

# XXI Scientific Convention

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



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## SCIENTIFIC PROGRAM - Preliminary

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Monday, March 13<sup>th</sup> 2023

10.00 – 14.00 *Registration and poster setting up*

**14.00 – 15.00 Welcome and Opening by Fondazione Telethon**

Francesca Pasinelli, Direttore Generale di Fondazione Telethon

**15.00 – 17.00 PLENARY SESSION 1**

**FROM BASIC MECHANISMS TO DISEASE TARGET IDENTIFICATION**

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 Biotecnologie Avanzate e Università degli Studi di Napoli “Federico II”

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**

**19.00 – 20.00 Spettacolo serale - “LIFE IS MAGIC”**

20.15 – 21.30 *Light dinner*

## Tuesday, March 14<sup>th</sup> 2023

08.15 – 09.00 *Registration and poster setting up*

### 09.00 – 11.00 PLENARY SESSION 2

#### THE PATH TOWARD PRE-CLINICAL DEVELOPMENT

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

13.00 – 14.00 *Light Lunch (buffet)*

### 14.00 – 15.30 PLENARY SESSION 3

#### GENE THERAPY – CHALLENGES AND LIMITATIONS

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**

15.30 – 16.00 **TIGEM Clinical trial updates**  
Istituto TIGEM, Pozzuoli (NA)

16.00 – 16.30 **SR-TIGET Clinical trial updates**  
Istituto SR-TIGET, Milano

16.30 – 17.00 **Tbd**

17.00 – 17.30 *Coffee break*

**17.30 – 18:30 POSTER SESSION 3**

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

**Wednesday, March 15<sup>th</sup> 2023**

**09.00 – 10.40 PLENARY SESSION 5 - NEW TECHNOLOGIES**

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**

11.45 – 12.05 **Cryptic or not? Pathogenic intronic variants detected from WES data**  
Fulvio D'Abrusco, Università di Pavia

**12.05 – 12.25 Telethon Undiagnosed Diseases Program (TUDP): a systematic study of 1,098 families with severe undiagnosed pediatric conditions**  
Vincenzo Nigro, Istituto TIGEM, Pozzuoli (NA)

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

**12.45 – 13.15 LATE BREAKING NEWS**

**13.15 – 13.30 BEST POSTER AWARDS**

*Prizes will be awarded to the three best posters*

**13.30 – 13.45 CONCLUDING REMARKS**