



NEUROACANTHOCYTOSIS
COHEN SYNDROME
OTHER **VPS13**-RELATED DISORDERS

2025

NA PATIENT-ORIENTED PROGRAMME

Friday 12 September - Sunday 14 September

Jules-Gonin Eye Hospital, Fondation Asile des Aveugles

Avenue de France 15, CH-1004 Lausanne, Switzerland

SCIENTIFIC PRESENTATIONS
The Auditorium

PATIENT RELATED SESSIONS
**Room Louis Braille 1, Room Louis Braille 2,
Room Elisabeth de Cejat**

LAST UPDATED: 2 SEPTEMBER 2025

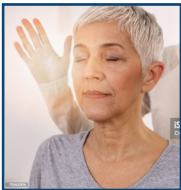
FRIDAY 12 SEPTEMBER

SPEAKERS

PARALLEL PATIENT SESSIONS:
NEUROACANTHOCYTOSIS SYNDROMES

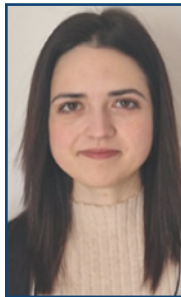
Chair: Ginger Irvine

11:00 - 13:00	REIKI SESSIONS (4 X 20 MINS) <i>Led by: Sally Cowan</i>
13:00 - 14:30	LUNCH BREAK
14:30 - 15:50	HOW CAN OCCUPATIONAL THERAPY HELP NA PATIENTS <i>Led by: Joana Valente</i> (to join remotely, please email info@naadvocacy.org and we'll send you the personalised MSTeams invitation)
15:30 - 16:00	GROUP PHOTO / COFFEE BREAK
16:00 - 17:30	REIKI SESSIONS (3 X 20 MINS) <i>Led by: Sally Cowan</i>
17:00 - 18:00	POSTER VIEWING SESSION / COFFEE BREAK Selected topics on VPS13-related studies, including cell and animal models
FROM 19:00	NETWORKING DINNER <i>Location: Aquatis Hotel Restaurant, Rte de Berne 148, 1010 Lausanne</i> <i>Sponsored by Cohen Syndrome Research Foundation</i>



ABOUT REIKI

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 instill a sense of well-being.



JOANA VALENTE

Occupational Therapist, Luxembourg

Joana is an occupational therapist working for the Luxembourg Red Cross in home-based care. Her practice focuses on older adults and individuals living with Parkinson's disease and other neurodegenerative conditions.

Joana is currently completing a Master's in Gerontology at the University of Luxembourg and she holds a Bachelor's degree in Occupational Therapy from Brussels.

As a trainer-in-training for ParkinsonNet Luxembourg, she is actively involved in promoting interdisciplinary collaboration and delivering practical strategies to support autonomy, upper limb function, and daily life participation. She is passionate about empowering both patients and caregivers through tailored interventions and education.



SALLY COWAN

Reiki Practitioner

Having lived in Japan in the 1990s, Sally is passionate about conscious ways of living, mindfulness practices and holistic health. She felt called to learn Reiki during the Covid pandemic as part of her own healing journey and qualified in Usui Reiki Levels 1 and 2 with the Reiki Maya London Centre in 2022.

SATURDAY 13 SEPTEMBER

SPEAKERS

PARALLEL PATIENT SESSION: NEUROACANTHOCYTOSIS SYNDROMES

Chair: Joy Willard-Williford

11:00 - 12:00

MENTAL HEALTH RESOURCES FOR THOSE AFFECTED BY NA SYNDROMES

Led by: Matt Bolz-Johnson

(to join remotely, please email info@naadvocacy.org and we'll send you the personalised MTeams invitation)

12:30 - 14:00

LUNCH BREAK

14:00 - 15:30

"IF I GET TO KOREA, I'LL TELL YOU"

Actor: José Miguel Abrantes Figueiredo

This is a theatre play performance in Portuguese - an English written translation will be provided for the audience.

The play was written by José Miguel, who is also delivering the monologue play. He is a VPS13A diagnosed patient. It is intended for a small audience and it lasts approximately 20 mins, so there may be two successive, identical performances.

