



## 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 30<sup>th</sup> - November 1<sup>st</sup>, 2014

La Palma Hotel - Stresa

## AGENDA

### THURSDAY, OCTOBER 30<sup>th</sup>

- 10 am-1 pm Parallel meeting of *EMINA 2*  
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: Parallel meeting of *EMINA 2*
- 3.15 Welcome by the Organizers: *Lucia De Franceschi (COST)-Sonia Levi-Valeria Tiranti*
- 3.15-3.30 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 *Ruth H. Walker*  
Diagnosis and treatment of chorea and dystonia
- 4.15-5.00 *Susan Hayflick*  
News on NBIA
- 5.00-5.45 *Tracey Rouault*  
Does identification of multiple NBIA disease genes implicate common pathways in pathogenesis?
- 6.00 pm: closing of work session
- 6.00-8.00 **Parallel meeting of TIRCON (members please refer to the separate agenda)**
- 8 pm: **Aperidinner**



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### FRIDAY, OCTOBER 31<sup>st</sup>

#### SESSION 1. PATHOPHYSIOLOGY AND BASIC RESEARCH IN CHOREA-ACANTHOCYTOSIS

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

- 8.30-9.00 *Susanne Schneider*  
Clinical spectrum and differential diagnosis of ChAc
- 9.00-9.15 *Siamak Karkheiran*  
Case Report: Chorea-acanthocytosis in Iran: focus upon therapies
- 9.15-9.45 *Giel Bosman*  
Progress on red cells in NA syndromes: focus on ChAc
- 9.45-10.15 *Aaron Neiman*  
Multiple functions of yeast VPS13
- 10.15-10.45 *Andreas Hermann (COST)*  
News from EMINA-2
- 10.45: **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
- 12.00- 12.15 Talk 1 selected from abstracts  
12.15-12.30 Talk 2 selected from abstracts
- 1 pm: **Lunch**



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#### SESSION 2. PATHOPHYSIOLOGY AND BASIC RESEARCH IN NBIA

*Chairs: Valeria Tiranti-Tracey Rouault*

- 2.15-2.45 *Ody Sibon*  
New insights in NA and NBIA using *Drosophila melanogaster*
- 2.45-3.15 *Holger Prokisch*  
On the way: From patients to genes to animal models to search for therapeutic approaches
- 3.15-3.45 *Manju Kurian*  
New Genetic Insights into Neurodegeneration with Brain Iron Accumulation
- 3.45-4.00 talk 1 selected from abstracts
- 4.00-4.15 talk 2 selected from abstracts

4.30: **Coffee break**

*Chairs: Enrico Bertini-Susan Hayflick*

- 4.45-5.15 *Thomas Klopstock*  
News on TIRCON
- 5.15-6.00 *Nardo Nardocci- Giovanna Zorzi*  
Master Clinician Forum: an interactive session on relevant case reports with videos  
2 case reports selected from abstracts with videos projection

*Zuhal Yapici*

6.00 -7.45 **Poster Session\***

*Chairs: Enza Maria Valente-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.



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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*  
The hematology of MLS

**9.30-11.00 Parallel meeting of TIRCON (members please refer to the separate agenda)**

9.30-9.45 Talk 1 selected from abstracts  
9.45-10.00 Talk2 selected from abstracts

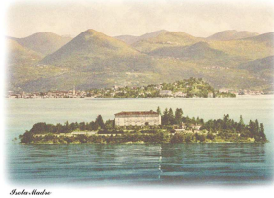
10.00-10.30 Late-breaking news

10.30 **Coffee break**

11- 12.30: **Contributions from Patients Associations**

12.30 am: Closing remarks, *Adrian Danek (COST)* and *Thomas Klopstock*

1 pm **Farewell Lunch**



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### HOW TO REACH THE CONFERENCE VENUE

The Symposium will be held at the **Hotel La Palma**, which is located on the lake side of Stresa, on the western shore of Lake Maggiore, 80 Km from Milan and 45 km from the Swiss border.

#### **By car:**

From downtown Milan, take the Highway "Autostrada dei Laghi" in direction of A8 - VARESE. After 30 km, take the Highway A26 in direction of GRAVELLONA TOCE. After 29 km, take the exit "CARPUGNINO" and then follow the directions to STRESA. Turn left on the lakeside SS33 and enter the city of STRESA. The Hotel La Palma is located on the left side on the road, just in front of the lake.

#### **From Milan Malpensa Airport:**

From Milano Malpensa Airport take the S.A.C.O bus until Gallarate Station and from here take a train to Stresa Station.

Please check the timetable of S.A.C.O buses at the following link:

<http://www.airpullman.com/altri/saco/ORARI%20GALL-MXP-CASTEL.%20DAL%2001.03.14.pdf>.

Please check the timetable of the **train** from Gallarate to Stresa here:

[www.trenitalia.com](http://www.trenitalia.com)

From Stresa train station, go right on Via G. Carducci; at the first cross, turn left on Via Duchessa di Genova; go straight until you find the lakeside and then turn right. The Hotel La Palma is on the right side of the road, facing the lake.

#### **By train:**

From Milano Centrale train station, take a train to the train station of Stresa.

The train takes about 70 minutes.

Please check the timetable on <http://www.trenitalia.it>.

From Stresa train station, go right on Via G. Carducci; at the first cross, turn left on Via Duchessa di Genova; go straight until you find the lakeside and then turn right. The Hotel La Palma is on the right side of the road, facing the lake.

