Each contribution, whether focused on NA syndromes or broader VPS13 biology, helps illuminate the path toward better understanding of the disease mechanisms, and ultimately how these can be repaired and made functional.

DAY 2 - 13 SEPTEMBER 2025

Keynote Lecture

VPS13 Family Proteins in Physiology and Disease
Professor Pietro de Camilli

Professor De Camilli opened the session with an overview of the four VPS13 genes found in humans, VPS13A, B, C, and D, and the distinct diseases linked to each. His talk focused especially on VPS13A and its partner protein XK, which are central to NA syndromes. Using powerful imaging tools like cryo-electron microscopy, his team has mapped the structure of VPS13A bound to XK, revealing how this complex helps move lipids between membranes inside cells. This lipid transfer is vital for keeping brain cells and red blood cells healthy.

He also explored how disruptions in this system might lead to disease. In striatal neurons, for example, faulty lipid handling at the plasma membrane could trigger stress, interfere with cell signalling, and contribute to the formation of acanthocytes (spiky red blood cells seen in NA syndromes). The presentation revisited the “bilayer couple” hypothesis from the 1970s, showing how changes in membrane shape and composition might explain some of the blood cell abnormalities. Professor De Camilli also shared striking images of VPS13A’s location in different cell types, including unexpected findings in cells missing XK suggesting there’s still much to learn.

Take-home message: By revealing how VPS13A and XK work together to maintain lipid balance, researchers are uncovering the cellular roots of NA syndromes, and opening new doors for understanding, monitoring, and eventually treating these rare conditions.

Scientific Session #4
Model Organisms

Human Anterior Neural Organoids as a Promising Model for Cohen Syndrome
Dr Woong Sun

Dr. Sun introduced a cutting-edge approach to studying Cohen Syndrome using human anterior neural organoids (tiny, lab-grown clusters of brain-like cells that mimic early development). These organoids offer a powerful new way to explore how mutations in the VPS13B gene affect brain structure and function, especially in regions linked to cognition and behaviour. By using patient-derived stem cells, researchers can observe how neural cells grow, connect, and respond to stress in a controlled environment. This model holds promise for uncovering the cellular changes that drive symptoms in Cohen Syndrome and may one day help test potential therapies.

Take-home message: Organoids are opening new doors to understanding Cohen Syndrome, bringing us closer to personalised insights and future treatments.

Characterization of Mouse Models of Cohen Syndrome to Better Understand Disease Pathogenesis
Dr Binnaz Yalcin

Dr Yalcin presented new research using mouse models to study Cohen Syndrome. These models help scientists observe how the disease develops over time, especially how it affects brain function, movement, and other systems. By comparing healthy and affected mice, researchers can pinpoint which biological pathways are disrupted and begin to understand why certain symptoms appear.

Take-home message: Mouse models are a powerful tool for uncovering how Cohen Syndrome works, and they may one day help guide treatments that target the root causes of the condition.

Muscle Dysfunction in VPS13A Disease: Progress from Animal Model to Human Muscle Biopsies
Professor Lucia De Franceschi

Professor De Franceschi shared new insights into how VPS13A disease affects muscle function, not just in the brain, but throughout the body. Her team has studied both animal models and human muscle biopsies, revealing signs of muscle damage and altered energy use. These findings suggest that VPS13A plays a role in keeping muscle cells healthy, and that its absence may lead to weakness or fatigue in some patients.

By comparing tissue samples from mice and people, the research helps bridge the gap between lab studies and real-life symptoms. It also opens the door to exploring new ways to monitor and support muscle health in those living with NA syndromes.

Take-home message: VPS13A disease may affect more than just movement and mood, it can impact muscle health too. Understanding this connection could lead to better care and even treatment options.

Patient-oriented Session #2

Mental Health Resources for Those Affected by NA Syndromes
Matt Bolz-Johnson

This session was led by Matt Bolz-Johnson, Mental Health Lead & Healthcare Advisor / Healthcare and Research Director, Eurordis - Rare Diseases Europe. Matt's talk explored emotional wellbeing, psychological support, and practical tools for coping with the challenges of living with a rare neurodegenerative condition. It also included reflections from breakout groups, where participants shared personal insights and experiences.

Click to read the comprehensive report on this session, which is available on our website.

The feedback received from the participants recommends it to anyone affected by either of the NA syndromes.

Scientific Session #5

Advances in VPS13A Research

An Update on Red Blood Cells as a Diagnostic Biomarker for Neuroacanthocytosis Syndromes

Professor Dr Lars Kaestner

Professor Kaestner presented new findings on how red blood cells, specifically the presence of acanthocytes, can help diagnose NA syndromes. Acanthocytes are red blood cells with spiky shapes, and they're often seen in people with VPS13A-related conditions.

