SCIENTIFIC PROGRAMME

(Sala 1000)

Monday, 09th March 2015

10.00 – 14.00 *Registration and poster setting up*

14.00 – 14.30 **Welcome and Opening address**
Lucia Monaco (Fondazione Telethon)

14.30 – 15.00 **OPENING LECTURE**
Role of the advocacy organization in rare disease research
Sharon Terry (Genetic Alliance, Washington DC, USA)

15.00 – 17.00 **PLENARY SESSION 1** – **Motorneuron diseases: from molecular targets to trial design**
Chairpersons: Francesco Muntoni, Maria Pennuto
Pathogenesis and therapeutic window in chromosome 5 spinal muscular atrophy
Francesco Muntoni (UCL, London, UK)
From standards of care to natural history: recipe for trial readiness
Eugenio Mercuri (Università Cattolica del Sacro Cuore, Rome)
SMN2 splicing regulation: from disease-linked mechanism to therapeutic opportunity in spinal muscular atrophy
Claudio Sette (University of Rome Tor Vergata, Fondazione Santa Lucia, Rome, Italy)
Skeletal muscle degeneration in spinal and bulbar muscular atrophy
Maria Pennuto (DTI, Università degli Studi di Trento, Trento)
The protein quality control system in motor neuron degeneration in spinal and bulbar muscular atrophy
Angelo Poletti (Università degli Studi di Milano, Milan)

17.00 – 17.30 *Coffee break*

17.30 – 19.00 **POSTER SESSION 1**

19.00 – 20.00 ***SCIENCE AND SOCIETY*** **- Animal experimentation**
Moderator: Anna Maria Zaccheddu (Fondazione Telethon)
Speakers: Elena Cattaneo (Università degli Studi di Milano and Italian senator for life)
Giuliano Grignaschi (IRCCS Mario Negri Institute, Milan)
Kirk Leech (European Animal Research Association, London, UK)

20.00 – 21.00 *Welcome cocktail*

Tuesday, 10th March 2015

08.30 – 09.00 *Registration and poster setting up*

09.00 – 10.00 **ROUND TABLE – From the lab to the clinic, yet remaining a basic scientist**

Moderators: Lana Skirboll, Lucia Faccio

Speakers: Lana Skirboll (Sanofi, Washington DC, USA)
Lucia Faccio (Fondazione Telethon)
Andrea Ballabio (TIGEM, Naples)
Luigi Naldini (OSR-TIGET, Milan)

10.00 – 11.00 **PLENARY SESSION 2 – Updates on the Telethon clinical trials**

Chairperson: Lana Skirboll

Gene therapy of Mucopolisaccharidosis VI

Alberto Auricchio (TIGEM, Naples)

Hematopoietic stem cell gene therapy for inherited disorders: 15 years of experience at TIGET

Alessandro Aiuti (OSR-TIGET, Milan)

Gene therapy for beta thalassemia: towards clinical application

Giuliana Ferrari and Fabio Ciceri (OSR-TIGET, Milan)

11.00 – 11.30 *Coffee break*

11.30 – 13.00 **POSTER SESSION 2**

13.00 – 14.00 *Buffet Lunch*

14.00 – 16.30 **PLENARY SESSION 3 – Hereditary ataxias: convergences and divergences**

Chairpersons: Laura Ranum, Enrico Bertini, Joel Gottesfeld

Introduction on hereditary ataxias

Enrico Bertini (Ospedale Pediatrico Bambino Gesù, Rome)

Domain-specific regulation of cerebellar neurogenesis by Zfp423, a gene implicated in Joubert Syndrome and cerebellar vermis hypoplasia

Gian Giacomo Consalez (Università Vita-Salute San Raffaele, San Raffaele Scientific Institute, Milan)

Molecular determinants of Marinesco-Sjogren syndrome: cell stress, and alterations of proteostasis

Michele Sallese (Fondazione Mario Negri Sud, Santa Maria Imbaro, Chieti)

Epigenetic therapy for Friedreich's ataxia

Joel Gottesfeld (The Scripps Research Institute, La Jolla, USA)

Preventing Frataxin degradation as a therapeutic strategy for Friedreich's ataxia

Roberto Testi (Università degli Studi di Roma Tor Vergata, Rome)

Repeat associated non-ATG (RAN) translation in spinocerebellar ataxia type 8

Laura Ranum (University of Florida, Gainesville, USA)

Genetic and pharmacological rescues of spinocerebellar ataxia in the SCA28 model open to human therapy

Giorgio Casari (Università Vita-Salute San Raffaele, San Raffaele Scientific Institute, Milan)

16.30 – 17.00 *Coffee break*

17.00 – 18.30 **POSTER SESSION 3**

18.30 – 19.30 ***SCIENCE POLICIES - Research integrity***

Michael Caplan (Yale School of Medicine, New Haven, USA)

Wednesday, 11th March 2015

09.00 – 11.00 **PLENARY SESSION 4 - The central role of the skeleton in human health**

Chairpersons: Anna Maria Teti, Marta Serafini

Can we treat systemic autosomal dominant diseases? An example from osteopetrosis

Anna Maria Teti (Università degli Studi dell'Aquila, L'Aquila)

Neonatal bone marrow transplantation for mucopolysaccharidosis type I: the earlier the better

Marta Serafini (DTI, San Gerardo Hospital/Fondazione MBBM, Università di Milano-Bicocca, Monza)

Congenital malformations of the limbs reveal complex genetic interactions between the p63 and Dlx5 genes: towards identification of exploitable targets

Luisa Guerrini (Università degli Studi di Milano, Milan)

In vivo models of chondrodysplasias caused by defects in proteoglycan biosynthesis: phenotyping and pharmacological approaches

Antonio Rossi (Università degli Studi di Pavia, Pavia)

New roles of cellular catabolism during skeletal development

Carmine Settembre (DTI, TIGEM, Naples)

11.00 – 11.30 *Coffee break*

11.30 – 13.00 **PLENARY SESSION 5 - Neurological disorders and DNA damage accumulation**

Chairpersons: Fabrizio d'Adda di Fagagna, Domenico Delia

DNA damage response activation in the brain: causes and consequences

Fabrizio d'Adda di Fagagna (IFOM the FIRC institute of Molecular Oncology, Milan, CNR Pavia)

Dissecting the neurogenic role of CSB protein and its implication in Cockayne syndrome

Luca Proietti de Santis (Università degli Studi della Tuscia, Viterbo)

Links between DNA repair and chromosome structural pathways revealed by cellular models of the Warsaw breakage syndrome cohesinopathy disorder

Takuya Abe (IFOM, the FIRC institute of Molecular Oncology, Milan)

Mechanisms of neurodegeneration in ATM-deficient ataxia-telangiectasia

Domenico Delia (Fondazione IRCCS Istituto Nazionale Tumori, Milan)

13.00 – 13.30 **LATE BREAKING NEWS**

The Intellectual disability protein RAB39B regulates selectively GluA2 trafficking determining synaptic AMPAR composition

Patrizia D'Adamo (DTI, San Raffaele Scientific Institute, Milan)

***COQ4* mutations cause a broad spectrum of mitochondrial disorders associated with CoQ10 deficiency**

Daniele Ghezzi (IRCCS Foundation of the Carlo Besta Neurological Institute, Milan)

Liver-directed lentiviral gene therapy in a dog model of hemophilia B

Alessio Cantore (OSR-TIGET, Milan)

13.30 – 14.00 **POSTER PRIZE AWARD AND CLOSING REMARKS**