

19th CATANIA INTERNATIONAL SUMMER SCHOOL OF NEUROSCIENCE

July 18-22, 2022

Coordinator: Prof. Filippo Drago
Scientific Director: Prof. Michelangelo Mancuso

Rare Diseases and Neurosciences

Hotel Mercure Catania Excelsior, Piazza Giovanni Verga 39, 95129 Catania, Italy

PROGRAM

18th JULY

COMPLEXITY AND PERSPECTIVES ACROSS RARE NEUROLOGICAL DISEASES

- 08:30 Opening
Prof. Filippo Drago and
Prof. Michelangelo Mancuso
- 9:00-10:30 Morning lecture
The future of research in Rare Neurological Diseases
Dr. Holm Graessner, Managing Director of the ZSE Tübingen and Head of the Research Management Unit of the Institute of Medical Genetics and Applied Genomics at the University Hospital Tübingen, Germany.
- 10:30-11:30 Interactive discussion with **Dr. Rebecca Schuele-Freyer**
- 15:30-17:00 Afternoon lecture
Exploring Rare Movement Disorders
Prof. Kailash Bathia, FRCP, Professor of Clinical Neurology, Institute of Neurology, UCL Queen Square, London, UK.
- 17:00-18:00 Interactive discussion
- 18:00-19:00 General discussion

19th JULY

EXPLORING COMPLEX PHENOTYPES, LOOKING FOR THE ARIADNE'S THREAD

- 9:00-10:30 Morning lecture
Hereditary Ataxias
Prof. Dr. Thomas Klopstock, MD, FEAN, Friedrich-Baur-Institute, Department of Neurology, LMU University Hospital, Ludwig-Maximilians-University, Munich, Germany.
- 10:30-11:30 Interactive discussion with **Dr. Holm Graessner**
- 15:30-17:00 Afternoon lecture
Mitochondrial diseases. A scientific challenge
Prof. Michelangelo Mancuso, MD, PhD, Department of Clinical and Experimental Medicine, Neurological Institute University of Pisa & AOUP, Italy.
- 17:00-18:00 Interactive discussion with **Prof. Holger Prokisch** and **Dr. Rebecca Schuele-Freyer**

20th JULY

GENETICS, GENOMICS, OMICS: FROM THE BENCH TO THE BEDSIDE

- 9:00-10:30 Morning lecture
Genetics, Genomics and Multiomics approaches in the field of the Rare Neurological Diseases
Prof. Holger Prokisch, PhD, Head of the Research Group "Genetics of Mitochondrial Disorders" at the Institute of Human Genetics of the Technical University Munich and of the Helmholtz Zentrum München, Neuherberg, Germany.
- 10:30-11:30 Interactive discussion with **Prof. Dr. Thomas Klopstock**

- 15:30-17:00 Afternoon lecture with
Challenges of iPSC-based diseases modelling in Rare Neurological Disorders
Dr. Valeria Tiranti, Head of Molecular Pathology of Mitochondrial Diseases, Unit of Medical Genetics and Neurogenetics, Fondazione IRCCS Istituto Neurologico Carlo Besta, Milano, Italy.
- 17:00-18:00 Interactive discussion with **Dr. Rebecca Schuele-Freyer**

21st JULY

GROWING EXPERIENCES AND CURRICULUM

- 9:00-10:00 Training lecture
How to write a paper/project
Prof. Michelangelo Mancuso
- 10:00-11:30 Training in small groups
Participants will write a one-page project in small groups
- 14:30-16:00 Afternoon lecture
Muscular diseases: is still pathology a crucial step?
Prof. Massimiliano Filosto, MD, PhD, FEAN Associate Professor of Neurology, Department of Clinical and Experimental Sciences, University of Brescia, Italy.
- 16:00-16:30 Interactive discussion with **Prof. Dr. Thomas Klopstock**, **Prof. Holger Prokisch** and **Dr. Rebecca Schuele-Freyer**
- 16:30-18:00 Presentation of morning projects by participants
- 19:00 Social Dinner

22nd JULY

CURRENT AND FUTURE PROSPECTS FOR GENE THERAPY FOR RARE NEUROLOGICAL DISEASES

- 9:00-10:30 Morning lecture
Mutation-specific RNA therapies for ultrarare diseases: the n-of-1 challenge
PD. Dr. Rebecca Schuele-Freyer, Department of Neurodegenerative Diseases, Hertie Institute for Clinical Brain Research (HIH), Center of Neurology, University of Tübingen, Tübingen, Germany. German Center for Neurodegenerative Diseases (DZNE), Tübingen, Germany.
- 10:30-11:30 Interactive discussion with **Prof. Holger Prokisch** and **Prof. Massimiliano Filosto**
- 12:00 Closing remarks



Organizing Secretariat

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