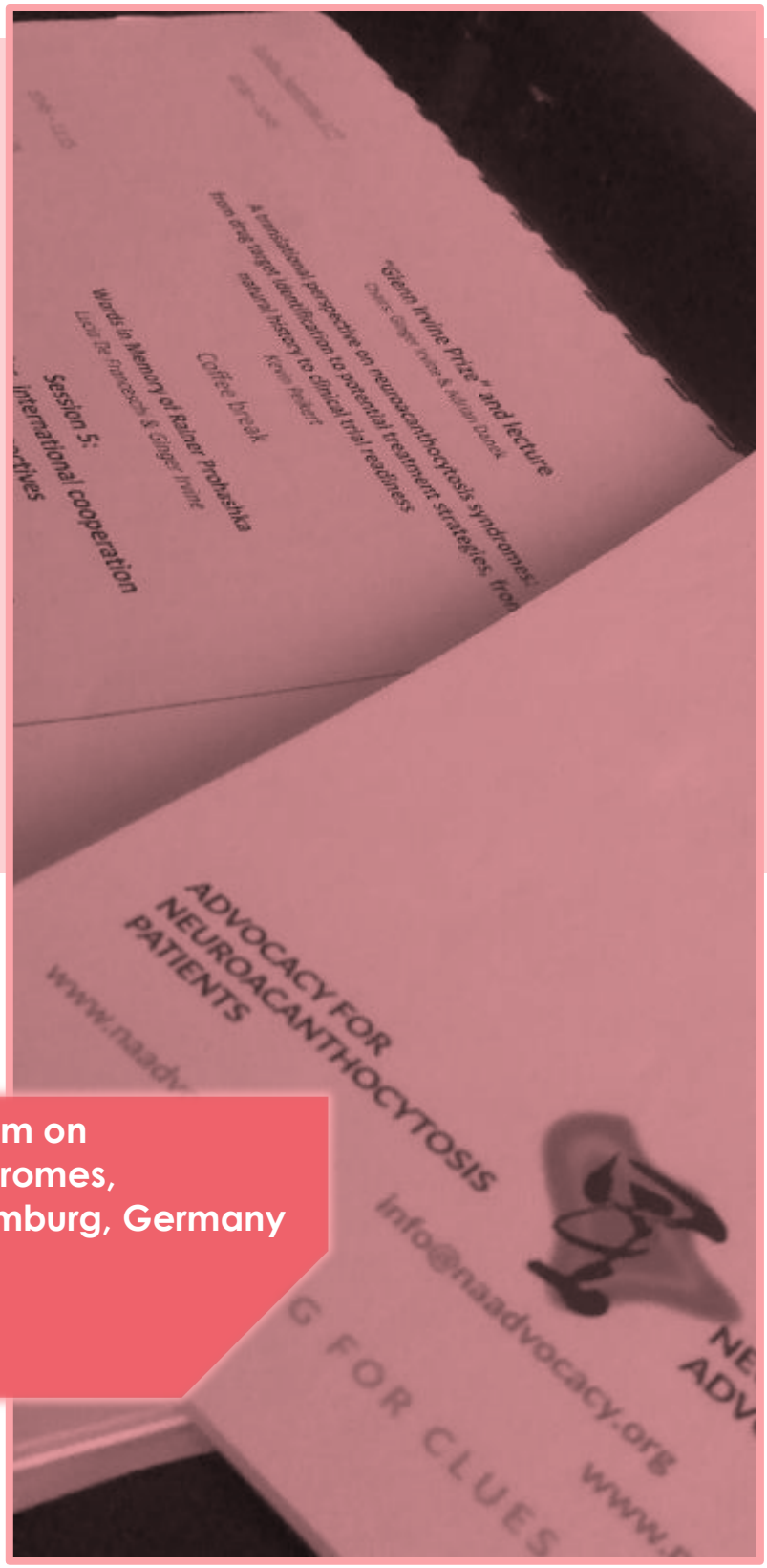

11th International Symposium on Neuroacanthocytosis Syndromes, 15-17 September 2023, Homburg, Germany

REPORT

Advocacy for Neuroacanthocytosis Patients

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DAY 1 - 15 SEPTEMBER 2023

The day started with an interesting opening from Professor Lars Kästner representing our hosts, Universität des Saarlandes Medical Centre in Homburg – thank you for hosting our event!

