

XXI Scientific Convention

PALAZZO DEI CONGRESSI - RIVA DEL GARDA (TN)
13-15 MARCH 2023



SCIENTIFIC PROGRAM

Monday, March 13th 2023

10.00 – 14.00 *Registration and poster setting up*

14.00 – 15.00 Welcome and Opening by Fondazione Telethon

Luca di Montezemolo, Presidente di Fondazione Telethon

Francesca Pasinelli, Direttore Generale di Fondazione Telethon

15.00 – 17.00 PLENARY SESSION 1

FROM BASIC MECHANISMS TO DISEASE TARGET IDENTIFICATION

The importance of supporting basic research - advancing knowledge in health and disease mechanisms is instrumental to foster innovation

Chairs: Ivana Trapani, Istituto TIGEM, Pozzuoli (NA) e Università “Federico II”, Napoli
Gaia Colasante, Università Vita-Salute San Raffaele, Milano

15.00 – 15.20 The role of SMN protein in translation: implications for Spinal Muscular Atrophy

Gabriella Viero, CNR – Istituto di Biofisica, Trento

15.20 – 15.40 Dissecting the contribution of nuclear mechanics for the treatment of Kabuki Syndrome

Alessio Zippo, Università di Trento

15.40 – 16.00 Amyloid aggregation and lysosomal membrane dynamics in Sanfilippo Disease

Antonio Monaco, CEINGE Biotec. Avanzate e Università degli Studi “Federico II”, Napoli

16.00 – 16.20 Selective autophagy of the Endoplasmic reticulum: mechanisms, regulation and therapeutic potential

Carmine Settembre, Istituto TIGEM, Pozzuoli (NA)

16.20 – 16.40 GABAA-receptor defects in CDKL5 disorder: molecular mechanisms and targeting by synthetic neuroactive steroids

Charlotte Kilstrup-Nielsen, Università dell’Insubria, Busto Arsizio (VA)

16.40 – 17.00 Unraveling PCDH19 neuronal role in a mouse model of DEE9

Silvia Bassani, CNR – Istituto di neuroscienze – Vedano al Lambro (MB)

17.00 – 17.30 *Coffee break*

17.30 – 18.45 POSTER SESSION 1

18.45 – 19.00 *(spostamento in sala Garda per lo spettacolo serale)*

19.00 – 20.00 “LIFE IS MAGIC”

Show di *Christopher Castellini – l'illusionista della mente*

20.15 – 21.30 *Light dinner*

Tuesday, March 14th 2023

08.15 – 09.00 *Registration and poster setting up*

09.00 – 11.00 PLENARY SESSION 2

THE PATH TOWARD PRE-CLINICAL DEVELOPMENT

Moving a step forward in basic research – challenges in pre-clinical development of biomolecules and small molecules

Chairs: Claudia Bagni, Università di Roma Tor Vergata e Università di Losanna
Emiliano Biasini, CIBIO Università di Trento

09.00 – 09.20 **Strategies to raise brain cholesterol in different mouse models of HD produce a complete and prolonged recovery of cognitive and motor deficits**
Elena Cattaneo, Università degli Studi di Milano

09.20 – 09.40 **AAV-mediated inhibition of miR-181a/b as gene-independent therapeutic tool for mitochondrial diseases**
Alessia Indrieri, Istituto TIGEM, Pozzuoli (NA)

09.40 – 10.00 **Finding a cure of Tubular aggregate myopathy**
Armando Genazzani, Università del Piemonte Orientale “A. Avogadro”, Novara

10.00 – 10.20 **The CFTR corrector C17 is effective in sarcoglycanopathies: proof of concept and ADME studies**
Dorianna Sandonà, Università di Padova

10.20 – 10.40 **Protein folding pathways across physiology and therapy**
Emiliano Biasini, Università di Trento

10.40 – 11.00 **Targeting Cl homeostasis in neurodevelopmental disorders with new selective NKCC1 inhibitors**
Savardi Annalisa, Istituto Italiano di Tecnologia, Genova

11.00 – 11.30 *Coffee break*

11.30 – 13.00 POSTER SESSION 2

This poster session will be shared with Patient Organizations: a precious opportunity to connect Researchers and Patients

13.00 – 14.00 *Lunch buffet*

14.00 – 15.30 PLENARY SESSION 3

GENE THERAPY – CHALLENGES AND LIMITATIONS

Gene therapy has been evolving for decades. While many hurdles remain to be overcome, still the spread of new strategies and tools keeps the field vibrant.

