

Lessons learned and further road to the therapies of genetic neurological disorders.

What we have, unmet needs and future perspectives

AND

XXXII Ottorino Rossi Award

IRCCS Mondino Foundation, Berlusconi Hall - November 18, 2022

The event will take place in hybrid form

Background to the conference

The starting point and inspiration for the scientific programme is the lecture by Prof. Muntoni, who will explain how, in recent years, translational research has managed to find a cure for spinal muscular atrophy, an extremely severe genetic disease that was long considered incurable. This was achieved through a combination of deep phenotyping, better understanding of the disease pathogenesis, and the development of innovative technologies aimed at correcting the underlying gene defect. Research into other dramatic neuromuscular diseases, such as Duchenne muscular dystrophy, on the other hand, has not yet been translated into effective therapeutic strategies, highlighting the existence of conceptual and methodological difficulties that remain hard to overcome.

The conference will highlight recent advances, unmet needs, and future perspectives in the quest for novel therapeutic strategies, looking at key examples in the fields of paediatric and adult inherited neurological disorders. Prof. Muntoni's lecture will be followed by two general lectures, the first dealing with the difficulties in designing clinical trials in rare diseases (especially those of childhood), and the second providing an overview of innovative technologies for the diagnosis and treatment of hereditary disorders. In the afternoon session, lectures will cover four neurological disease types (frontotemporal dementia, amyotrophic lateral sclerosis, metabolic diseases, and Parkinson's disease), focusing on their genetic basis, pathogenetic mechanisms, and most importantly, on current and novel therapeutic perspectives, whose development, although still conditioned by criticalities and difficulties, is destined to change the natural history of these still incurable conditions.

8.45 Registration and welcome coffee

9.30 Greetings from the Authorities

XXXII OTTORINO ROSSI AWARD CEREMONY

9.45 Presentation of the Winner

*Angela Berardinelli - Research Unit of Neuromuscular Diseases of Childhood and Adolescence,
IRCCS Mondino Foundation (Pavia)*

10.00 Lecture by the Winner

Lesson learned from novel therapies for childhood neuromuscular disorders

Francesco Muntoni - Dubowitz Neuromuscular Centre, University College London (UCL, London)

11.00 Delivery of the Prize

*Francesco Svelto (President) and Roberto Bergamaschi (Scientific Director), IRCCS Mondino
Foundation (Pavia)*

CONFERENCE

SESSION I

Chairpersons: Renato Borgatti, Stefania Corti - IRCCS Mondino Foundation (Pavia) and University of Pavia;
IRCCS Ca 'Granda Foundation, Polyclinic Hospital (Milan)

11.20 Why is so difficult to design trials in childhood rare diseases?

Eugenio M. Mercuri - Department of Women's and Child Health and Public Health Sciences, IRCCS Gemelli University Hospital, Catholic University (Rome)

12.00 New technologies in the diagnosis and treatment of inherited neurological disorders

Enza Maria Valente - Neurogenetic Research Center, IRCCS Mondino Foundation (Pavia) and University of Pavia

12.40 Discussion

13.00 Lunch

SESSION II

Chairpersons: Stefano Cappa, Barbara Garavaglia - IRCCS Mondino Foundation (Pavia) and IUSS (Pavia);
Genetics of Movement Disorders and Energy Metabolism Disorders, IRCCS Carlo Besta Neurological Institute Foundation (Milan)

14.00 Genetic Frontotemporal Dementia: from pathogenic mechanisms to disease modifying drugs

Daniela Galimberti, University of Milan and IRCCS Ca 'Granda Foundation, Polyclinic Hospital (Milan)

14.40 Strategies for gene therapy in Amyotrophic Lateral Sclerosis (ALS)

Vincenzo Silani - Department of Neurology and Neuroscience Laboratory, IRCCS Auxologico Italian Institute (Milan) and University of Milan

15.20 Gene therapy for inborn errors of metabolism

Nicola Brunetti-Pierri, Department of Translational Medical Sciences, University of Naples Federico II

16.00 Similarities and differences between genetic and pharmacological models of Parkinson's disease: pathophysiological implications

Antonio Pisani - Research Centre of Movement Disorders, IRCCS Mondino Foundation (Pavia) and University of Pavia

16.40 Discussion

17.00 Concluding Remarks

Scientific Supervisor: Roberto Bergamaschi, Scientific Director IRCCS Mondino Foundation (Pavia)

Scientific Committee: Angela Berardinelli*, Alfredo Costa[°], Luca Diamanti*, Claudio Pacchetti*, Cristina Tassorelli[°] (*IRCCS Mondino Foundation - Pavia, [°]University of Pavia)

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