

XX Scientific Convention

PALAZZO DEI CONGRESSI - RIVA DEL GARDA (TN)
28-30 OCTOBER 2019

FONDAZIONE



Monday, October 28th 2019

10.00 – 14.00 *Registration and poster setting up*

14.00 – 15.00 Welcome and Opening by Fondazione Telethon

Francesca Pasinelli, General Manager
Manuela Battaglia, Head of Research

**15.00 – 17.00 PLENARY SESSION 1 - GENETIC MUSCLE DISEASES: FROM KNOWLEDGE ON
PHYSIOPATHOLOGY TO IMPLEMENTATION OF ADVANCED THERAPIES**

The complexity of the pathological mechanisms underlying muscular dystrophies and myopathies will be discussed.

Chair: Enrico Bertini - Ospedale Pediatrico Bambino Gesù, Roma

15.00 – 15.15 **Introduction**

15.20 – 15.40 **Store-operated calcium entry (SOCE): role in skeletal muscle function and disease**

Feliciano Protasi – Università degli Studi G. d'Annunzio Chieti, Pescara

15.40 – 16.00 **Autophagy as a key pathogenic mechanism in COL6-related diseases and its significance
for prospective therapies**

Paolo Bonaldo – Università degli Studi di Padova

16.00 – 16.20 **From reverse genetics to gene therapy: Duchenne muscular dystrophy**

Alessandra Ferlini – Università degli Studi di Ferrara

16.20 – 16.40 **Cross-fertilization between motor neuron disorders and muscular dystrophies: improving
care and targeting treatments in Myotonic Dystrophy type 1**

Valeria Sansone – Centro Clinico Nemo and Università degli Studi di Milano

16.40 – 16.50 **Flash Talk – Neuromuscular biobanks: a support to research and patients' needs**

Giacomo Comi – Ospedale Maggiore Policlinico, Università degli Studi di Milano

17.00 – 17.30 Flash poster presentations

*The selected poster presenters will have the chance to give a Flash Talk presentation of 3-minutes to
“advertise” their work prior to the poster session.*

17.30 – 18.00 *Coffee break*

18.00 – 19.15 POSTER SESSION 1

**19.15 – 20.15 SCIENCE AND SOCIETY - WHOSE DATA ARE MY DATA? SHARING AND PROTECTING
PERSONAL HEALTH DATA**

An interactive session addressing patients' needs, concerns and expectations on personal health data and illustrating key principles relevant to health data management in research.

Moderator: Lucia Monaco – Research Impact and Strategic Analysis, Fondazione Telethon
Sandra Courbier – EURORDIS, Paris

Michela Maggi – Fondazione Telethon Data Protection Officer (DPO), Studio Legale Maggi,
Milano

20.15 – 21.30 *Welcome buffet*

Tuesday, October 29th 2019

08.30 – 09.00 *Registration and poster setting up*

09.00 – 11.00 PLENARY SESSION 2 - NEW THERAPEUTIC AVENUES FOR MITOCHONDRIAL DISEASES

Different approaches to tackle mitochondrial diseases will be presented and discussed.

Chairs: Giorgio Casari – TIGEM, Pozzuoli (Napoli) and Università Vita Salute San Raffaele, Milano
Gabriele Siciliano – Clinica Neurologica, Università degli Studi di Pisa

9.00 – 9.15 **Introduction**

9.20 – 9.40 **Omic approaches to improve diagnostics (and optimize treatment) for patients with mitochondrial disease**

Daniele Ghezzi – Fondazione IRCCS Istituto Neurologico "C. Besta", Milano

9.40 – 10.00 **Mitochondrial disorders: from gene discovery to pathomechanisms and experimental therapy**

Massimo Zeviani – Università degli Studi di Padova

10.00 – 10.20 **miR-181a and miR-181b downregulation ameliorates mitochondrial-associated neurodegeneration by enhancing mitochondrial biogenesis and mitophagy**

Brunella Franco – TIGEM, Pozzuoli (Napoli)

10.20 – 10.40 **Mitochondria as signalling hubs in neurodegeneration**

Rosario Rizzuto – Università degli Studi di Padova

10.40 – 10.50 **Flash Talk - The 'Cell line and DNA Bank of Genetic Movement Disorders and Mitochondrial Diseases' of the TNGB as a key resource for research**

Barbara Garavaglia – Fondazione IRCCS Istituto Neurologico "C. Besta", Milano

11.00 – 11.30 *Coffee break*

11.30 – 13.00 POSTER SESSION 2 – Meeting Patient Organizations

This poster session will be shared with Patient Organizations who were informed of the posters of interest. A useful slot for connecting Researchers and Patients.

13.00 – 14.00 *Buffet lunch*

14.00 – 14.15 Fondazione Telethon partners with Patient Organizations in a Seed Grant Special Program

Alessandra Camerini, Patient Associations Relationship – Fondazione Telethon
Manuela Battaglia, Head of Research – Fondazione Telethon

14.15 – 15.30 FOCUS - INTERNATIONAL ACTIVITIES AT FONDAZIONE TELETHON

International partnerships and European projects on research infrastructures, policy recommendations and strategic research agendas: why to engage and the potential impact for a Researcher.