Chairs: Ivana Trapani, Istituto TIGEM, Pozzuoli (NA) e TIGEM e Università “Federico II”, Napoli
Renato Ostuni, Istituto SR-TIGET e Università Vita-Salute San Raffaele, Milano

14.00 – 14.20 **Development and application of targeted epigenetic silencing**

Angelo Lombardo, Istituto SR-TIGET, Milano

14.20 – 14.40 **Nuclease-free targeted integration of a promoter-less mini-ATP7B confers proliferative advantage to edited hepatocytes and corrects Wilson disease**

Pasquale Piccolo, Istituto TIGEM, Pozzuoli (NA)

14.40 – 15.00 **Next-generation gene editing in human hematopoietic stem/progenitor cells**

Samuele Ferrari, Istituto SR-TIGET, Milano

15.00 – 15.20 **EXPEDITing AAV gene therapy**

Alberto Auricchio, Istituto TIGEM, Pozzuoli (NA)

15.30 – 17.00 PLENARY SESSION 4

UPDATES FROM THE TELETHON CLINICAL RESEARCH

Overview of the Telethon clinical research and discussion on current status

Chairs: Graciana Diez-Roux, Istituto TIGEM, Pozzuoli (NA)
Anna Villa, Istituto SR-TIGET e IRGB-CNR, Milano

15.30 – 16.00 **Duchenne Muscular Dystrophy: from natural history to innovative experimental therapies**

Eugenio M Mercuri, Università Cattolica del Sacro Cuore e Fondazione Policlinico Universitario “A. Gemelli” IRCCS, Roma

16.00 – 16.30 **SR-TIGET Clinical trial updates**

Alessandro Aiuti, Istituto SR-TIGET, Milano

16.30 – 17.00 **Updates of two AAV-mediated, liver-directed gene therapy clinical trials**

Nicola Brunetti-Pierri, Istituto TIGEM, Pozzuoli (NA)

17.00 – 17.30 *Coffee break*

17.30 – 18:30 POSTER SESSION 3

18:30 – 20:00 SCIENCE & SOCIETY – Round Table

PNRR: un'occasione per la ricerca biomedica italiana?

Alla luce delle scelte fatte sinora in termini di priorità e di modalità di allocazione dei fondi, qual è l'impatto atteso del PNRR sulla ricerca, in particolare sulle malattie genetiche rare?

Wednesday, March 15th 2023

09.00 – 10.40 PLENARY SESSION 5

NEW TECHNOLOGIES

Scientific and technological advances break down barriers and create new frontiers of research in health and disease.

Chairs: Emiliano Biasini, CIBIO Università di Trento
Carmine Settembre, Istituto TIGEM, Pozzuoli (NA) e Università “Federico II”, Napoli

- 09.00 – 09.20 **Fluorescent measurement of PIDDosome activation enables high throughput screening for modulators of centriolar distal appendage functionality**
Luca Fava, Università di Trento
- 09.20 – 09.40 **Cellular and transcriptional dynamics of human neutrophils at steady state and upon stress**
Renato Ostuni, Istituto SR-TIGET, Milano
- 09.40 – 10.00 **Human Hematopoietic Stem/Progenitor cell trafficking and clonal tracking**
Serena Scala, Istituto SR-TIGET, Milano
- 10.00 – 10.20 **A functional genomics framework to investigate the molecular bases of rare genetic diseases**
Davide Cacchiarelli, Istituto TIGEM, Pozzuoli (NA)
- 10.20 – 10.40 **Explainable Artificial Intelligence and fractal dimension of brain MRI in Friedreich ataxia and SCAs**
Stefano Diciotti, Università di Bologna
- 10.40 – 11.10 *Coffee break*
- 11.10 -11.40 *Photo time!*

11.45 – 12.45 PLENARY SESSION 6

IMPROVING THE DIAGNOSIS OF RARE GENETIC DISEASES

Improving genetic diagnosis when it comes to rare diseases and genetic syndromes is still a challenge. A timely and accurate diagnosis improves quality of life of rare patients and allows quicker access to possible treatments and therapies.

Chair: Giorgio Casari, Università Vita-Salute San Raffaele, Milano

- 11.45 – 12.05 **Telethon Undiagnosed Diseases Program (TUDP): a systematic study of 1,098 families with severe undiagnosed pediatric conditions**
Vincenzo Nigro, Istituto TIGEM, Pozzuoli (NA)
- 12.05 – 12.25 **Cryptic or not? Pathogenic intronic variants detected from WES data**
Fulvio D’Abrusco, Università di Pavia

12.25 – 12.45 **Responsible Implementation of Newborns Genome Sequencing (RINGS): a technical and interpretative feasibility study**
Maria Iascone, ASST Papa Giovanni XXIII, Bergamo

12.45 – 13.15 LATE BREAKING NEWS

Authors of freshly published results will have the chance to present their findings

Chairs: Graciana Diez-Roux, Istituto TIGEM, Pozzuoli (NA)
Gaia Colasante, Università Vita-Salute San Raffaele, Milano

13.15 – 13.30 BEST POSTER AWARDS

Prizes will be awarded to the three best posters

Chairs: Graciana Diez-Roux, Istituto TIGEM, Pozzuoli (NA)
Gaia Colasante, Università Vita-Salute San Raffaele, Milano

13.30 – 13.45 CLOSING REMARKS

Fondazione Telethon