MEETING REPORT
26 October 2026

Thank you

To the organisers and hosts:

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

To all our speakers!
To everyone who sent in patient related questions, and to the international panel who answered them.

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

TABLE OF CONTENTS

Introduction.....2
Advocacy Updates 2
12th Symposium 3
Q&A Session 3
Next VPS13 Forum 13

INTRODUCTION

The Forum was moderated by Professor Ruth Walker (RW), Department of Neurology, Mount Sinai School of Medicine, New York, USA.

The Forum was focused on the Q&A for patients, families and carers, as well as researchers and healthcare professionals interested in neuroacanthocytosis (NA) syndromes.

ADVOCACY UPDATES

Despina Dinca, Charity Manager for NA Advocacy gave a brief overview of what NA Advocacy and NA Advocacy USA stand for and what they've been up to in the past few months since the Forum in July:

- Continued collaboration
- Website redevelopment work
- Preparations for Rare Disease Day 2027.

RW announced the 13th Symposium joint with NBIA (neurodegeneration with brain iron accumulation) in Warsaw, Poland, between 16 and 18 September 2027. More details will follow as released by the organisers.

Q&A SESSION

Once a year the Forum opens a session for the questions asked by patients, their families and carers. The answers were discussed by an international panel of clinicians with extensive experience in VPS13A disease and XK disease.

The discussion focused on practical aspects of living with these conditions, current treatment options, ongoing research, and areas where important questions remain unanswered. While no single treatment works for everyone, the session highlighted the value of sharing clinical experience across centres and countries, helping families and healthcare professionals learn from one another.

The main clinicians who formed the ad-hoc panel:

- Professor Ruth Walker (RW), Department of Neurology, Mount Sinai School of Medicine, New York, USA

- Dr Kevin Peikert, Department of Neurology, University Medicine Rostock, Germany
- Professor Dr Hans Jung, Department of Neurology, University Hospital Zurich, Switzerland
- Professor Dr Adrian Danek, Department of Neurology, Ludwig-Maximillians University of Munich, Germany.

Thank you to them as well as everyone else who contributed along to the discussions.

Feeding dystonia

Are there any new treatments for tongue dystonia apart from botulinum toxin (Botox) injections? My son has been treated for about four years with Botox injections and they seem to be not as effective as earlier.

Answer:
Feeding dystonia is one of the most challenging symptoms experienced by many people with VPS13A disease.

Clinicians explained that feeding dystonia can cause involuntary tongue movements, forceful tongue protrusion, difficulties keeping food inside the mouth, biting of the lips and tongue, and significant challenges with eating and nutrition. Because these movements are often triggered by eating itself, they can be particularly distressing and difficult to manage.

The panel discussed the use of botulinum toxin (Botox being one of the best-known brand names) as one of the most widely used symptomatic treatments. When successful, injections can weaken overactive tongue muscles and reduce involuntary movements for several months before treatment needs to be repeated.

However, responses vary considerably between individuals.

Important: While some patients experience significant improvement, others receive little or no benefit despite careful targeting of different muscle groups. Clinicians emphasised that lack of response does not necessarily mean the treatment has failed permanently; adjustments to targeting or dosage may still be worthwhile. One potential side effect to be careful of is worsening of swallowing.

The discussion also highlighted how complex these symptoms can be. Some individuals lose benefit over time because different muscles begin contributing to the abnormal movements, while others never respond from the outset. The reasons for these differences remain poorly understood.

Medication options (for movement symptoms)

What about Tetrabenazine, has this medication been tried?

Answer:
Tetrabenazine remains one of the most commonly used treatments internationally and can reduce both chorea (sudden, uncontrollable movements) and dystonia (uncontrolled, repetitive, or sustained muscle contractions) in some patients. Clinicians noted that, for certain individuals, tetrabenazine has been transformative, particularly in reducing severe feeding dystonia for most known cases. However, in other cases, tetrabenazine didn't alleviate the intended symptoms and the side effects were outweighing the benefits.

Several other medications used to reduce involuntary movements were also discussed, including deutetrabenazine and valbenazine. In some countries, they are used to treat chorea associated with Huntington's disease and tardive dyskinesia (a persistent, movement disorder causing uncontrollable, repetitive, and abnormal muscle movements) in adults.

However, these treatments are not yet widely available throughout Europe, and clinicians expressed mixed opinions regarding whether they offer substantial advantages over the older and less expensive tetrabenazine.

There was interest also about the medication quetiapine. Although not traditionally used to treat dystonia, some clinicians reported surprisingly positive results when used at relatively high doses in a small number of patients whose feeding dystonia had not responded to tetrabenazine. While these experiences remain anecdotal and require further study, they provided a hopeful example of how sharing experiences across different clinical centres can uncover useful treatment approaches.