Keynote Lecture

Professor Adrian Danek took us through a fascinating evolution of the names for what we know nowadays as neuroacanthocytosis (NA) syndromes and the association with other groups of diseases. He also acknowledged the Public Service Award from the International Parkinson and Movement Disorder Society received by Ginger Irvine, our Chair and Co-Founder at their 2023 International Congress Welcome Ceremony.
Take-home point: Nomenclature changes - Chorea-acanthocytosis (ChAc) becomes VPS13A, McLeod Syndrome becomes XK Disease.

Keynote lecture 'Retiring the neuroacanthocytosis syndromes at 70 years?'- Professor Adrian Danek

Scientific session #1

The first scientific session saw three presentations on a newly acknowledged protein super-family, Bridge-Like Lipid Transfer Proteins (BLTP).

Patient session #1

The parallel session for the lay audience had two presentations:

- Dr Sriramya Lapa, University Hospital Frankfurt spoke about the swallowing difficulties (dysphagia) associated with VPS13A and XK diseases and how it can be treated and improved
- Dr Elina Tripoliti, University College London, spoke about communication, ways to improve it by tailoring the therapy for the VPS13A and XK patients.

It was so successful that an ad-hoc second part was organised after lunch at the request of the patients to answer more questions.

Take-home point: While there is a need for more research to be carried out in both areas, there are already many therapies and ways to work on improvement of both swallowing and communication difficulties of those affected.

Patient session #1 - Dr Elina Tripoliti

Scientific session #2

The second scientific session looked at the recent developments in the bulk lipid transfer disorders with four presentations from research groups in the USA, Switzerland, Poland and Germany.

Poster blitz session

The poster blitz session gave the PhD students whose posters were selected the opportunity to summarise their work in front of the entire audience. A total of eight posters were displayed in the foyer of the Library building. The discussions were lively, and we have the photos to prove it!

Take-home point: Research teams across the world are hard at work looking to understand faster the causes of the diseases and the to find ways to accelerate the paths towards effective treatments and therapies.

Poster session

Patient session #2

This is a long section, but so was the Q&A session due to its popularity!
An illustrious panel of clinicians and researchers facing a range of questions from a mixed audience of patients, family members, carers and advocacy representatives. Professor Adrian Danek, Professor Bernhard Landwehrmeyer, Professor Ruth Walker, Professor Andreas Hermann and Dr Kevin Peikert have kindly covered the topics raised by the audience in both English and German.

- There is no particular pattern on the geographical spread of the cases known to date.

- It's important to work with a local neurologist and where possible with a multidisciplinary team, who can be easily accessible. It's recommended to find a specialist in movement disorders or other disorders with similar manifestations to VPS13A and XK. It is recognised that the level of interest of a clinician unfamiliar with either VPS13A or XK diseases may vary. It is also recognised that different countries have different systems, but by getting in touch with us, the advocacy organisations, Advocacy for Neuroacanthocytosis Patients or NA Advocacy USA, Inc., you will have access to the clinicians involved with us which should be able to advise further based on the respective personal circumstances. It's important to provide comprehensive context which will help the clinicians to get a better understanding of each case.

- Continue to raise awareness in the local communities, as much as in the medical community. One of our colleagues, Joy Willard-Williford gave an example of how she volunteered Mark, her husband, to be examined by a specialist team and how persistence led to a good outcome. Speak about the symptoms, known facts and research which you are aware of. Use your own information and signpost those interested to our websites (which will be refreshed shortly!!) and social media for even more details.

- For the XK patients only – it's very important to remember to consider donating and have the blood stored accordingly and easily retrievable in case of emergency. An XK patient may have one first blood transfusion with normal blood, but after that they will require only their own blood.

- Work with local organisations which have care systems in place for those with similar disorders to VPS13A and XK (such as Huntington's Disease). If you are seeking relief as a carer or a family member, this may be a solution. Also keep looking into therapies and alternative therapies available and easily reachable for the patients. The earlier talks about the treatments to help with swallowing and speech mentioned this and any other therapies that encourage exercise (water exercise therapy, chair exercising, walking where possible, etc). When you find something that works, please don't hesitate to share your experience with others. Social media is usually the easiest way for this, but if in doubt, just contact either of our organisations.

Take-home point: Get in touch with us anytime, we're here to help!

