SYMPOSIU[m]

12th International Meeting on Neuroacanthocytosis, Cohen Syndrome and other VPS13-related Disorders

Friday, September 12 - Sunday, September 14, 2025

Auditorium of the Jules Gonin Eye Hospital
Lausanne, Switzerland

Jules Gonin Eye Hospital
University department of ophthalmology
Fondation Asile des aveugles

About the Conference

This 12th edition of the International Meeting on Neuroacanthocytosis, Cohen Syndrome and other VPS13-Related disorders (this year organized by the Hôpital Ophtalmique Jules-Gonin and Fondation Asile des Aveugles and the Cohen Syndrome Research Foundation) aims to unite clinicians, researchers, patients, families, and advocacy organizations to foster collaboration, enhance disease management, and explore innovative treatment strategies for conditions associated with pathogenic variants in the VPS13 gene family. These include Neuroacanthocytosis syndromes (XK disease, VPS13A disease), Cohen syndrome, VPS13C-related Parkinsonism, and VPS13D-related ataxia.

The event's primary goal is to provide a platform for knowledge exchange, focusing on the latest advancements in understanding and managing VPS13-associated diseases. Patient and caregiver involvement will be central, ensuring their perspectives shape clinical care and future research directions. Through this collaborative approach, we aim to advance multidisciplinary solutions and accelerate the development of cutting-edge therapies.

Key Objectives

1. Enhance collaborations between scientists and clinicians researching VPS13-associated diseases.
2. Offer a forum for clinicians to discuss evidence-based best practices.
3. Involve patients, families, and advocacy organizations to guide research priorities and address daily challenges.
4. Explore novel treatment approaches leveraging modern biotechnologies and therapies.

Conference Rooms

Auditorium, Jules-Gonin Eye Hospital, Fondation Asile des Aveugles
Rooms Louis Braille 1 and 2 – Patients and relatives – Neuroacanthocytosis
Room Elisabeth de Cejat – Patients and relatives – Cohen Syndrome

Friday, September 12, 2025

08h30 - 09h00 **Registration and coffee**

Auditorium

09h00 - 09h30 **Conference Opening**
Chair: Muhammad Ansar

Welcome address
Thomas Wolfensberger

09h30 - 10h30 **Keynote Lecture**
Chair: Ruth Walker

Disorders of bulk lipid transfer, an emerging disease category
Adrian Danek

10h30 - 11h00 *Coffee Break*

11h00 - 12h30 **Session 1: Clinical Perspectives of VPS13A/XK/Cohen syndrome**
Chair: Lucia De Franceschi

Clinical Management in VPS13A/XK
Ruth Walker

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

Update on VPS13A Disease
Kevin Peikert

12h30 - 13h00 **Session 2: Lipid transport proteins**
Chair: Fabrizio Vacca

Partner proteins of yeast VPS13
Aaron M. Neiman

13h00 - 14h30 *Lunch Break*

14h30 - 15h30 **Session 2: Lipid transport proteins**
Chair: Fabrizio Vacca

The Alkuraya-Kučinskas Syndrome
Alexandre Reymond

Architecture of a native bridge-like lipid transport protein complex
Sarah A. Clark

15h30 - 16h00 *Group Photo and Coffee Break*

16h00 - 17h00 **Session 2: Lipid transport proteins**
Chair: Howard Riezman

The molecular mechanism of lipid transport by bridge-like lipid transfer proteins
Stefano Vanni

Revealing the regulation of VPS13 through the analysis of physical interactions
Joanna Kaminska

Room De Cerjat

14h30 - 15h30 **Parallel patient session 1: learning from the evolution of Cohen Syndrome**
Chair: Ashley Waterman

Evolution and management of retinal dystrophy and other eye-related phenotypes in Cohen syndrome
Nathalie Voide

Open discussion on overall disease management – parent's perspective

15h30 - 16h00 *Group Photo and Coffee Break*

16h00 - 17h00 **Parallel patient session 1: learning from the evolution of Cohen Syndrome**
Chair: Ashley Waterman

