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

08h00 - 09h00 **Welcome Coffee and Registration**

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**
Chairs : Fabrizo Vacca

Partner Proteins of Yeast Vps13
Aaron M. Neiman

13h00 - 14h30 *Lunch Break*

14h30 - 15h30 **Session 2 : Lipid transport proteins**
Chairs : Fabrizo Vacca

The Alkuraya-Kučinskas Syndrome
Alexandre Reymond

A Journey on the Interorganelle Lipid Transportation Network
Benoit Kornmann

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

16h00 - 17h00 **Session 2 : Lipid transport proteins**
Chairs : Fabrizo Vacca

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

Revealing the regulation of Vps13 through the analysis of physical interactions
Joanna Kaminska

14h30 - 15h30

Parallel patient session 1 : learning from the evolution of Cohen Syndrome

Chairs : 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

Chairs : Ashley Waterman

15h45 - 16h15 **Discussion on orthopedic complic**
Pierre-Yves Zambelli

16h15 - 16h45 **Neuromodulation with microcurrent stimulation for low vision restoration and rehabilitation**
Prof. Bernhard Sabel, University of Magdeburg (GE)

Open discussion on overall disease management – parent's perspective

14h30 - 15h30

Parallel patient session 2 : Neuroacanthocytosis syndromes (XK disease - disease evolution)

Chairs : Despina Dinca

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XXX

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

16h00 - 17h00

Parallel patient session 2 : Neuroacanthocytosis syndromes (XK disease - disease evolution)

Chairs : Despina Dinca

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17h00 - 18h00 **Poster session**
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

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

VPS13 family proteins in physiology and disease
Pietro de Camilli

10h30 - 11h00 *Coffee Break*

11h00 - 12h30

Session 3 : 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

12h30 - 14h00 *Lunch Break*

14h00 - 15h30 **Session 4 : 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

14h30 - 16h00 **Parallel patient session 3 : 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

14h30 - 16h00 **Parallel patient session 3 : 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

14h30 - 16h00 **Parallel patient session 4 : Neuroacanthocytosis syndromes (XK disease - disease evolution)**
Chair : NA Advocacy – yet to be planned

15h30 - 16h00 *Coffee Break*

16h00 - 18h00 **Session 5 : Short poster presentations**
Chair : Fabrizio Vacca

Sunday, September 14, 2025

16h00 - 18h00 **Session 6 : 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*

16h00 - 18h00 **Session 5 : Short poster presentations**
Chair : Fabrizio Vacca

16h00 - 18h00 **Session 7 : 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

12h30 - 15h00 **Networking Lunch and discussion on potential collaborations**

End of Conference

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RESEARCH FOUNDATION

Swiss National
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