



NEW FRONTIERS IN NEUROACANTHOCYTOSIS AND NEURODEGENERATION WITH BRAIN IRON ACCUMULATION: FROM BENCHSIDE TO BEDSIDE

THE THIRD JOINT SYMPOSIUM ON NEUROACANTHOCYTOSIS AND NEURODEGENERATION WITH BRAIN IRON ACCUMULATION

October 30th - November 1st, 2014

La Palma Hotel - Stresa

AGENDA

THURSDAY, OCTOBER 30th

- 10 am-1 pm: 1st Parallel meeting of EMINA 2 (members please refer to the separate agenda)
1 pm - 3 pm: Parallel meeting of TIRCON (members please refer to the separate agenda)
- 1 pm - 3 pm: **Opening of the Secretariat and registrations**
- 2 pm- 3 pm: 2nd Parallel meeting of EMINA 2 (members please refer to the separate agenda)
- 3.15-3.30 pm: Welcome by the Organizers: **Lucia De Franceschi (COST)-Sonia Levi-Valeria Tiranti**
3.15-3.30 pm: Presentation of the COST action in EU research community: **Ricardo Escalante (COST)**

OPENING LECTURES

Chairs: **Lucia De Franceschi (COST) - Sonia Levi - Valeria Tiranti**

- 3.30-4.15 pm: **Ruth H. Walker**
Diagnosis and treatment of chorea and dystonia
- 4.15-5.00 pm: **Susan Hayflick**
News on NBIA
- 5.00-5.45 pm: **Tracey Rouault**
Does identification of multiple NBIA disease genes implicate common pathways in pathogenesis?
- 5.45-6.00 pm: Discussion
- 6.00-6.15 pm: Patricia Wood-Angelika Klucken: Announcement of Alliance Travel Grant Awards
- 6.15-8.00pm: Parallel meeting of TIRCON (members please refer to the separate agenda)
- 8 pm: **Aperidinner**



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FRIDAY, OCTOBER 31st

SESSION 1. PATHOPHYSIOLOGY AND BASIC RESEARCH IN CHOREA-ACANTHOCYTOSIS

Chairs: Adrian Danek (COST)-Andreas Hermann (COST)

- 8.30-9.00: **Susanne A. Schneider**
Clinical spectrum and differential diagnosis of ChAc
- 9.00-9.30: **Siamak Karkheiran**
Case Report: Chorea-acanthocytosis in Iran: focus upon therapies
- 9.30-10.00: **Giel Bosman**
Progress on red cells in NA syndromes: focus on ChAc
- 10.00-10.30: **Aaron Neiman**
Multiple functions of yeast VPS13
- 10.30-11.00: **Andreas Hermann (COST)**
News from EMINA-2
- 11.00-11.30: **Coffee break**
- 11.00-1 pm: **Parallel meeting of TIRCON (members please refer to the separate agenda)**
- 11.30-12.00: **Wenke Seifert**
Functional studies of the Cohen syndrome-associated protein VPS13B

Selected oral communications

- 12.00-12.15: **Mario Mairhofer**
Analysis of the effects of the knock-down of VPS13A in cell lines, iPS cells and primary cultures
- 12.15-12.30: **Sandra Muñoz-Braceras,**
The chorea-acanthocytosis protein VPS13A regulate autophagy in Dictyostelium and human HeLa cells
- 12.00-12.30: Discussion



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1 pm: **Lunch**

SESSION 2. PATHOPHYSIOLOGY AND BASIC RESEARCH IN NBIA

Chairs: Susan Hayflick-Tracey Rouault

2.15-2.45 pm: **Ody Sibon**
New insights in NA and NBIA using *Drosophila melanogaster*

2.45-3.15 pm: **Holger Prokisch**
On the way: From patients to genes to animal models to search for therapeutic approaches

3.15-3.45 pm: **Manju Kurian**
Novel Therapeutic Perspectives for PLA2G6-Associated Neurodegeneration