Discussion on orthopedic complications
Pierre-Yves Zambelli

Open discussion on overall disease management – parent's perspective

Room Braille

14h30 - 15h30 **Parallel patient session 2: Neuroacanthocytosis syndromes**
Chair: Ginger Irvine

How can occupational therapies help NA patients
Joana Valente

15h30 - 16h00 *Group Photo and Coffee Break*

16h00 - 17h00 **Parallel patient session 2: Neuroacanthocytosis syndromes**
Chair: Ginger Irvine

To be planned
To be planned

Auditorium

17h00 - 18h00 **Session 3: Short poster presentations**
Chair : Fabrizio Vacca

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

19h00 **Networking Dinner at the Aquatis Hotel Restaurant**
(Rte de Berne 148, 1010 Lausanne)

Saturday, September 13, 2025

Auditorium

09h30 - 10h30 **Keynote Lecture**
Chair: Adrian Danek

VPS13 family proteins in physiology and disease
Pietro de Camilli

10h30 - 11h00 *Coffee Break*

11h00 - 12h30 **Session 4: Model organisms**
Chair: Muhammad Ansar

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

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

Muscle dysfunction in VPS13A disease: progress from animal model to human muscle biopsies
Lucia De Franceschi

Room Braille

11h00 - 12h30 **Parallel patient session 3: Neuroacanthocytosis syndromes**
Chair: Joy Willard-Williford

Mental health resources for those affected by NA syndromes
Matt Bolz-Johnson

12h30 - 14h00 *Lunch Break*

14h00 - 15h30 **Session 5: Advances in VPS13A Research**
Chair: Ruth Walker

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

VPS13 expression in red blood cells
Lesley Bruce

Phenotypes in iPSC-derived neurons from patients with VPS13A disease – an update
Dajana Grossmann

Room De Cerjat

14h00 - 15h30 **Parallel patient session 4: Learning from the evolution of Cohen Syndrome**
Chair: Ashley Waterman

Understanding the genetics of CS and updates on therapeutic development
Muhammad Ansar

Disease management – clinical perspective
Heng Wang

Discussion with parents and caregivers on the progression and evolution of Cohen syndrome

Room Braille

14h00 - 15h30 **Parallel patient session 3: Neuroacanthocytosis syndromes**
Chair: Joy Willard-Williford

How to keep the joy of communication and eating and drinking
Dr Elina Tripoliti

15h30 - 16h00 *Coffee Break*

Auditorium

16h00 - 17h00 **Session 6: Short poster presentations**
Chair: Fabrizio Vacca

17h00 - 18h00 **Poster viewing session**

Sunday, September 14, 2025

Auditorium

09h00 - 10h30 **Session 7: Genes to therapy for Cohen syndrome**
Chair: Binnaz Yalcin

Understanding the genetics of Cohen Syndrome and potential therapeutic options
Muhammad Ansar

Drug discovery for the treatment of Cohen Syndrome
Fabrizio Vacca

Consequences of VPS13B deficiency beyond the Golgi
Jens Lüders

10h30 - 11h00 *Coffee Break*

11h00 - 13h00 **Session 8: Clinical New directions**
Chair: Lars Kaestner

The Clinical Perspective of Neuroacanthocytosis
Hans Jung

Exploring the Role of VPS13B in Cohen Syndrome
Wenke Seifert

The Glenn Irvine Prize talk
To be announced

13h00 - 15h30 **Networking Lunch and discussion on potential collaborations**

End of Conference

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RESEARCH FOUNDATION

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Science Foundation

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Access

No parking available at the venue
Parking du Centre SA, Rte de Genève 31
Parking de Beaulieu, Av. des Bergières 10

www.pms-parkings.ch

By bus from Lausanne train station :

- Bus 3 direction Bellevaux
- Bus 20 and 21 direction Blécherette

Stop at Chauderon

www.ophtalmique.ch