Monday, 13th March 2017

10.00 – 14.00 *Registration and poster setting up*

14.00 – 14.30 **Welcome and Opening address**
Lucia Monaco, Fondazione Telethon, Milan

14.30 – 15.00 **OPENING LECTURE** *(Talk 1)*
Yann Le Cam, EURORDIS, France

15.00 – 17.00 **PLENARY SESSION - Neurodevelopmental disorders: from molecular mechanisms to therapeutic inroads**
Chairpersons: Enrico Cherubini, EBRI Rome, and Claudia Bagni, Tor Vergata University Rome - University of Lausanne

Synaptic dysfunctions leading to intellectual disabilities in Autism Spectrum Disorders
Enrico Cherubini, European Brain Research Institute (EBRI), Rome *(Talk 2)*

Fragile X Syndrome and Autism: the molecular mechanisms underlying brain plasticity and therapy *(Talk 3)*
Claudia Bagni, Tor Vergata University, Rome and University of Lausanne, Switzerland

Integrating 2D and 3D patient-specific models for the molecular elucidation of Williams and 7q11.23 microduplication syndromes *(Talk 4)*
Giuseppe Testa, Istituto Europeo di Oncologia, Università degli Studi di Milano, Milan

Protein substitution therapy: an innovative approach to treat CDKL5 disorder
Elisabetta Ciani, University of Bologna, Bologna *(Talk 5)*

Drug repurposing in neurodevelopmental disorders as a faster track from mouse models to clinical trials: the case of Down syndrome *(Talk 6)*
Laura Cancedda, Dulbecco Telethon Institute, Istituto Italiano di Tecnologia, Genoa

17.00 – 17.30 *Coffee break*

17.30 – 20.00 **POSTER SESSIONS - 1 & 2**

20.00 – 21.00 *Welcome buffet*

Tuesday, 14th March 2017

08.30 – 09.00 *Registration and poster setting up*

09.00 – 11.00 **PLENARY SESSION - Share for Rare**
Clinical data, biological samples and research results to fight genetic diseases
Chair: William A. Gahl, National Human Genome Research Institute (NHGRI), NIH, Bethesda, USA

09.00 – 09.30 **The NIH Undiagnosed Diseases Program, Network, and Network International**
William A. Gahl, NHGRI, NIH, Bethesda, USA *(Talk 7)*

09.30 – 10.00 **Telethon Undiagnosed Disease Program**
Vincenzo Nigro, Tigem, Pozzuoli (Naples) *(Talk 8)*

10.00 – 10.20 **Telethon Network of Genetic Biobanks: sharing of human biological material for research**
Mirella Filocamo, Istituto Giannina Gaslini, Genoa *(Talk 9)*

10.20 – 10.40 **Cross-Cutting bottlenecks and solutions in rare diseases research**
Hanns Lochmüller, Newcastle University, Newcastle upon Tyne, UK *(Talk 10)*

10:40 – 11:00 **Discussion**

11.00 – 11.30 *Coffee break*

11.30 – 12.30 **ROUND TABLE - Ethical, legal and social implications (ELSI) in resource sharing**

Moderators: Yann Le Cam, EURORDIS, and Domenica Taruscio, Istituto Superiore di Sanità

11:30 – 11:45 **Practical implications for researchers under the new EU "General Data Protection Regulation"**
Marta Tomasi, University of Trento, Trento *(Talk 11)*

11:45 – 12:30 **Discussion**
Yann Le Cam, Domenica Taruscio, Marta Tomasi, William A. Gahl, Vincenzo Nigro, Mirella Filocamo, Hanns Lochmüller, Sharon Terry

12.30 – 13.30 *Buffet lunch*

13.30 – 15.00 **PLENARY SESSION - Fondazione Telethon and clinical trials**

Chair: Luigi Naldini, SR-Tiget, San Raffaele Scientific Institute and Vita-Salute San Raffaele University, Milan

Gene therapy clinical trial for mucopolysaccharidosis type VI
Nicola Brunetti Pierri, Tigem, Pozzuoli (Naples) *(Talk 12)*

Hematopoietic stem cell gene therapy for inborn errors: from clinical studies to approved drugs
Alessandro Aiuti, SR-Tiget, San Raffaele Scientific Institute and Vita-Salute San Raffaele University, Milan *(Talk 13)*

Gene therapy for beta-thalassemia: initial results from TIGET BTHAL clinical trial
Giuliana Ferrari, SR-TIGET, Scientific Institute San Raffaele and Vita-Salute San Raffaele University, Milan *(Talk 14)*

15.00 – 16.30 **PLENARY SESSION - Preclinical approaches to correct neurological defects**
Chair: Joel Gottesfeld - The Scripps Research Institute, La Jolla, USA

CNS therapy for Lysosomal Storage Disorders
Alessandro Fraldi, Tigem, Pozzuoli (Naples) *(Talk 15)*

Lysosomal Storage Disorders: modeling the disease complexity to refine gene and cell therapy treatment strategies
Angela Gritti, SR-Tiget, San Raffaele Scientific Institute, Milan *(Talk 16)*

