Scientific conference
The event will take place in hybrid form

NOVEMBER
18th 2022

Lessons learned and further road to the therapies of genetic neurological disorders. What we have, unmet needs and future perspectives

XXXII OTTORINO ROSSI AWARD

IRCCS Mondino Foundation
Pavia, via Mondino 2, *Berlucchi Hall*
entrance from via Magenes

www.mondino.it

FONDAZIONE MONDINO
Istituto Neurologico Nazionale a Carattere Scientifico | IRCCS
Sistema Sanitario Regione Lombardia

UNIVERSITÀ DI PAVIA

Ottorino Rossi was born on 17th January, 1877, in Solbiate Comasco, near Como, Italy. In 1895 he enrolled at the medical faculty of the University of Pavia as a student of the Ghislieri College and during his undergraduate years was an intern pupil of the Institute of General Pathology and Histology, headed by Camillo Golgi. In 1901 Rossi obtained his medical doctor degree with the highest grades and a distinction. In October 1902 he went on to the Clinica Neuropatologica (Hospital for Nervous and Mental Diseases) directed by Casimiro Mondino to continue his studies. At the same time, he continued to frequent the Golgi Institute which was the leading Italian centre for biological research. Having completed his clinical preparation in Florence under Eugenio Tanzi, and in Munich at the Institute directed by Emil Kraepelin, he taught at the Universities of Siena, Sassari and, from 1925, Pavia. In Pavia he was made Rector of the University (serving from 1925 to 1936), and during his tenure he was instrumental in getting the buildings of the new San Matteo General Hospital completed.

Ottorino Rossi made many important scientific contributions to the fields of neurology, neurophysiopathology and neuroanatomy. These include: the identification of glucose as the reducing agent of cerebrospinal fluid, the demonstration that fibres from the spinal ganglia pass into the dorsal branch of the spinal roots, and the description of the cerebellar symptom which he termed "the primary asymmetries of positions". Moreover, he conducted important studies on the immunopathology of the nervous system, the serodiagnosis of neurosyphilis and the regeneration of the nervous system. He was the author of major scientific works including an extensive investigation of arteriosclerosis in the brain, L'Arteriosclerosi dei Centri Cerebrali e Spinali (1906), which dealt with the development of lesions of vascular origin. He died in 1936 at the age of 59, having named the Ghislieri College as his heir. Ottorino Rossi was one of Camillo Golgi's most illustrious pupils as well as one of the most eminent descendants of Pavia's medico-biological tradition.

Since 1990, thanks to an initiative of the then new Scientific Director (prof. Giuseppe Nappi), the IRCCS Mondino Foundation has held an annual Ottorino Rossi Award Conference at which the award is presented to a scientist who has made an important contribution to research in the field of the neurosciences.

In the course of its 30-year history, the Ottorino Rossi Award has, on two occasions, been theme based. In the period 2010-2012, it was was devoted to *The Founders of Neurology*, namely the three founders of the most important Italian Schools of Neurology of the twentieth century, while the awards assigned from 2017 to 2019 celebrated the *Pavia Legacy*. This latter series stemmed from the desire to recognise eminent researchers with strong scientific and cultural links with the city of Pavia.

Unfortunately, due to the restrictions imposed by the Covid-19 pandemic, it was not possible to stage the Ottorino Rossi Award Conference in 2020, but the tradition was resumed the following year. This year, 2022, brings the 32nd edition of the Award.

Previous Winners / Ottorino Rossi Award

1990
Vittorio Erspamer
Rome (Italy)

1991
Paolo Pinelli
Milan (Italy)

1992
Giovanni Di Chiro
Bethesda (USA)

1993
Clarence Joseph Gibbs
Bethesda (USA)

1994
David Zee
Baltimore (USA)

1995
Elio Lugaresi
Bologna (Italia)

1996
Michel Fardeau
Paris (France)

1997
Salvador Moncada
London (UK)

1998
Alain Berthoz
Paris (France)

1999
Ottar Sjaastad
Trondheim (Norway)

2000
John Timothy Greenamyre
Atlanta (USA)

2001
Salvatore Di Mauro
New York (USA)

2002
Elio Raviola
Boston (USA)

2003
Michael Welch
Chicago (USA)

2004
François Boller
Paris (France)

2005
Jes Olesen
Copenhagen (Denmark)

2006
Stanley Finger
S. Louis (USA)

2007
Michael A. Moskowitz
Boston (USA)

2008
Patricia Smith Churchland
San Diego (USA)

2009
Stephen P. Hunt
London (UK)

2010
Vincenzo Bonavita
Naples (Italy)

2011
Cesare Fieschi
Rome (Italy)

2012
Giorgio Bernardi
Rome (Italy)

2013
Henry Markram
Lausanne (Switzerland)

2014
Emmanuele A. Jannini
L'Aquila (Italy)

2015
Roberto Crea
Hayward (USA)

2016
Richard Stanislaus Joseph Frackowiak
Lausanne (Switzerland)

2017
Pierluigi Nicotera
Bonn (Germany)

2018
Gianvito Martino
Milan (Italy)

2019
Adriano Aguzzi
Zurich (Switzerland)

2021
Rigmor Højland Jensen
Copenhagen (Denmark)

Francesco Muntoni was born in Cagliari in 1959. He studied in Italy, graduating in medicine at the University of Cagliari in 1984, and specialising in child neurology and psychiatry at the University of Sassari in 1989. He started his medical career in the Department of Child Neuropsychiatry at the University Hospital of Cagliari.

