Monday, 17th March 2025 - *afternoon*

10 am – 2 p.m.	*Registration and poster setting up*	
2 – 3 p.m.	**Welcome and opening by Fondazione Telethon** *Ilaria Villa - General Manager, Fondazione Telethon ETS* *Celeste Scotti, R&D Director, Fondazione Telethon ETS*	*Anfiteatro*
3 – 4.35 p.m.	Session 1 - **From genome to genes and their expression** *Chairs: Andrea Ditadi, Anna Maria Cariboni* **Plenary talks** **Sestrin2 drives ER-phagy in response to protein misfolding** *Chiara De Leonibus - Telethon Institute of Genetics and Medicine (Pozzuoli)* **Regulation of gene expression at the level of translation: unexpected checkpoints and future perspectives** *Stefano Biffo – University of Milan & INGM (Milan)* **Brain organoids modelling of hormonal impact on neurodevelopment to unravel the molecular basis of Congenital Adrenal Hyperplasia neuropsychiatric symptoms** *Nicolò Caporale - University of Milan, Human Technopole (Milan)* **Flash presentations** • Anna Maria Gambetta, University of Padua - Genome-wide screening in pluripotent cells identifies novel suppressors of mutant huntingtin toxicity • Carlo Giaccari, CNR - IGB (Naples) - Exploring the role of the maternal-effect Padi6 gene in early mouse embryogenesis • Luca Frosio, San Raffaele Telethon Institute for Gene Therapy (Milan) - Mitotic kinase Aurora B control interferon responses in macrophages via histone phosphorylation • Leandro Soria, Telethon Institute of Genetics and Medicine (Pozzuoli) - Impaired nuclear glycogen metabolism affects liver homeostasis in Argininosuccinic aciduria • Martina Zobel, University of Milan - DNA as driver of its own instability within the Huntington's disease locus • Samuele Crotti, University of Modena and Reggio Emilia - Boosting HSPB3 to prevent neuromuscular degeneration in peripheral neuropathies **NEGEDIA – sponsored flash talk** Bridging Advanced Sequencing Technology and Clinical Diagnostics: Unlocking the Next Generation of Medicine *Beatrice Salvatori, Scientist and Project Manager Negedia*	*Anfiteatro*
4.35 – 5.20 p.m.	**Keynote Lecture** *Chair: Leopoldo Staiano* **Applications of human brain organogenesis** *Alysson R. Muotri - UC San Diego, CA*	*Anfiteatro*
5.30 – 7.30 pm	**Coffee break & Poster Session 1** *Scientists and Patient Association Representatives*	*Sala della Piazza*
7.30 pm	***Il Ritmo della Ricerca - cena romagnola e galà*** *Entertainment by the Raoul & Swing Orchestra*	*Foyer Domus*

Tuesday, 18th March 2025 - *morning*

8.30 – 9 am — *Registration and poster setting up*

9 – 10.30 am — Session 2 - **Editing strategies for rare diseases** — *Anfiteatro*

Chairs: Anna Cereseto, Angelo Lombardo

Plenary talks

Towards one-and-done gene therapies for defect of hepatic metabolism in pediatric patients
Alessio Cantore - San Raffaele - Telethon Institute for Gene Therapy (Milan)

Towards customized allele-specific CRISPR/Cas gene editing for the treatment of ocular surface disorder in EEC syndrome
Laura De Rosa - University of Modena and Reggio Emilia

Mutation-independent genome editing approaches for treatment of Stargardt disease
Ivana Trapani - Telethon Institute of Genetics and Medicine (Pozzuoli)

SIAL Srl – sponsored flash talk

Genome of Europe: building an unbiased genome reference across Europe for improving precision medicine
Graziano Pesole, Università degli Studi di Bari "A. Moro"

Flash presentations

- Fanny Jaudon, University of Trieste - New genome editing strategies for episodic ataxia and absence epilepsy: Optimizing splice isoforms for enhanced calcium channel function
- Federico Midena, San Raffaele - Telethon Institute for Gene Therapy (Milan) - Tridimensional scaffolds enable efficient and safe genetic engineering of hematopoietic stem and progenitor cells across multiple gene therapy platforms
- Agnese Padula, Telethon Institute of Genetics and Medicine (Pozzuoli) - Copper-mediated DNA damage influences AAV integration profiles in gene therapy and genome editing for Wilson disease
- Martino Cappelluti, San Raffaele - Telethon Institute for Gene Therapy (Milan) - Allele-selective silencing of the mutant Huntingtin gene by epigenetic editing for the treatment of Huntington's Disease
- Martina Orefice, University of Pisa - Modeling Pitt-Hopkins syndrome and new pathogenetic variants of TCF4 by gene editing: a step forward toward precision medicine

10.30 – 11.15 am — **Coffee Break** — *Sala della Piazza*

11.15 a.m. – 12 pm — **Keynote Lecture** — *Anfiteatro*

Chair: Anna Cereseto

Genome Editing by Homology Directed Repair (HDR) to Create Stem Cell Based Drugs
Matthew Porteus - Stanford University (CA)

12 p.m. – 1 p.m. — **From project to product: how can a scientist trigger pharma interaction?** — *Anfiteatro*

***Round Table** - Chair: Valentina Vavassori*

Alberto Auricchio, Tigem Scientific Director
Emiliano Biasini, Associate Professor at Department of Cellular, Computational and Integrative Biology (CIBIO), University of Trento
Samuele Ferrari, Project Leader at SR-Tiget
Irene Manfredi, ADV IP Srl (former IP Manager at Fondazione Telethon ETS)

1 – 2.20 p.m. — **Lunch buffet** — *Foyer Domus*

Enjoy lunch and see you at 2.30 p.m. in Anfiteatro…sharp!