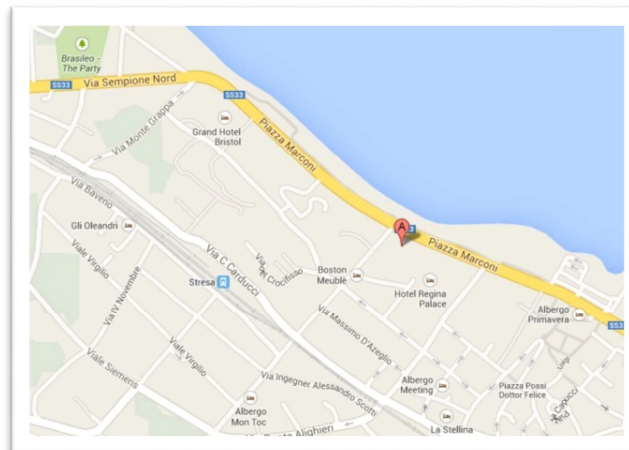


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### TOURIST INFORMATION AND USEFUL LINKS

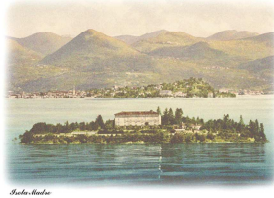
Stresa, the elegant internationally famous holiday resort, offers a unique variety and multiplicity of excursion possibilities: it is possible to visit Isola Bella, Isola Pescatori and Isola Madre; Villa Pallavicino, Villa Taranto, the Alpina Botanical Garden; Lago d'Orta, Mottarone and the ski resort of Macugnaga; the ancient monastery of Santa Caterina del Sasso, the Centovalli, not to mention Milan, capital of fashion and window shopping. Stresa is located near Milan's international airports: Malpensa (35 km) and Linate (110 km).

*The tourist office is located near by the Hotel La Palma, inside the lake port of Stresa.*

### STRESA - what to do

#### Villa Ducale and Rosminian Sites

The Villa Ducale is the oldest villa in Stresa. It was built by the Bolongaro family around 1770 and is located on the lakefront. From 1850 to 1855 it became the residence of the abbot and philosopher Antonio Rosmini who was considered the greatest 19th century Italian thinker. The most well known personalities of the Renaissance were guests at the villa. The philosophical conversations and discussions between Manzoni and Rosmini are particularly well known. In 1857, Elizabeth of Saxony, Duchess of Genoa decided to live in the Villa (hence the name Villa Ducale). Her daughter Margherita married Umberto I of Savoia and became Queen of Italy. Since 1966 the villa has been the headquarters of the International Centre for Rosminian Studies with the aim of promoting the thoughts



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and work of Rosmini and culture in general. The Study Centre is open from Monday to Friday 09.00-11.45 / 15.00-17.45 and you can visit the following: the Antonio Rosmini museum, Antonio Rosmini's room and the gardens. After Villa Ducale our itinerary continues to the monument dedicated to the friendship between Manzoni and Rosmini. It is located in Via Manzoni, half way to the Colle Rosmini, and was erected in 1932. The crucifix and the bas-relief are work of the sculptor Pietro Canonica. Following the road further you will reach the SS. Crocifisso Church at the Colle Rosmini. From here you can enjoy wonderful panoramic views. Here you can pause in front of the monument dedicated to Rosmini which is work of the sculptor Vincenzo Vela from Ticino. You can also descend into the crypt where the mortal remains of the philosopher are buried and where the tomb of the Rosminian religious poet, Clemente Rebora, is located.

#### **The Museum Pietro Canonica**

From 1898, for a period of seventeen years, Canonica was a frequent visitor to Stresa due to his friendship with the Duchess of Genoa, mother of Queen Margherita of Savoia. As a regular guest at the duchess's villa in Stresa he sculpted several bronze monuments for the town over a period of more than thirty years. In 1953, prompted by his strong ties with the town, Canonica donated a large number of his plaster and marble works to the City of Stresa, to be exhibited in a hall dedicated to him. Following a temporary arrangement in the Palazzo dei Congressi (Congress Palace), the sculptures by Canonica have now been restored and have found a more suitable home in a special exhibition room reserved expressly for his works within the Town Hall.

#### **BORROMEAN ISLANDS**

Isola Bella, named for Isabella, countess Borromeo, was originally a largely barren rock; after first improvements and buildings, opened by count Carlo III between 1629 and 1652, his son Vitaliano the 6th built an attractive summer palace, bringing in vast quantities of soil in order to build up a system of ten terraces for the garden. The unfinished building displays paintings by Lombard artists and Flemish tapestries.

Isola Madre, the largest of the three, is also noted for its gardens, which are maintained from about 1823 in an English style. Its palace, though uninhabited, is splendidly furnished with 16th- to 19th-century Italian masterpieces and paintings.

Isola dei Pescatori or Isola Superiore is now the only inhabited island in the archipelago. It has a fishing village, which in 1971 had a population of 208.

More information are available on <http://www.stresaturismo.it>.

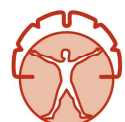
#### **Boats**

From the port of Stresa it is possible to reach every city located on the Lake Maggiore, including the beautiful Borromee Islands (Isola Bella, Isola dei Pescatori and Isola Madre). <http://www.navigazionealaghi.it/eng/>.



**PROTEOSTASIS**  
COST

**COST Action BM1307, PROTEOSTASIS**



International Parkinson and  
Movement Disorder Society