Important: A recurring theme throughout the discussion was that treatment remains highly individualised. What works extremely well for one patient may provide little benefit to another, meaning that symptom management often involves carefully trying different approaches and monitoring outcomes.

Deep Brain Stimulation (DBS)

Any known cases and known outcomes?

Answer:
The panel explained that DBS can sometimes be considered when involuntary movements are severe and significantly affect quality of life. Although the treatment has shown benefit in selected patients with VPS13A disease, the experts emphasised that DBS is a symptomatic treatment rather than a cure.

The greatest improvements are usually seen in excessive limb or body movements, whereas symptoms such as feeding difficulties, speech problems, gait impairment and other "axial" symptoms often improve less consistently. Benefits are typically seen for 1-2 years, and then the effect seems to wear off.

Important: Clinicians stressed the importance of setting realistic expectations. While DBS may reduce troublesome movements and improve daily functioning for a period of time, it does not stop the underlying disease process. Decisions about DBS therefore require careful consideration of both the potential benefits and limitations.

Understanding acanthocytes

Could a transfusion of clean blood cells have a positive impact to the patients?

Answer:
Acanthocytes are the "spiky" red blood cells seen in many patients, but not all. The panel explained that current evidence suggests acanthocytes themselves are unlikely to be the direct cause of neurological symptoms. Instead, they are viewed primarily as a visible sign of the underlying biological changes occurring in VPS13A and XK disease.

Researchers discussed how acanthocytes differ from normal red blood cells in their flexibility and membrane structure. Some scientists continue to investigate whether these abnormalities could affect blood flow through the smallest vessels of the brain, but at present there is no convincing evidence that replacing acanthocytes would alter disease progression. Also, these abnormal cells do not seem to cause any issues apart from a mild anemia. They don't clot or bleed any differently than regular cells.

Important: AD noted that most experts currently regard acanthocytes as an important clue to understanding disease mechanisms rather than a primary therapeutic target.

Gene Therapy

Hope, progress and remaining challenges

Answer:
The panel acknowledged the excitement surrounding recent advances in other neurological disorders, particularly Huntington's disease. However, they explained that translating those successes to VPS13A and XK disease is far from straightforward.

One major challenge is that the genetic defects differ fundamentally. Many Huntington's disease strategies aim to reduce production of a toxic protein, whereas VPS13A and XK disease are generally caused by the absence of a functioning protein. This means that researchers need to replace or restore the missing protein rather than switch it off.

For VPS13A disease specifically, researchers face the additional challenge that the VPS13A gene is exceptionally large. Current gene delivery systems struggle to carry genetic material of that size, making replacement therapies technically difficult.

The panel also discussed how much remains to be learned about disease mechanisms before genetic therapies can be developed. Understanding where the proteins act, which cells are most affected, and how the disease process unfolds remains essential groundwork for future treatments.

Important: The overall message was one of cautious optimism. New technologies continue to emerge, and lessons learned from other rare neurological diseases will likely benefit VPS13A and XK research in the future.

Research spotlight

"VPS13A Disease (Chorea-Acanthocytosis) in Puerto Rico; New Genetic and Clinical Observations"

While this was not a question per se, it's research work that's just been published in Movement Disorders Clinical Practice a week ago and it's available here: https://movementdisorders.onlinelibrary.wiley.com/doi/10.1002/mdc3.70866.

In this study, clinicians and scientists identified 18 people in Puerto Rico or from Puerto Rican families on the US mainland with confirmed VPS13A disease. They found that many of them shared the same four genetic changes, which suggests these may be "founder mutations" (variants that have been passed down through families over generations). This helps explain why VPS13A disease appears to occur more often in Puerto Rican families than expected. VPS13A disease is found worldwide, but there are a very large number of variants which cause the disease, and very often these are not the same, even in particular regions.

The study also showed that seizures were the first symptom for more than half of the patients. This is important because it reminds clinicians that VPS13A disease doesn't always start with movement symptoms, and that earlier genetic testing can help families get answers sooner. The average delay to diagnosis was almost 9 years.

Overall, the research highlights the need for better clinical awareness and easier access to genetic testing, so people can receive a diagnosis and support earlier in their journey. Because this condition is ultra rare, every piece of research helps families and clinicians understand it better.

LOOKING AHEAD

The Forum highlighted both the progress already made and the many questions that remain to be explored. It reinforced the importance of practical symptom management while researchers continue their search for disease-modifying therapies.

Although many challenges remain, the discussion demonstrated the strength of a collaborative community united by a common goal: improving understanding, treatment and, ultimately, outcomes for people living with VPS13A and XK disease.

NEXT VPS13 FORUM

28th Forum - Monday, 18 January 2027, 13:00 – 15:00 GMT. The topic will be announced nearer the time. The exact timings and agenda will be confirmed through the invitation from Dr Kevin Peikert.

Thank you!

ADVOCACY FOR NEUROACANTHOCYTOSIS PATIENTS

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