

Saturday, Sept. 28

- 11.40 Mirella Domenica Elia:
Nutritional issues
Comparison of experiences
- 12.00 Antonella Cersosimo:
Neurorehabilitation in Italy
- 12.20 Carlos Maragoto:
Neurorehabilitation in Cuba
- 12.40 **Discussion: How to Manage Critical Situations**
Chair: Nardo Nardocci
All speakers and families
- 13.15 **Lunch**

English/Italian translator available

with unconditional contributions from

Retrophin[®]



Associazione Italiana
Sindromi Neurodegenerative
da Accumulo di Ferro

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2^o Family Meeting



Associazione Italiana
Sindromi Neurodegenerative
da Accumulo di Ferro

Rimini 27-28 Sept. 2019
Hotel Continental Rimini

Under the auspices of



Fondazione I.R.C.C.S.
Istituto Neurologico Carlo Besta

Sistema Socio Sanitario



Regione
Lombardia



Comune di Rimini



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PROGRAM

Friday, Sept. 27

Introduction to the Works

- 14.30 Welcome and opening message
Natale Antonio Scalise
- 14.40 Introduction to the meeting
Nardo Nardocci
- 14.50 Aisnaf research funding initiatives – quick update
Francesca Sofia

NBIA Research - State of Art and Perspectives for the Future

Chairs: Massimo Pandolfo ed Enrico Bertini

- 15.10 Agnès Rötig - “PANK2, PLA2G6, FA2H, C19ORF12, CRAT, REPS1: several NBIA genes result in the same deregulation of iron uptake”

PKAN

- 15.30 Ivano Di Meo - “Mitochondrial inborn errors of Coenzyme A biosynthesis-associated neurodegeneration: implementation of new disease models”
- 15.50 Lauriel Earley - “PANK2 gene therapy for treatment of PKAN and elucidation of disease biology”
- 16.10 Dario Finazzi - “Understanding and curing PKAN: advancements from the phenotypic rescue of a zebrafish model”

MPAN

- 16.30 Arcangela Iuso - “Development of a treatment strategy for MPAN”

Coffee break

BPAN

- 17.30 Robin Ketteler - “Modulation of autophagy as a potential therapy for BPAN”
- 17.50 Lena Burbulla - “Mechanistic insights into iron accumulation in WDR45 mutant neurons linked to β -propeller-associated neurodegeneration”
- 18.00 Penny Hogarth - “Update on BPAN”

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FAHN

- 18.20 Filippo Santorelli - “WHAT & FA2H: touring in a “simple” enzyme protein”

Overview on Ongoing Clinical Studies

- 18.40 Giovanna Zorzi – “Deferiprone and Fosmetpantotenate”
- 19.00 Barbara Garavaglia - Patient registers and biobanks: essential resources for research”
- 19.20 Susan Hayflick - “CoA-Z in PKAN”
- 19.40 Discussion
- 20.00 **Closing and Dinner**

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Therapies and Care: How to Improve the Quality of Life of People with NBIA Disorders

Chairs: Roberta Battini and Anna Scalise

- 09.00 Roberta Scalise:
Good clinical practices
- 09.20 Miryam Carecchio:
Medical therapy
- 09.40 Paola Soliveri:
Focal dystonia therapy
- 10.00 Francesco Motta:
Intrathecal baclofen therapy
- 10.20 Angelo Franzini:
Deep Brain Stimulation
- 10.40 **Coffee break**
- 11.00 Marco Piastra:
Respiratory Issues
- 11.20 Paola De Angelis:
Digestive issues