

XIX Scientific Convention

PALAZZO DEI CONGRESSI - RIVA DEL GARDA (TN)

13-15 MARCH 2017

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** *(Talk 5)*
Elisabetta Ciani, University of Bologna, Bologna
- 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 – 21.30 *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, Scienzairete) 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

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 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

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, 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 articles*
- Citron Kinase deficiency leads to chromosomal instability and TP53-sensitive microcephaly.** Cell Reports, February 14, 2017.
Federico Bianchi, University of Turin
- A fully organic retinal prosthesis restores vision in a rat model of degenerative blindness.** Nature Materials, Mar 6, 2017.
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**