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
Claudia Bagni, Tor Vergata University, Rome and University of Lausanne, Switzerland  (Talk 3)

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

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
Laura Canciedda, Dulbecco Telethon Institute, Istituto Italiano di Tecnologia, Genoa  (Talk 6)

17.00 – 17.30  Coffee break

17.30 – 20.00  POSTER SESSIONS - 1 & 2

20.00 – 21.30  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)
Introduction by Guglielmo Lorenzo, Content Manager of Fondazione Telethon
Round table with Roberta Villa (Corriere Salute, Scienzainrete) and Michela Vuga (circuito Cnr radio, OK salute). Moderator: Annamaria Zaccheddu, Fondazione Telethon.

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 (Talk 21)
Vittorio Bellotti, University of Pavia, Pavia, University College London, London

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

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

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

B. Translational research in muscle diseases: from biology to bedside
Chairpersons: Petra Kaufmann, 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 (Talk 25)
Irene Bozzoni, Sapienza University of Rome, Rome

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

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

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

11.00 – 11.30 Coffee break
11.30 – 13.00  PARALLEL WORKSHOPS

A. Data Management  

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

13.00 – 13.30  LATE BREAKING NEWS

Original research articles
Federico Bianchi, University of Turin

Maurizio Mete, Sacro Cuore Hospital-Don Calabria, Negrar (Verona)

Consensus Report
Human genome editing: Science, ethics, and governance (2017).  
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