3.45-4.15 pm: **Thomas Klopstock**
News on TIRCON

Selected oral communications

4.15-4.30 pm: **Anthony Drecourt**
REPS1 is a novel gene of Neurodegeneration with Brain Iron Accumulation

4.30-4.45 pm: **Suh Young Jeon**
Disrupted blood-brain-barrier integrity is restored by CoA synthesis pathway intermediate in the Pank2^{-/-} mouse endothelial cells

4.45-5.15 pm: **Coffee break**

5.15-5.40 pm:

MASTER CLINICAL FORUM: AN INTERACTIVE SESSION ON RELEVANT CASE REPORTS WITH VIDEOS.

Chairs: Nardo Nardocci- Giovanna Zorzi

Selected oral communications

5.40-5.50 pm: **Petr Dusek**
Brain iron levels are not increased in heterozygous PANK2 mutation carriers



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5.50-6.00 pm: **Belen Pérez-Dueñas**

Pantotenate Kinase Associated Neurodegeneration: Clinical Assessment and Genetic Characterization by means of a Spanish Multi-Centre Research Network.

6.00-6.15 pm: Discussion

6.15 -7.45 pm: **Interactive Poster Session***

Chairs: **Enrico Bertini-Adrian Danek (COST)- Lucia De Franceschi (COST)**

*Posters will be hung starting from the morning and will be commented on by chairs during an itinerant discussion. Authors are required to stay at posters during this interactive session.

8 pm: **Conference dinner**

SATURDAY, NOVEMBER 1st

SESSION 3. PATHOPHYSIOLOGY AND BASIC RESEARCH IN MCLEOD SYNDROME

Chairs: Ruth Walker-Enrico Bertini

8.30-9.00: **Hans Jung**
News on McLeod Syndrome

9.00-9.30: **Beat Frey**
McLeod Mutations – Diagnostic Challenges

Selected oral communications

9.30-9.45: **Alicia Rivera**
Alterations in Magnesium and Potassium but Not Sodium Transporters Characterized
McLeod Syndrome Erythrocytes

8.30-10.00: **Parallel meeting of TIRCON (members please refer to the separate agenda)**



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LATE-BREAKING NEWS

Chairs: Ruth Walker-Enrico Bertini

Selected oral communications

- 10.00-10.15: **Stephen Zano**
Correction of Coenzyme A Deficiency using RE024 Phosphopantothenate Prodrug
- 10.15-10.30: **Balaji Srinivasa,**
External supplementation of Coenzyme A can directly influence cellular Coenzyme A levels and its dependent processes to rescue phenotypes associated with pantothenate kinase associated neurodegeneration
- 10.30-11.00: **Coffee break**

CONTRIBUTIONS FROM PATIENTS ASSOCIATIONS

NA Advocacy

- 11.00-11.30: NA Advocacy presents their work and Q&A (The NA Advocacy would like to present a timeline of their Advocacy work, both a chronological history and a yearly tasks/processes/events activities, sharing patients' stories, fundraising events and research ideas)

NBIA Alliance

- 11.30-11.45: **Natale Scalise**
NBIA Advocacy in Italy: a brief history of AISNAF and its aims for the future
- 11.45-11.55: **Patricia Wood**
The NBIA Alliance at a glance: short overview of the Alliance's development since 2012
- 11.55-12.15: **Antonio Lopez-Maayke van Schijndel/Alie Nagel-Jean-Loup Vasseur- Naveed Ali Self-Portraits of NBIA advocacies, developed under the patronage of TIRCON**
- 12.15-12.20: **Gustavo Andrés Marino-Michelle Whalen**
The situation for NBIA families in Argentina and Canada
- 12.20-12.30: **Question Time**
- 12.30-1 pm: Closing remarks, **Adrian Danek** (COST) and **Thomas Klopstock**
- 1 pm: **Farewell Lunch**