



Chelsea's Hope

Lafora Children
Research Fund

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Istituto delle Scienze Neurologiche
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Lafora disease science Symposium

Bologna, October 9th - 10th, 2023

Welcome

Dear colleagues and families, we are deeply honored to host the 2023 Lafora disease science symposium in Bologna, for the first time in Italy.

Though being an ultra-rare condition, a cluster of the disease is present in Italy and other Mediterranean areas, with a not negligible number of patients being observed with this disease in our country. Lafora disease has come to gradually unravel its pathogenesis and mechanisms, yet we are faced daily with the patients and their families in search of a cure.

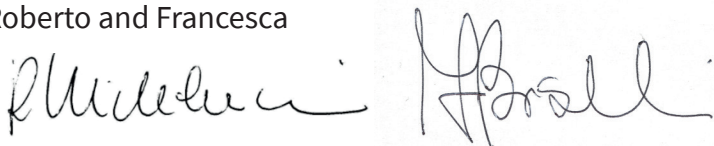
Bologna, since the Middle Ages, has been a place for people from all over the world to meet and exchange knowledge. The famous UNESCO World Heritage “porticoes,” which we invite you to visit in the city center, are symbolic of this fruitful cultural exchange for the development of new ideas and solutions, as they are a safe place where people can meet in any season and at any time of day.

We therefore trust that this meeting may bring substantial progress in the understanding of this condition and generate new insight into its potential treatments, giving the chance of a beneficial exchange between scientists, clinicians and families.

We thank Chelsea’s Hope for its unvaluable help in the organization of the meeting and financial support, along with the Italian Associations of patients (AILA and Tempo Zero) who sponsored some events of the symposium.

Therefore, again welcome to everyone!

Roberto and Francesca

Two handwritten signatures in black ink. The first signature is 'Roberto' and the second is 'Francesca'.

Gonzalo Lafora - oil painting on canvas
Fromkes, Maurice - Vilna (Lituania), 19.2.1872 - París, 27.9.1931
Madrid, Museo del Prado

Program

Monday, October 9th, 2023

ROOM GARDENIA

14:00 - 14:15 **Opening Remarks and talk**

Roberto Michelucci, Francesca Bisulli

14:15 - 14:45 **Welcome greetings**

- *Paolo Bordon* - General Director AUSL of Bologna
- *Raffaele Lodi* - Scientific Director IRCCS-ISNB
- *Pietro Cortelli* - Operative Director IRCCS-ISNB
- *Piero Cavallotti, Tullio Bressanello* - Italian Patient Associations

14:45 - 15:10 **Remarks from Chelsea's Hope**

Kit Donohue

Preview of Fighting the Rare Documentary

Chairpersons: *Roberto Michelucci, Berge Minassian*

15:10 - 15:30 **Rare genetic disease: how to stimulate research and enable clinical applications**

Francesca Pasinelli - General Director of Telethon Foundation

15:30 - 16:00 **Preclinical Therapies Developed for LD: where we are and future perspectives**

Matthew Gentry

16:00 - 16:20 **Discussion**

16:20 - 16:40 *Coffee break*

Pharmaceutical Session

Chairpersons: *Francesca Bisulli, Matthew Gentry*

16:40 - 17:00 **Current epilepsy medications for LD patients**

Alison Dolce

17:00 - 17:20 **ANLBIO& Gene Therapy for Lafora**

Christopher Lim

17:20 - 17:40 **New therapeutic approaches for Adult polyglucosan body disease**

Or Kakhlon

17:40 - 18:00 **Clinical markers and efficacy endpoints: proposal of a study protocol**

Lorenzo Muccioli, Luca Vignatelli

18:00 - 18:30 **General Discussion**

Program

Tuesday, October 10th, 2023

ROOM GARDENIA

Translational Science Research Session

Chairperson: *Matthew Gentry, José M. Serratosa*

- 08:30 - 08:50 **Gene replacement therapy in LD**
José M. Serratosa
- 08:50 - 09:10 **Hexose channeling in brain metabolism**
Matthew Gentry
- 09:10 - 09:30 **Gene and Antisense Oligonucleotide Therapies for Lafora disease**
Berge Minassian
- 09:30 - 09:50 **Reducing polyglucosan accumulation in astrocytes**
Mireia Moreno
- 09:50 - 10:10 **Zebrafish: A new tool to study Lafora Disease**
Stefania Della Vecchia
- 10:10 - 10:30 **New insight into LD pathogenesis from instant microwave-assisted brain fixation**
Felix Nitschke (remote)
- 10:30 - 10:50 General discussion
- 10:50 - 11:50 Coffee and Poster Session

Split Sessions: **LD Pathology & Mechanisms Track Vs. Patient Care Track**

ROOM GINESTRA

LD Pathology & Mechanisms Session

Chairpersons: *Jordi Duran, Pascual Sanz*

- 11:50 - 12:10 **Neuroinflammation and LD: Facts and opportunities**
Pascual Sanz
- 12:10 - 12:30 **Lessons learned from LD: toxic role of glycogen in ALS**
Jordi Duran
- 12:30 - 12:50 **Understanding the role of E3 ligase malin**
Sharmi Mitra
- 12:50 - 13:10 **New Knock-in LD mouse models**
Nerea Iglesias
- 13:10 - 13:30 **Discussion**
- 13:30 - 15:00 Lunch

ROOM GARDENIA

Patient Care Session

Chairpersons: *Francesca Bisulli, Lena Ismail*

- 11:50 - 12:10 **The Childhood Dementia Initiative**
Kris Elvidge (remote)
- 12:10 - 12:30 **LD in Italy: the perspectives from the parents and caregivers**
Piero Cavallotti
- 12:30 - 12:50 **Ask a Pharmacologist**
Dawn Eliashiv
- 12:50 - 13:10 **Patient Registry participation**
Maria Machio
- 13:10 - 13:30 **Chelsea's Hope: Caregiver and Sibling Support**
Niki Markou

ROOM GARDENIA

Clinical Markers Session

Chairpersons: *José M. Serratosa, Federico Vigevano*

- 15:00 - 15:20 **Lessons from the Natural History Study**
Dawn Eliashiv
- 15:20 - 15:40 **Lessons from the Patient Registry**
Maria Macchio
- 15:40 - 15:50 **Discussion on the basis of the prospective data of the Italian cohort**
Roberto Michelucci, Maria Tappatà
- 15:50 - 16:10 **Electro-clinical Features and Management of the late stage of LD**
Giuseppe d'Orsi
- 16:10 - 16:30 **Early biomarkers in six stages of LD: visual seizure, rising spike-wave index, REM loss**
Antonio Delgado-Escueta
- 16:30 - 16:50 **Awake and sleep EEG features in the natural history study**
Elena Pasini, Greta Mainieri
- 16:50 - 17:10 **Non-invasively Mapping Brain Glycogen with MRI**
Nirbhay Yadav (remote)
- 17:10 - 17:30 **FDG-PET Assessment and Metabolic Patterns**
Andrea Farolfi
- 17:30 - 18:00 **Discussion**
- 18:00 - 18:10 **Upcoming Patient Advocacy Events**
Chelsea's Hope, A.I.L.A, TempoZero, France Lafora, AEVEL
- 18:10 - 18:30 **Closing Remarks**
- 20:00 *Social Dinner at Restaurant Corbezzoli*

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