His talk explored how these cells form, what they reveal about underlying disease mechanisms, and how they might be used more reliably in diagnosis. While acanthocytes aren't always present in every patient, advances in imaging and analysis are helping researchers better understand their role. Professor Kaestner emphasized the importance of combining red blood cell studies with other clinical signs to build a clearer picture of each person's condition.

Take-home message: Red blood cells may hold important clues for diagnosing NA syndromes, and ongoing research is helping make those clues easier to spot and understand.

VPS13 Expression in Red Blood Cells

Dr Lesley Bruce

Dr Bruce shared new findings on how VPS13 proteins, especially VPS13A, are expressed in red blood cells. Her team is exploring how these proteins contribute to the shape and stability of red blood cells, and what happens when they're missing or altered.

By studying VPS13 expression in blood samples, scientists hope to uncover patterns that could help with diagnosis or tracking disease progression. It's a step toward connecting what's happening inside cells with the symptoms patients experience.

Take-home message: Looking closely at red blood cells may help us understand how VPS13A works, and why its absence leads to the changes seen in NA syndromes.

Phenotypes in Induced Pluripotent Stem Cells (iPSC)-derived Neurons from Patients with VPS13A Disease – An Update

Dr Dajana Grossmann

Dr Grossmann shared new findings from studies using iPSC-derived neurons (lab-grown brain cells created from the skin or blood of patients with VPS13A disease). These cells allow researchers to observe how the condition affects neurons at a microscopic level, without needing invasive procedures. Her team has identified specific changes in cell shape, behaviour, and stress responses that may help explain symptoms like movement difficulties and cognitive changes.

This research brings scientists closer to understanding how VPS13A disease unfolds in the brain, and how future treatments might target the earliest signs of dysfunction.

Take-home message: Patient-derived neurons offer a powerful window into VPS13A disease, helping researchers track its effects and search for new ways to intervene.

Patient-oriented Session #3

"If I Get to Korea, I'll Tell You"
Theatre Play

José-Miguel Abrantes Figueiredo

One of the most striking moments of the Symposium came not from a lecture hall, but from a transformed meeting room-turned stage (through the kind support of our local hosts) for the performance of "If I Reach Korea I'll Tell You". This intimate, boundary-pushing work by Teatro do Zero featured actor and creator José-Miguel (Zé) Figueiredo, who lives with VPS13A disease.

The play is not about the disease, but the disease was present. It was not autobiography, yet it carried the weight of lived experience. What unfolded was not spectacle, but a raw, immersive act of creation, one that challenged the very structures of performance and perception.

Through silence, gesture, and vulnerability, Zé invited us into a space where difference is not a limitation but a source of artistic power. The performance asked difficult questions about who gets to be seen, heard, and valued in art and society. It offered no easy answers, only the courage to listen more deeply.

The theatrical moment was inspiring and a very different feature in the context of the scientific symposium. It reminded us that the body is not just a subject of science, but a force of expression. And that art, at its most honest, is a place where truth can be felt, not just understood.

With the help of his friend, Mauro Corage, who is also the director of the play, Zé incorporates theatre and some other art-based therapies in his wellbeing regime.

While we had permission to record the performance, we don't have the rights to share it publicly. Please email info@naadvocacy.org to request a link to watch it, but please do not distribute any further. Thank you for your understanding.

Patient-oriented Session #4

How to Keep the Joy of Communication and Eating and Drinking

Dr Elina Tripoliti

This session was led by Dr Elina Tripoliti, Clinical Specialist Speech and Language Therapist, University College London (UCL) - National Hospital for Neurology and Neurosurgery.

In this fourth and final session, Elina brought her deep clinical experience and warmth to topics that sit at the heart of living well with NA syndromes: communication, eating and drinking. She offered a compassionate look at how changes in speech, voice, and expression affect not only practical communication, but also identity, confidence, and connection.

Her presentation has also a call to action: to recognise communication as a core part of care, and to support people in expressing who they are.

Click to read the comprehensive report on this session, which is available on our website.

The feedback received from the participants recommends it to anyone affected by either of the NA syndromes.

DAY 3 - 14 SEPTEMBER 2025

Scientific Session #7
Genes to Therapy for Cohen Syndrome

This entire session spotlighted recent advances in understanding and addressing Cohen Syndrome. Researchers presented updates on how VPS13B mutations affect cellular function, particularly in neurons and the Golgi apparatus, and how these disruptions contribute to the syndrome's clinical features. Mouse models and patient-derived cells were used to explore disease mechanisms and identify potential therapeutic targets.

The session also included early-stage efforts toward therapy development, including gene correction strategies and functional rescue experiments. While the focus was distinct from NA syndromes, the shared involvement of VPS13 family proteins offered valuable context and potential crossover insights, especially in understanding how different VPS13 paralogs contribute to cellular health and disease.

Scientific Session #8
Clinical New Directions

The Clinical Perspective of Neuroacanthocytosis
Professor Dr Hans Jung

Professor Jung opened the clinical session with a comprehensive overview of NA syndromes, now more precisely defined as VPS13A and XK diseases. He emphasized their rarity, fewer than 1,500 cases worldwide, but also the likelihood of underdiagnosis due to overlapping symptoms with other movement disorders.