15:30 - 16:00

COFFEE BREAK

16:00 - 17:30

HOW TO KEEP THE JOY OF COMMUNICATION AND EATING AND DRINKING

Led by: Dr Elina Tripoliti

(to join remotely, please email info@naadvocacy.org and we'll send you the personalised MTeams invitation)

17:00 - 18:00

POSTER PRESENTATIONS / VIEWING

Selected topics on VPS13-related studies, including cell and animal models



MATT BOLZ-JOHNSON

Mental Health Lead & Healthcare Advisor / Healthcare and Research Director, EURORDIS – Rare Diseases Europe

Matt has worked at EURORDIS since 2014 as a patient advocate. Matt has over 15 years experience working in the rare diseases field specialising in the selection and approval of expert centres in the NHS, England and for the European Reference Networks used by the European Commission. He also has supported the development of clinical networks for rare diseases at a national, regional and international level, ensuring patient groups to be active and be leaders in these networks.

Matt is leading the development of EURORDIS Mental Health Programme and Mental Health Partnership Network and coordinating advocacy actions as well as leading the development of a Mental Health Toolkit for Rare Conditions.

Matt is also an active advocacy lead for ATMPs working on EURORDIS Rare Impact initiative, Join4ATMP & ERDERA ATMP research, to streamline the ATMP development pathway and propose actionable solutions to improve patient access to cell and gene therapies in Europe.

Previously, Matt worked in NHS, England as a hospital manager and commissioner and specialised in rare disease and highly specialised healthcare.



DR ELINA TRIPOLITI

Clinical Specialist Speech and Language Therapist

Elina is a Clinical Specialist Speech and Language Therapist working at the National Hospital for Neurology and Neurosurgery, Queen Square, and an Honorary Senior Lecturer at the Department of Clinical and Movement Neurosciences, Institute of Neurology, UCL.

She is responsible for treating communication and swallowing problems in patients with Movement Disorders, mainly PD, PSP, MSA and Dystonia. She provides intensive therapies such as the Lee Silverman Voice Treatment (www.lsvtglobal.com) and the Expiratory Muscle Strength Training (emst150.com) and she has been a pioneer in delivering therapy through the internet. She founded the Movement Disorders Speech Lab for the investigation of acoustical, perceptual and aerodynamic aspects of speech.

She has a degree in Clinical Psychology from the University of Athens and a postgraduate degree in Speech & Language Therapy from City University, London. She was awarded her PhD in Neurological studies from UCL Institute of Neurology in 2010 (with Professor Limousin and Prof Hariz as mentors and supervisors) investigating the speech changes after Deep Brain Stimulation for patients with PD and Dystonia.

She is a founding member and a Trustee of the Sing for Joy Choir (singforjoychoirs.org.uk), and a firm advocate of the role of music, singing and dancing in speech rehabilitation.

"In the truest sense, Elina makes sure patients are heard" (www.parkylife.com/dr-elina-tripoliti)

SUNDAY 14 SEPTEMBER

09:00 - 10:30

REIKI SESSIONS (3 X 20 MINS)

Led by: Sally Cowan

12:00 - 12:30

THE 2025 GLENN IRVINE PRIZE WINNER TALK

Speaker: Marianna Leonzino



We are 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 presentation is titled "Getting to know VPS13 proteins".

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.”

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.

The first winner of the Glenn Irvine Prize was [Jae-Sook Park in May 2020](#). Dr Park is Senior Research Scientist in Professor Aaron Neiman's lab at Stony Brook, Long Island, NY, USA where she works with VSP13 proteins. Professor Neiman expects Dr Park will continue to make significant contributions to our understanding of how this protein family functions both in yeast and in human cells.

The second winner of the Glenn Irvine Prize was [Dr Kevin Peikert in 2023](#). The award was presented to the winner by Ginger Irvine and Professor Adrian Danek on 17 September 2023 at the 11th International Symposium in Homburg (Saar), Germany. Kevin is the organiser of the VPS13 Forum, which unites the international community of clinicians, scientists and families dealing with the VPS13-related disorder. The meetings are quarterly and [lay language reports](#) are published afterwards.

12:30 - 15:00

NETWORKING LUNCH / LUNCH ON THE GO

SEE YOU IN 2027!

NOTES



ADVOCACY FOR
NEUROACANTHOCYTOSIS
PATIENTS

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**A world where neuroacanthocytosis
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