Moderator: Graciana Diez Roux - TIGEM, Pozzuoli (Napoli)

RESTORE - Hans-Dieter Volk, BCRT and Charité – Universitätsmedizin Berlin, Germany

EJP rare diseases - Daria Julkowska, Inserm, Paris, France

IRDiRC - Lucia Monaco, Fondazione Telethon and IRDiRC

RARE 2030 - Stefano Benvenuti, Fondazione Telethon

ENMC - Anna Ambrosini, Fondazione Telethon

15.10 - 15.30 **Roundtable**

15.30 – 17.30 PLENARY SESSION 3 - GOING TRANSLATIONAL

The path towards therapy development: what it takes, the tools available and some good news.

Chair: Michael Caplan – Yale University, New Haven, CT, USA

15.30 – 15.40 **Introduction**

15.40 – 16.00 **What it takes to go translational**

Dieter Volk – BCRT and Charité – Universitätsmedizin Berlin, Germany

16.00 – 16.10 **The Sofinnova - Telethon fund: a new opportunity**

Annamaria Merico - Fondazione Telethon

16.10 – 16.30 **Orphan drug development guidebook: the IRDiRC toolkit to increase efficiency in the development of new treatments for rare disease**

Diego Ardigò - Chiesi Farmaceutici, Parma

16.30 – 16.50 **Artificial Intelligence in Biomedical Research: where we are and where we are going**

Robert Alexander - IBM Healthcare & Life Science, Roma

Chair: Robertson Parkman – Stanford University, Stanford, CA, USA

16.50 – 17.00 **Update on clinical trials**

Alessandro Aiuti – SR-TIGET, Milano

Nicola Brunetti Pierri – TIGEM, Pozzuoli (Napoli)

17.00 – 17.30 **A focus on mucopolysaccharidoses (MPS) I**

Maria Esther Bernardo and Bernhard Gentner – SR-TIGET, Milano

17.30 – 18.30 **Poster Session & Coffee Break**

18.30 – 19.30 SCIENCE and SOCIETY- ACCESSIBILITY AND SUSTAINABILITY OF ADVANCED THERAPIES

The roadblocks between EMA positive opinion for marketing authorization and patients: analysis of challenges, possible solutions and pioneers' experiences.

Moderator: Francesco Macchia – Osservatorio Terapie Avanzate

Diego Ardigò – Chiesi Farmaceutici, Parma

Christos Sotirelis - Patient Expert in the Committee for Advanced Therapies (CAT), European Medicines Agency (EMA)

Simona Paratore – Novartis Oncology

Roundtable

Wednesday, October 30th 2019

9.00 – 10.30 FOCUS - GOING BEYOND ANIMAL EXPERIMENTATION: WHEN AND WHERE

Cutting-edge alternatives to animal experimentation: are we there yet?

Moderator: Manuela Battaglia - Fondazione Telethon

9.00 – 9.20 **Let's hear from the Telethon-Institutes' Directors**

Andrea Ballabio - TIGEM, Pozzuoli (Napoli)

Luigi Naldini - SR-TIGET, Milano

- 9.20 – 9.40 **How can C. elegans worm your way into the study of genetic diseases**
Elia Di Schiavi - CNR, Napoli
- 9.40 – 10.00 **One by one: convergence, multiplexing and single-cell resolution in the study of neurodevelopmental disorders through brain organoids**
Giuseppe Testa - Università degli Studi di Milano - Istituto Europeo di Oncologia, Milano
- 10.00 – 10.20 **Organs-on-Chips: the promises and limits of microfluidics**
Diego Di Bernardo - TIGEM, Pozzuoli (Napoli)
- 10.20 – 10.30 **Concluding remarks: MANIFESTO “Salviamo la ricerca Biomedica” and next steps**
Giuliano Grignaschi - Research4life, IRCCS Istituto M. Negri, Milano

10.30 – 11.00 *Coffee break*

11.00 – 13.00 PLENARY SESSION 4 - FIGHTING BLINDNESS: AN EYE ON NEW THERAPEUTIC APPROACHES AND CHALLENGES

Novel and various approaches to tackle eye diseases will be presented and discussed.

Chair: Sandro Banfi - TIGEM, Pozzuoli (Napoli)

11.00 – 11.15 **Introduction**

11.20 – 11.40 **Expanding AAV transfer capacity in the retina**

Alberto Auricchio – TIGEM, Pozzuoli (Napoli)

11.40- 12.00 **Targeting common cell death pathways for the neuroprotection of degenerating photoreceptors**

Valeria Marigo, Università degli Studi di Modena e Reggio Emilia, Modena

12.00– 12.20 **Cone dystrophies and retinal degeneration from protein structures to biological networks**

Daniele Dell’Orco, Università degli Studi di Verona

12.20– 12.40 **Inhibition of autophagy curtails visual loss in a model of autosomal dominant optic atrophy**

Luca Scorrano, Università degli Studi di Padova

12.40– 12.50 **Flash Talk – Fondazione Telethon and the Unit of Advanced Therapies for Hereditary Ocular Diseases**

Stefano Zancan, Fondazione Telethon

13.00 – 13.30 LATE BREAKING NEWS

Authors of just-published important results will have the chance to present their data.

Endothelial cell clonal expansion in the development of cerebral cavernous malformations

Matteo Malinverno, IFOM-The FIRC Institute of Molecular Oncology Foundation, Milano

Inhibition of DNA damage response at telomeres improves the detrimental phenotypes of Hutchinson-Gilford Progeria Syndrome

Francesca Rossiello, IFOM-The FIRC Institute of Molecular Oncology, Milano

13.30 – 13.45 CLOSING REMARKS

Manuela Battaglia – Head of Research, Fondazione Telethon