Tuesday, 18th March 2025 – *afternoon*

2.30 – 3.30 p.m. Session 3 - **Strategies and challenges in biomedical research** *Anfiteatro*

Safety and Efficacy in Gene Therapy: A Balancing Act
Eugenio Montini - San Raffaele - Telethon Institute for Gene Therapy (Milan)

Round table
Celeste Scotti, R&D Director, Fondazione Telethon ETS
Alberto Auricchio, Tigem Scientific Director
Luigi Naldini, SR-Tiget Scientific Director

3.30 – 5.30 p.m. Session 4 - **Mitochondrial biology** *Anfiteatro*

Chair: Giorgio Casari

Plenary talks

A mitochondrial therapy for muscular dystrophies
Paolo Bernardi, University of Padua

Posterior Column Ataxia and Retinitis Pigmentosa: new pathogenetic insights from the study of mitochondria-associated membranes
Deborah Chiabrando, University of Turin

Dysregulation of lipid droplet dynamics induces mitochondrial stress and promotes fibrosis, driving the progression of chronic kidney disease in Lowe syndrome
Leopoldo Staiano, Telethon Institute of Genetics and Medicine (Pozzuoli)

EUROCLONE – sponsored flash talk
Innovative solutions for protein detection in Spatial Biology and in in Single Cell Analysis
Maxime Jacquet, Field Application Scientists EMEA, Cell Signaling Technology

Flash presentations

- Michael Donadon, University of Padua - Exploiting redox-active molecules against mitochondrial diseases linked to complex III and complex I dysfunction
- Iva Cantando, University of Rome "La Sapienza" - Mitochondrial dysfunctions in 22q11 Deletion Syndrome: a novel target for therapeutic intervention
- Lucia Iannotta, CNR - Neuroscience Institute (Milan) - Uncovering VPS13D's function to understand SCAR4 pathogenesis
- Silvia Sighinolfi San Raffaele - Telethon Institute for Gene Therapy (Milan) - Intracellular Iron Overload rewires HSC metabolism by impairing mitochondrial fitness in in β-thalassemia
- Erica Tagliatti, Humanitas University (Milan) - Microglial NHD Trem2 gene controls neuronal metabolism and synapses during development

5.30 – 7.30 p.m. **Coffee break & Poster Session 2** *Sala della Piazza*

Wednesday, 19th March 2025 – *morning*

9 – 10.30 a.m. Session 5 - **RNA technologies for therapy development** *Anfiteatro*

Chairs: Marco Sandri, Laura Cancedda

Plenary talks

A new RNA-based therapy for the Fragile X Syndrome
Claudia Bagni, University of Rome "Tor Vergata"

mRNA-replacement therapy for Glycogen Storage Disease type 1b
Lucia De Stefano, Telethon Institute of Genetic and Medicine (Pozzuoli)

Developing a strategy to target AR coactivators in preclinical models of SBMA
Manuela Basso, University of Trento

PRODOTTI GIANNI – sponsored flash talk

New Generation Drug Delivery Systems: intracellular uptake and trafficking studies
Barbara Canonico, Professor at University of Urbino Carlo Bo

Flash presentations

- Alessia Oppezzo, IFOM ETS (Milan) - Selective control of DNA damage response at telomeres as an innovative therapeutic approach for Dyskeratosis Congenita
- Roberta Benfante, National Research Council (Milan) - The natural antisense lncRNA PHOX2B-AS1 in the pathogenesis and as potential drug target in Congenital Central Hypoventilation Syndrome (CCHS)
- Roberto Oleari, University of Milan - Combining spatial and gene transcriptomics to reveal NKTR as nuclear splicing protein implicated in head development and in a novel spliceosomopathy
- Dalila Capasso, Telethon Institute of Genetic and Medicine (Pozzuoli) - Gaining insight into the role of noncoding RNAs in Inherited Retinal Disease by RNA-seq-based approaches

10.30 – 11.30 a.m. **Coffee break** *Sala della Piazza*

with take-away

11.30 – 12.45 p.m. Session 6 - **Innovative therapeutic approaches** *Anfiteatro*

Chairs: Maria Pennuto, Emiliano Biasini

Plenary talks

A membrane-targeted photoswitch restores physiological ON/OFF responses to light in the degenerate retina
Fabio Benfenati, Italian Institute of Technology (Genova)

A Pipeline for drug discovery in Lowe Syndrome
Antonella De Matteis, Telethon Institute of Genetics and Medicine (Pozzuoli)

Classification and rescue of HCN1 mutations in EIEE patients
Anna Moroni, University of Milan

Flash presentations

- Francesco Gentile, Università Vita-Salute San Raffaele (Milan) - Genetic and pharmacologic modulation of the ATF6 UPR-related pathway affect disease pathogenesis in mice models of CMT1B
- Angela Della Sala, University of Torino - Rescue of Mutant CFTR Chloride Channels by a Mimetic Peptide Targeting the A-Kinase Anchoring Function of PI3Kγ
- Sandro Montefusco, Telethon Institute of Genetics and Medicine (Pozzuoli) - Combining phenotypic high-content imaging with the repurposing of drugs to tackle lysosomal storage disorders
- Enrica Federti, University of Verona - Resolvin D1 protects against sickle cell related inflammatory cardiomyopathy in humanized mice
- Nicole Innocenti, University of Trento - Targeting the undruggable: chemical exploration of a prion protein degrader compound

12.45 – 13.25 p.m. **Late Breaking News** *Anfiteatro*

13.25 – 13.30 p.m. **Closing** *Anfiteatro*