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*	*Anfiteatro*
3 – 4.35 p.m.	Session 1 - **From genome to genes and their expression** *Chairs: Andrea Ditadi, Anna Maria Cariboni*	*Anfiteatro*
3 – 4 pm	**Plenary talks**	
3.00 – 3.20	**Sestrin2 drives ER-phagy in response to protein misfolding** *Chiara De Leonibus - Telethon Institute of Genetics and Medicine (Pozzuoli)*	
3.20 – 3.40	**Regulation of gene expression at the level of translation: unexpected checkpoints and future perspectives** *Stefano Biffo – University of Milan & INGM (Milan)*	
3.40 – 4.00	**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)*	
4.00 – 4.20	**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	
4.20 – 4.35	**NEGEDIA – sponsored flash talk** Bridging Advanced Sequencing Technology and Clinical Diagnostics: Unlocking the Next Generation of Medicine *Beatrice Salvatori, Scientist and Project Manager Negedia*	
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** *Chairs: Anna Cereseto, Angelo Lombardo*	*Anfiteatro*
9 – 10 am	**Plenary talks**	
9.00 – 9.20	**Towards one-and-done gene therapies for defect of hepatic metabolism in pediatric patients** *Alessio Cantore - San Raffaele - Telethon Institute for Gene Therapy (Milan)*	
9.20 – 9.40	**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*	
9.40 – 10.00	**Mutation-independent genome editing approaches for treatment of Stargardt disease** *Ivana Trapani - Telethon Institute of Genetics and Medicine (Pozzuoli)*	
10.00 – 10.15	**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"*	
10.15 – 10.30	**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** *Chair: Anna Cereseto* **Genome Editing by Homology Directed Repair (HDR) to Create Stem Cell Based Drugs** *Matthew Porteus - Stanford University (CA)*	*Anfiteatro*
12 p.m. – 1 p.m.	**From project to product: how can a scientist trigger pharma interaction?** *Round Table - Chair: Valentina Vavassori* *Irene Manfredi, ADV IP Srl* *Alberto Auricchio, Tigem Scientific Director* *Luigi Naldini, SR-Tiget Scientific Director* *Samuele Ferrari, Project Leader at SR-Tiget*	*Anfiteatro*
1 – 2.20 p.m.	**Lunch buffet** *Enjoy lunch and see you at 2.30 p.m. in Anfiteatro…sharp!*	*Foyer Domus*

Tuesday, 18th March 2025 – *afternoon*

2.30 – 3.30 p.m. Session 3 - **Fondazione Telethon** *Anfiteatro*

Strategies and challenges in biomedical research for rare diseases

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

Chair: Giorgio Casari

3.30 – 4.40 **Plenary talks**

3.30 – 4.00 **A mitochondrial therapy for muscular dystrophies**
Paolo Bernardi, University of Padua

4.00 – 4.20 **Posterior Column Ataxia and Retinitis Pigmentosa: new pathogenetic insights from the study of mitochondria-associated membranes**
Deborah Chiabrando, University of Turin

4.20 – 4.40 **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)

4.45 – 5.00 **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

5.00 – 5.15 **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

9.00 – 10.00 **Plenary talks**

9.00 – 9.20 **A new RNA-based therapy for the Fragile X Syndrome**
Claudia Bagni, University of Rome "Tor Vergata"

9.20 – 9.40 **mRNA-replacement therapy for Glycogen Storage Disease type 1b**
Lucia De Stefano, Telethon Institute of Genetic and Medicine (Pozzuoli)

9.40 – 10.00 **Developing a strategy to target AR coactivators in preclinical models of SBMA**
Manuela Basso, University of Trento

10.00 – 10.15 **PRODOTTI GIANNI – sponsored flash talk**
New Generation Drug Delivery Systems: intracellular uptake and trafficking studies
Barbara Canonico, Professor at University of Urbino Carlo Bo

10.15 – 10.30 **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, Elena Cattaneo

11.30 – 12.30 **Plenary talks**

11.30 – 11.50 **A membrane-targeted photoswitch restores physiological ON/OFF responses to light in the degenerate retina**
Fabio Benfenati, Italian Institute of Technology (Genova)

11.50 – 12.10 **A Pipeline for drug discovery in Lowe Syndrome**
Antonella De Matteis, Telethon Institute of Genetics and Medicine (Pozzuoli)

12.10 – 12.30 **Classification and rescue of HCN1 mutations in EIEE patients**
Anna Moroni, University of Milan

12.30 – 12.45 **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 – 13.10 p.m. **Late Breaking News** *Anfiteatro*

13.10 – 13.15 p.m. **Closing** *Anfiteatro*