He has worked in the UK since 1993, where he is currently Head of the Dubowitz Neuromuscular Centre at UCL Institute of Child Health and Great Ormond Street Hospital for Children.

His London career began in 1993, and over the first few years saw him working initially as a lecturer, and then senior lecturer in paediatric neurology at the Royal Postgraduate Medical School, Hammersmith Hospital, and then as a reader and honorary consultant in paediatric neurology at Imperial College London, Hammersmith Hospital. In 1996, he was made Clinical and Research Director at the Hammersmith Hospital Neuromuscular Centre, linked to the hospital's Department of Paediatrics and Neonatal Medicine, and in 1998, he was appointed Professor of Paediatric Neurology at Imperial College London. In 2001, he became head of the national referral centre for congenital muscular dystrophy at Hammersmith Hospital.

He was head of the Developmental Neuroscience Programme at Hammersmith Hospital from 2008 to 2018, where he is still Theme Lead in Novel Therapies at the Biomedical Research Centre. He is also Co-Director of Medical Research at the MRC Translational Research Centre at UCL.

In the clinical and research sphere, Professor Muntoni has always focused mainly on novel gene identification, deep phenotyping, and translational research, especially in the area of Duchenne muscular dystrophy (DMD), congenital muscular dystrophy, and spinal muscular atrophy (SMA), although his interest extends to all developmental neuromuscular diseases. He had conducted and continues to conduct numerous natural history studies, has designed multiple clinical trials aimed at the development of therapies for neuromuscular diseases, and has thus contributed significantly to the revolution in the field of SMA therapy that has taken place in recent years, transforming a very serious and fatal disease into a treatable condition. His collaborations with colleagues in the UK, Europe, USA

and Australia have made it possible to identify over 30 genes responsible for neuromuscular diseases. Overall, Professor Muntoni's work has been shaped by his strong interest in clinical aspects, which are both the starting point and the ultimate target of his pathogenetic, molecular and deep phenotyping studies, but he never lost sight of how the evolution of scientific knowledge can impact patients and their expectations.

Professor Muntoni is currently participating in 17 funded studies (as PI in 12 of them and Co-PI in another two), mainly focusing on clinical aspects, the study of biomarkers, and the development of therapies for DMD and SMA. They include, in particular, the over six-million-pound BIND (Brain Involvement in Dystrophinopathies) project, and a 2.4-million-euro project focusing on multisystemic aspects of SMA.

He also sits on the editorial boards of various journals devoted to neuropaediatrics and neuromuscular disorders.

Francesco Muntoni's incessant research activity has resulted in over 600 peer-reviewed publications and his work has a very high impact (he has an H-index of 127). Professor Muntoni is a member of numerous scientific societies including the European Paediatric Neurological Society (EPNS) and the World Muscle Society (WMS), as well as many professional bodies, and since 1996 has held prestigious institutional roles. Between 1994 and 2017, he was the recipient of nine scientific awards.

Alongside his scientific work, which includes the supervision of high-calibre researchers engaged in scientific research in the field of neuromuscular disease, he also boasts great clinical expertise. The centre he directs sees more than 2,000 children affected by neuromuscular diseases each year, and is therefore an essential point of reference for many clinicians wishing to specialise in this field, and for researchers interested in investigating pathogenetic aspects of neuromuscular disease.

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

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9.40 Presentation of the Winner
Angela Berardinelli (Pavia)

Lecture by the Winner
Lesson learned from novel therapies for childhood neuromuscolar disorders
10.00 **Francesco Muntoni** (UCL, London)

11.00 Award ceremony
Francesco Svelto (Pavia)
Roberto Bergamaschi (Pavia)

CONFERENCE

SESSION I
Chairpersons:
Renato Borgatti (Pavia)
Stefania Corti (Milan)

11.20 *Why is so difficult to design trials in childhood rare diseases?*
Eugenio M. Mercuri (Rome)

12.00 *New technologies in the diagnosis and treatment of inherited neurological disorders*
Enza Maria Valente (Pavia)

12.40 *Discussion*

13.00 *Lunch*

SESSION II

Chairpersons:
Stefano Cappa (Pavia)
Barbara Garavaglia (Milan)

14.00 *Genetic Frontotemporal Dementia: from pathogenic mechanisms to disease modifying drugs*
Daniela Galimberti (Milan)

14.40 *Strategies for gene therapy in Amyotrophic Lateral Sclerosis (ALS)*
Vincenzo Silani (Milan)

15.20 *Gene therapy for inborn errors of metabolism*
Nicola Brunetti-Pierri (Naples)

16.00 *Similarities and differences between genetic and pharmacological models of Parkinson's disease: pathophysiological implications*
Antonio Pisani (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 (Pavia)

Speakers and Chairpersons

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Research Unit of Neuromuscular Diseases of Childhood and Adolescence, IRCCS Mondino Foundation

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Accreditamento ECM-CPD

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Sono stati preassegnati n. 6 crediti ECM-CPD per le seguenti figure professionali:

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- Assistente Sanitario
- Biologo
- Chimico
- Farmacista (farmacia ospedaliera)
- Infermiere
- Infermiere pediatrico
- Psicologo (Psicologia, Psicoterapia)
- Tecnico Sanitario di Laboratorio Biomedico

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L'iscrizione, gratuita, è obbligatoria al seguente link: **http://corsi.mondino.it/corsi_list.php**

Other information

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Registration Participants, Speakers and Chairpersons
Entrance from Via Magenes

Under the patronage of

AIM Associazione Italiana Miologia – Italian Association of Myology

SINPIA Società Italiana di Neuropsichiatria dell'Infanzia e dell'Adolescenza

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