GLUT1 deficiency syndrome: biochemical basis of the neurologic defect and possible therapeutic approaches in preclinical models
Maurizio Crestani, University of Milan, Milan *(Talk 17)*

Novel therapeutic strategies for hereditary Cerebral Cavernous Malformations
Elisabetta Dejana, Fondazione Istituto Firc di Oncologia Molecolare, Milan *(Talk 18)*

16.30 – 17.00 *Coffee break*

17.00 – 18.30 **POSTER SESSION - 3**

18.30 – 19.30 ***SCIENCE AND MEDIA: DISSEMINATION OF RESEARCH*** *(Talk 19)*

Guglielmo Lorenzo, Content Manager of Fondazione Telethon will give an introduction, after which Annamaria Zaccheddu, Fondazione Telethon, will lead the discussion with two science communication professionals.

Wednesday, 15th March 2017

09.00 – 11.00 **PARALLEL SESSIONS**

A. From molecular insights to development of therapeutic approaches in amyloidosis
Chairpersons: Giampaolo Merlini, Policlinico San Matteo - University of Pavia, and
Serena Carra, Università degli Studi di Modena e Reggio Emilia, Modena

Unfolding amyloid diseases: challenges and advances *(Talk 20)*
Giampaolo Merlini, Foundation IRCCS Policlinico San Matteo, University of Pavia, Pavia

Transthyretin related amyloidosis: toward a proper therapy based on the right target
Vittorio Bellotti, University of Pavia, Pavia, University College London, London *(Talk 21)*

From protein structure to novel therapeutics against gelsolin amyloidosis
Matteo de Rosa, CNR, Milan *(Talk 22)*

Defining common pathogenic mechanisms elicited by amyloids in the central nervous system
Emiliano Biasini, Dulbecco Telethon Institute (DTI), Center for Integrative Biology (CIBIO), University of Trento, Italy *(Talk 23)*

Molecular chaperones and protein aggregation: from cellular function to disease
Serena Carra, Università degli Studi di Modena e Reggio Emilia, Modena *(Talk 24)*

B. Translational research in muscle diseases: From biology to bedside
Chairpersons: Petra Kaufman, National Institutes of Health, Bethesda USA, and
Irene Bozzoni, Sapienza University of Rome, Rome

RNA-based studies of Duchenne Muscular Dystrophy: post-transcriptional control and role of non-coding RNAs in normal and dystrophic muscle development
Irene Bozzoni, Sapienza University of Rome, Rome *(Talk 25)*

Transcriptional regulation of muscle metabolism in response to physical exercise
Andrea Ballabio, Tigem, Pozzuoli (Naples) *(Talk 26)*

Framing clinical and pathological variability of muscle disorders and genetic heterogeneity: nosography, registries and molecular targets
Giacomo Comi, Università degli Studi di Milano, Fondazione I.R.C.C.S. Ca' Granda Ospedale Maggiore Policlinico, Milan *(Talk 27)*

Setting the stage for successful trials in muscle disease
Petra Kaufmann, Petra Kaufmann, National Center for Advancing Translational Sciences, National Institutes of Health, Bethesda (USA) *(Talk 28)*

11.00 – 11.30 *Coffee break*

11.30 – 13.00 **PARALLEL WORKSHOPS**

A. Data Management *(Talk 29)*

11.30 – 12.30 **Introduction to *-omics* data management in life sciences**
Diego Di Bernardo, Tigem, Pozzuoli (Naples)

Transcriptomics data:

How to submit transcriptomic data to the Gene Expression Omnibus database
Annamaria Carissimo, Tigem, Pozzuoli (Naples)

How to access and analyse transcriptomics data via interactive web tools: GEO2R and GEO Profiles
Rossella De Cegli, Tigem, Pozzuoli (Naples)

Genomics data:

Online repositories of sequencing data: The European Nucleotide Archive and the European Genome-Phenome Archive
Margherita Mutarelli, Tigem, Pozzuoli (Naples)

12.30 – 13.00 **The Telethon-CINECA Genomics Repository**
Mattia D'Antonio, Cineca, Rome

B. Clinical Development and enabling Regulatory Steps *(Talk 30)*

Filling the gap: what I need to know, plan and do to enter into the clinics
Stefano Zancan, Fondazione Telethon, Milan

How to obtain ODD and Scientific Advice at EMA
Michela Gabaldo, Fondazione Telethon, Milan

13.00 – 13.30 **LATE BREAKING NEWS**

Original research article
Citron Kinase deficiency leads to chromosomal instability and TP53-sensitive microcephaly. Cell Reports, February 14, 2017
Federico Bianchi, University of Turin

Consensus Report
Human genome editing: Science, ethics, and governance.
National Academy of Sciences and National Academy of Medicine. nationalacademies.org/gene-editing

Luigi Naldini, San Raffaele Telethon Institute for Gene Therapy (SR-TIGET), Vita Salute San Raffaele University, Milan

13.30 **CLOSING REMARKS**