MEETING REPORT
February 2025

Thank you

To the organisers and hosts:

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

To all our 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, who is now Professor Emeritus, and he is based in Munich, Germany.

The agenda had four main points:

- Regular updates from Advocacy for Neuroacanthocytosis Patients (NA Advocacy) and NA Advocacy USA
- Updates on the 12th International Symposium
- Presentation on lipid transport in cells
- Presentation on calcium homeostasis.

The audience was formed mainly of clinicians and researchers, some of them regular to the forum and some new. A few patients were also in attendance.

ADVOCACY UPDATES

Despina Dinca, Charity Manager at NA Advocacy gave a brief introduction of the charitable organisations, their purpose and aims:

- Information and support to potential or diagnosed patients worldwide
- Improvement of diagnosis times and available therapies
- Research funding.

It was highlighted that this year the Glenn Irvine Prize will be open for entries and the winner will be announced at the 12th Symposium in September 2025. The £5,000 is awarded for development and progress of young scientists in neuroacanthocytosis (NA) related fields.

Patients who are championing fundraising for the charities were acknowledged and thanked for their valuable contributions. Drew, based in USA organised special Super Bowl campaigns for two consecutive years; Alex published an art book and pledged the profit made from sales to the two charities. Her work will also be featured in “More than you can imagine: an anthology of rare experiences” which will be launched on Rare Disease Day on 28 February 2025.

UPDATE–12TH INTERNATIONAL SYMPOSIUM

Dr Muhammad Ansar presented the summary of the preparations for the 12th International Symposium, which this time will be a collaborative project between NA, Cohen Syndrome and other VPS13-related disorders.

When: Friday, 12 September to Sunday, 14 September 2025
Where: Jules-Gonin Eye Hospital, Lausanne, Switzerland

Most speakers in the provisional program are now confirmed and preparations are underway to finalise the exact schedule. The registration for patients, their family members and carers is free. We will publicise the registration link as soon as it becomes available.

The closest hotel which offers easy access to the venue is Ibis Lausanne Centre. We are checking the accessibility facilities at a number of hotels and we will update those interested in the invitation to the Symposium. Follow us online, on social media for the latest updates.

PRESENTATION – LIPID TRANSPORT

Dr André Nadler from Max Planck Institute of Molecular Cell Biology and Genetics, Dresden, Germany gave a presentation titled **"Quantitative Imaging of species-specific lipid transport in cells"**.

He explained the chemical diversity of lipids. A typical mammalian cell contains approximately 2000 lipid species. Lipid dysregulation appears when the body's lipid metabolism is disrupted. It can be linked to a number of health conditions, including obesity, diabetes, and neurodegenerative disorders.

Gaining an insight into the mechanisms of the lipids in cells requires the ability to track these lipids. For the proteins there are certain strategies available; for the lipids it's all novel and Dr Nadler's team researched a number of areas:

- Determine fundamental parameters (quantitative kinetics) of the lipid transport
- Investigate if the lipid composition of organelles (organised or specialised structures within a living cell) is determined by transport or metabolism
- Determine what drives the directionality of transport (from outside to the inside).

After the presentation several questions from researchers and patients were answered. We learned that the project was done with a small team, specialist roles were required, and it's all quite unique at the moment. The team had to build some of the equipment they used for their research, because what was on the market was not suitable for the intended purposes. Unfortunately, this research set up could not be extended to be applied at the ongoing research on post-mortem tissue which takes places currently at the Icahn School of Medicine. However, there is subsequent research work carried out looking at the other direction of the lipid transport discussed here.

The patient question was, “How does this impact VPS13A?’
In essence, these new research avenues show clear progress towards treatment approaches by better understanding the mechanisms of the lipids and their transport.

PRESENTATION – CALCIUM HOMEOSTASIS

Dr Dajana Grossmann from the Translational Neurodegeneration Section “Albrecht Kossel”, University of Rostock, Germany gave a presentation titled **“Membrane contact sites and calcium homeostasis in fibroblasts of VPS13A disease patients”**.

A fibroblast is a type of cell that contributes to the formation of connective tissue, a fibrous cellular material that supports and connects other tissues or organs in the body.

Calcium homeostasis is the process of maintaining a stable level of calcium in the cells.

Dr Grossmann’s research team explored the role of VPS13A in maintaining the health and function of cells focusing on how its absence can affect those with the condition. The research showed that VPS13A is an important organizer and regulator at sites of contact in different parts of the cell, particularly those involved in handling lipids and calcium.

Some of the key findings are:

- Lipid droplets: VPS13A is essential for the proper formation and function of lipid droplets, which are like storage bubbles for fats within the cell. Without VPS13A, these lipid droplets don't form correctly, suggesting a problem with the distribution and storage of fat within cells.
- Organelle communication: VPS13A helps different parts of the cell, called organelles communicate effectively. When VPS13A is missing, these communication lines are disrupted.
- Calcium regulation: VPS13A is vital for controlling calcium levels within the cell.
- Autophagy: VPS13A also plays a role in autophagy, the cell's recycling system. When VPS13A is deficient, this recycling process is impaired, potentially leading to a build-up of damaged components within the cell.
- Mitochondrial health: Mitochondria, the cell's powerhouses, are particularly affected by VPS13A deficiency. The mitochondrial function is compromised when VPS13A is missing, likely due to the combined problems with lipid handling, calcium regulation, and organelle communication.

In essence, the absence of VPS13A affects several critical cellular processes. This likely contributes to the symptoms of VPS13A disease, which include movement disorders and neurological problems.

This research helps us understand the complex role of VPS13A. By uncovering the specific processes affected by VPS13A deficiency, researchers hope to develop potential therapies for neuroacanthocytosis syndromes.

There were some specific questions at the end of the presentation which were answered by Dr Grossmann and sparked scientific exchange of useful information between the researchers and clinicians present.

NEXT VPS13 FORUMS

21st Forum – Monday, 28 April 2025
22nd Forum – Monday, 28 July 2025

The time will likely be 13:00 – 15:00 GMT.

The exact timings and agenda will be confirmed through the zoom invitation from Dr Kevin Peikert and through 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