

XXI Scientific Convention

PALAZZO DEI CONGRESSI - RIVA DEL GARDA (TN)

13-15 MARCH 2023



PROGRAM

Monday, March 13th 2023

10.00 – 14.00 *Registration and poster setting up* (Pala Vela)

14.00 – 15.00 Welcome and Opening by Fondazione Telethon (Sala Garda)

Francesca Pasinelli, Direttore Generale di Fondazione Telethon ETS

15.00 – 17.00 PLENARY SESSION 1

FROM BASIC MECHANISMS TO DISEASE TARGET IDENTIFICATION

Chairs: Ivana Trapani, Istituto TIGEM, Pozzuoli e Università degli Studi "Federico II", Napoli
Gaia Colasante, Università Vita-Salute San Raffaele, Milano

(Sala Garda)

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 Univ. 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* (Pala Vela)

17.30 – 18.45 POSTER SESSION 1 (Pala Vela)

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

19.00 – 20.00 “LIFE IS MAGIC” *(Sala Garda)*

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

20.15 – 21.30 *Light dinner* *(Pala Vela)*

Tuesday, March 14th 2023

08.15 – 09.00 *Registration and poster setting up* *(Pala Vela)*

09.00 – 11.00 PLENARY SESSION 2

THE PATH TOWARD PRE-CLINICAL DEVELOPMENT

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

(Sala Garda)

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* *(Pala Vela)*

11.30 – 13.00 POSTER SESSION 2

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

(Pala Vela)

13.00 – 14.00 *Lunch buffet* *(Pala Vela)*

14.00 – 15.30 PLENARY SESSION 3

GENE THERAPY – CHALLENGES AND LIMITATIONS

Chairs: Ivana Trapani, Istituto TIGEM, Pozzuoli e TIGEM e Univ. degli Studi “Federico II” Napoli
Renato Ostuni, Istituto SR-TIGET e Università Vita-Salute San Raffaele, Milano

(Sala Garda)

- 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

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

- 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* *(Pala Vela)*

17.30 – 18:30 POSTER SESSION 3 *(Pala Vela)*

18:30 – 20:00 SCIENCE & SOCIETY – Round Table
PNRR: un'occasione per la ricerca biomedica italiana? *(Sala Garda)*

Wednesday, March 15th 2023

09.00 – 10.40 PLENARY SESSION 5
NEW TECHNOLOGIES

Chairs: Emiliano Biasini, CIBIO Università di Trento
Carmine Settembre, Istituto TIGEM, Pozzuoli e Univ. degli Studi "Federico II", Napoli *(Sala Garda)*

- 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*

(Pala Vela)

11.10 -11.40 *Photo time!*

(Outdoor)

11.45 – 12.45 **PLENARY SESSION 6** **IMPROVING THE DIAGNOSIS OF RARE GENETIC DISEASES**

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

(Sala Garda)

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 Iacone, ASST Papa Giovanni XXIII, Bergamo

12.45 – 13.15 **LATE BREAKING NEWS**

(Sala Garda)

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

Antagonistic effect of cyclin-dependent kinases and a calcium-dependent phosphatase on polyglutamine-expanded androgen receptor toxic gain of function

Piol et al, Jan 2023, Sci. Adv.

Maria Pennuto

Antibody gene transfer treatment drastically improves epidermal pathology in a keratitis ichthyosis deafness syndrome model using male mice

Peres et al, Feb 2023, eBioMedicine

Chiara Peres

TFEB and TFE3 drive kidney cystogenesis and tumorigenesis

Di Malta et al, accepted for publication,

Chiara Di Malta

EMBO Mol Med

13.15 – 13.30 **BEST POSTER AWARDS**

(Sala Garda)

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

13.30 – 13.45 **CLOSING REMARKS**

(Sala Garda)

Fondazione Telethon ETS