Q&A session

DAY 2 - 16 SEPTEMBER 2023

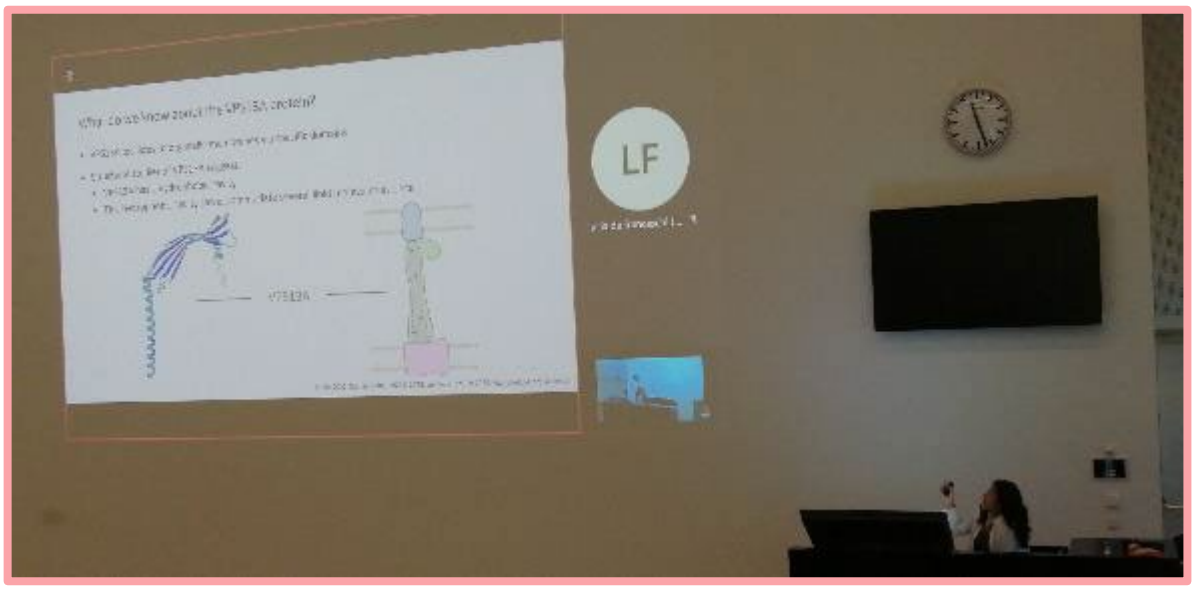
The first part of the day was dedicated to scientific sessions number three and four.

Scientific session #3

Disease insights from cell models – one research team from Germany and one from Spain presented their findings on VPS13A in their respective researched models. Another team from Germany spoke about their findings on VPS13B (Cohen syndrome) which is part of the VPS13 family of lipid transporters.

Scientific session #4

Disease insights from model organisms – this session saw four research teams from USA (New York and Michigan), Poland and Italy presenting their comprehensive findings.



Session #4 - Presentation 'Molecular mechanisms underlying chorea-acanthocytosis' - Genesis Rodriguez

Time out (patients and scientists)

After such an intensively scientific first part of the day, everyone had a chance to let their hair down and spend a few hours in nature at the Saarschleife Treetop Walk. This is a wooden trail running through the native mixed deciduous forest leading to a 42-meter-high platform that unravels amazing 360 degrees views (even when the sky was so overcast) of the area where the river Saar chose to make a spectacular U-turn on its course. The day finished with a wine tasting session in the small town of Perl situated at the intersection point of three borders of Germany, France and Luxembourg.
Take-home point: This one has very much reinforced the point of the first day, about the fact that there are research teams across the world hard at work approaching many angles and areas which will hopefully lead to finding the paths towards effective treatments and therapies faster.


View from deck of Saarschleife Treetop Walk


The viewing deck

DAY 3 - 17 SEPTEMBER 2023

Glenn Irvine Prize and lecture

Back into Symposium mode, the final day debuted with a very special session, the Glenn Irvine Prize and lecture. The Glenn Irvine Prize is awarded by the NA Advocacy to young scientists who demonstrate a keen interest in research related to VPS13A and XK disorders. Dr Kevin Peikert is based at the University of Rostock, and you can find out more about his work in our top story article in the current edition of NA newsletter.
His lecture 'A translational perspective on neuroacanthocytosis syndromes: from drug target identification to potential treatment strategies, from natural history to clinical trial readiness' raised very important points about where we are and the next steps we should look at taking.

