

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**

Pharmaceutical Session

Chairpersons: *Francesca Bisulli, Matthew Gentry*

16:20 - 16:40 **Current epilepsy medications for LD patients**

Alison Dolce

16:40 - 17:00 **ANLBIO& Gene Therapy for Lafora**

Christopher Lim

17:00 - 17:20 **New therapeutic approaches for Adult polyglucosan body disease**

Or Kakhlon

17:20 - 17:40 **Clinical markers and efficacy endpoints: proposal of a study protocol**

Lorenzo Muccioli, Luca Vignatelli

17:40 - 18:10 **General Discussion**

Program

Tuesday, October 10th, 2023

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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 **Increasing GBE1 activity to prevent LB formation**
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: *Pascual Sanz, Jordi Duran*

- 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**

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Patient Care Session

Chairpersons: *Lena Ismail, Francesca Bisulli*

- 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 Macchio
- 13:10 - 13:30 **Discussion**

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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**