Drawing on decades of clinical experience, he outlined hallmark features such as progressive chorea, elevated CK levels, and the presence (though not always) of acanthocytes in blood tests.

He also highlighted the evolving understanding of the VPS13 gene family, noting how VPS13A and XK form a functional complex involved in lipid transport across cell membranes. This disruption in lipid handling may underlie both neurological symptoms and the red blood cell abnormalities seen in these conditions. Professor Jung stressed the importance of raising awareness among neurologists, adapting care strategies from related diseases like Huntington's, and building stronger networks between clinicians, researchers, and patient advocates.

Take-home message: NA syndromes are rare but increasingly understood. With better awareness, collaboration, and patient-cantered care, we can improve diagnosis and support for those affected.

Exploring the Role of VPS13B in Cohen Syndrome
Dr Wenke Seifert

Dr Seifert's presentation focused on the VPS13B gene and its role in Cohen Syndrome, rare condition affecting development, vision, and immune function. Her team is investigating how changes in VPS13B disrupt normal cell processes, especially in the brain and immune system. By studying patient cells and genetic models, they aim to better understand how the disease develops and how symptoms vary from person to person.

Take-home message: Understanding how VPS13B functions in cells is key to unlocking the causes of Cohen Syndrome, and may guide future treatments.

Glenn Irvine Prize Lecture
2025 Winner

Getting to Know VPS13 Proteins - VPS13D and the Cellular Landscape of Disease
Dr Marianna Leonzino

We were delighted to award this year's Glenn Irvine Prize to Dr Marianna Leonzino, in recognition of her outstanding contributions to research on the VPS13 protein family.

Her work has spanned from groundbreaking discoveries during her postdoctoral training in Pietro De Camilli's lab at Yale, to leading her own research group at the Institute of Neuroscience of the CNR at Humanitas.

In her words: "My interest in the VPS13 protein family began during my postdoctoral training [...] at Yale University. I contributed to seminal discoveries showing that VPS13 proteins act as lipid transfer proteins and localize at specific points where cell membranes meet. I also helped identify their binding partners and clarified how different human VPS13 family members (especially A, C, and D) connect to distinct cellular functions.

In recent years, I have been leading my own research group focusing on VPS13D. Because completely removing this protein is lethal for cells, we developed innovative models that allow us to study its function in living cells and in human neurons. These models are now giving us highly relevant insights into how VPS13D works, why its loss causes disease, and how this knowledge may guide future therapies."

Dr Leonzino delivered a powerful and visually rich presentation exploring the role of VPS13D in cellular health and disease.

Her talk traced the journey from molecular structure to patient impact, highlighting how VPS13D helps move lipids between organelles and supports mitochondrial function. Using advanced imaging and genetic tools, her team has shown that VPS13D is essential for maintaining healthy mitochondria, regulating autophagy (the cell's recycling system), and shaping neuronal development.

She introduced a new cellular model that allows researchers to "switch off" VPS13D temporarily, revealing how its absence leads to changes in mitochondrial shape, lipid composition, and neuron behaviour. Patient-derived stem cells carrying VPS13D mutations showed reduced neurite growth and altered electrical activity, offering clues to the movement and developmental symptoms seen in VPS13D-related disorders. The presentation closed with a heartfelt acknowledgment of collaborative efforts across continents and a call to keep pushing the boundaries of rare disease research.

The Glenn Irvine Prize honours the memory of our charity's co-founder and celebrates the work of young scientists whose research advances understanding of VPS13 and XK diseases. The idea and a large part of the Prize fund was a generous contribution from Carl and Betty Pforzheimer. Thanks to them, the Prize can continue to be awarded for many years to come, and for this we are very grateful.

Take-home message: VPS13D is a key player in keeping cells, and especially neurons, healthy. Dr Leonzino's work brings us closer to understanding how its disruption leads to disease, and how future therapies might restore balance.

Closing Remarks

Over three days in Lausanne, researchers and clinicians gathered to share the latest discoveries across the VPS13 spectrum, from molecular mechanisms to patient care. They were joined by amazing patients, family members, carers and members of the advocacy organisations.

Thank you to the organisers, the attendees and everyone else who supported the Symposium directly and indirectly!

The meeting offered a rich blend of scientific depth and community focus, with NA syndromes at the heart of many discussions, whether scientific or patient-oriented. Each presentation added a piece to the puzzle, bringing us closer to understanding, diagnosing, and ultimately treating NA syndromes and the other VPS13-related disorders. The work continues, but the path forward is becoming clearer one step at a time!

SEE YOU IN 2027 IN WARSAW, POLAND!

Thank you!

ADVOCACY FOR NEUROACANTHOCYTOSIS PATIENTS

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