Take-home points:

- We look forward to hearing about Kevin's progress!
- We are keen to hear from any young scientists about their research interest related specifically to VPS13A and XK disease, so please don't hesitate to get in touch with us if this is you or you know of someone who would be suitable for the next Glenn Irvine Prize.

Professor Adrian Danek, Dr Kevin Peikert and Ginger Irvine

There was a short session in memory of Professor Reiner Prohaska, who passed away recently. He was involved in pioneering research into red blood cells and made important contributions to NA research between 2013 and 2017.

Scientific session #5 (panel – scientists and patients)

The fifth scientific session on 'Patient registries, international cooperation and future perspectives' had a slightly different format and reunited all the scientists present together with the lay audience. The kick-off presentation from Professor Bernhard Landwehrmeyer looked at the possibility of a global patient registry for NA syndromes ('Huntingtonism'), i.e., VPS13A and XK diseases. He then chaired the subsequent conversations with the panel formed of Professor Adrian Danek, Ginger Irvine, Michaela Winkelmann, Dr Kevin Peikert, Susan Wagner, Professor Ruth Walker, Bob Metzger and Joy Willard-Williford. There is no doubt that there is a need for such a registry and its existence will be beneficial for both scientists and patients; however, the logistics of putting it into place and maintaining it has its challenges. But such registers do exist, and we heard the success story of 'Enroll-HD' which is the registry for Huntington Disease (HD) cases.

Take-home points:

- There is a lot of work to be done to develop the patient registry, but there are projects that were started on this previously and we could capitalise on them while we look at the success stories of existing registries and learn what helped to make them work.
- The advocacy organisations are here to help and support this work and inform the patients, their families and carers about it. Money is needed!

Scientific session #5 - Panel debate on 'Patient registries, international cooperation and future perspectives'

Scientific session #6

The last scientific session which looked at red blood cell related research took place simultaneously and it also went on while the final patient session brought together all the chairs of all the sessions (scientific and non-scientific).

Patient session #3

From the previous session there was a somewhat seamless segue into the final patient session, 'Learning from Huntington Disease experiences' with two very interesting presentations:

- Dr Alzbeta Mühlbäck, specialist in psychiatry and psychotherapy based in Munich, spoke to us about the stages of HD, how they are assessed, how they progress and their management and what support is in place. While she referred to Germany, the concept of the multidisciplinary care team approach for HD patients is very similar to the VPS13A and XK patients. The HD clinics in Germany are looking to coordinate:
    - Medical and clinical resources
    - Care services
    - Social and community resources
    - Long term & respite care.
- Michaela Winkelmann, President of the Deutsche Huntington-Hilfe e.V. (German Huntington's Disease Association), told us about the self-support organisation for those affected by HD. It's structured as a national non-profit association with regional branches, support groups and contact persons (volunteers). They organise meetings to exchange experiences (online or in person), they host lectures from experts which are open to families and specialists interested in HD. They also offer comprehensive information materials (printed or via the website) and a quarterly magazine. They are working tirelessly on communications campaigns and collaborate internationally with other HD-related organisations. The association is confronted with very similar challenges and unmet needs we have for the VPS13A and XK patients: rare disease and genetic disorder status; the currently incurable nature of the diseases; barriers in talking about the diseases from those affected.

Take-home points: We are grateful for all the information shared with us. We found it inspiring to understand how others make things work and we'll be looking to apply their success stories into our organisations to improve the support we offer, as well as developing other collaborations and communications channels to help us reach further and wider. Thank you to the speakers and we'll definitely stay in touch!

Last patient session - Michaela Winkelmann, Deutsche Huntington-Hilfe e.V.

The 11th International Symposium on Neuroacanthocytosis Syndromes came to a close late Sunday afternoon.

One last thing to add would be the dedicated experiences for the patients, family members and carers who were able to attend in person these extra options of the symposium:

- The acupuncture session
- Demonstration of Erysense live measurement of red blood cells
- Exercise diagnostics testing as part of a research project carried out at the University of Vienna.

Thank you to everyone involved in this and who took part.

Summary of Symposium (lay audience)

We say it was successful and inspiring! And we're supported in this by the panel which reunited the Chairs of all the sessions (scientific and non-scientific) who very kindly summarised the works of the three days for all the lay audience.

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