

XX Scientific Convention

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

FONDAZIONE



Monday, 28th October 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.

15.00 - 15.15 **Introduction**
15.15 – 15.35 **Store-operated calcium entry (SOCE): role in skeletal muscle function and disease**
Feliciano Protasi - Università degli Studi G. d'Annunzio Chieti, Pescara
15.35 – 15.50 **Flash Talk - Neuromuscular biobanks: a support to research and patients' needs -**
Monica Sciacco, Centro Dino Ferrari, Università degli Studi di Milano
15.50– 16.10 **Autophagy as a key pathogenic mechanism in COL6-related diseases and its significance
for prospective therapies**
Paolo Bonaldo - Università degli Studi di Padova
16.10 – 16.30 **From reverse genetics to gene therapy: Duchenne muscular dystrophy**
Alessandra Ferlini - Università degli Studi di Ferrara
16.30-16.50 **Cross-fertilization between motor neuron disorders and muscular dystrophies: improving
care and targeting treatments in Myotonic Dystrophy type 1**
Valeria Sansone - Centro clinico Nemo, Milano
16.50-17.00 **Telethon Science Highlights – past, present and future**
17.00-17.30 **Flash poster presentations**
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 the management of health data in the research setting.

Moderator: Lucia Monaco – Fondazione Telethon, Irdirc

Sandra Courbier – Eurordis, Paris

Michela Maggi - Telethon Data Protection Officer (DPO), Studio legale Maggi, Milan

20.15 – 21.30 *Welcome buffet*

Tuesday, 29th October 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.

09.00 – 9.15 **Introduction**

Chair: Giorgio Casari, Università Vita Salute San Raffele, Milano

9.15 - 9.40 **Omics approaches to improve diagnostics (and optimize treatment) for patients with mitochondrial disease**

Daniele Ghezzi – Fondazione I.R.C.C.S Istituto Neurologico "C.Besta"

9.40 - 10.00 Massimo Zeviani – Università di Padova

10.00 - 10.20 Brunella Franco – TIGEM, Pozzuoli (Napoli)

10.20 - 10.40 **Mitochondria as signaling hubs in neurodegeneration**

Rosario Rizzuto –Università 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 I.R.C.C.S Istituto Neurologico "C.Besta"

10.50-11.00 Telethon Science Highlights – past, present and future

11.00 – 11.30 *Coffee break*

11.30 – 13.00 POSTER SESSION 2 – Meeting Patient Organizations

13.00 – 14.00 *Buffet lunch*

14.00-14.15 Fondazione Telethon partners with Patient Association in a Seed Grant Special Program

Alessandra Camerini, Patient Association - 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, strategic research agendas: why to engage and what could be the impact for a researcher.

Introduction - *Chair:* 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

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.

- 15.30-15.40 **Introduction**
Chair: Michael Caplan – Yale University
- 15.40 – 16.00 **What it takes to go translational**
 Dieter Volk - BCRT and Charité – Universitätsmedizin Berlin, Germany
- 16.00 – 16.15 **The Sofinnova - Telethon fund: a new opportunity**
 Annamaria Merico, Head of Technology Transfer – Fondazione Telethon
- 16.15 - 16.30 Angelo Lombardo – SR-TIGET, Milano
- 16.30 - 16.50 **Orphan drug development guidebook: the IRDiRC's toolkit to increase efficiency in the development of new treatments for rare disease**
 Diego Ardigò
- 16.50 – 17.00 **Update clinical trials**
 Alessandro Aiuti - SR-TIGET, Milano
 Nicola Brunetti Pierri - TIGEM, Pozzuoli (Napoli)
- 17.00-17.30 **MPS I**
 Bernardo Maria Esther 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**
 Moderator: Francesco Macchia - Osservatorio Terapie Avanzate
 Diego Ardigò, responsabile del dipartimento malattie rare di Chiesi

Wednesday, 30th October 2019

- 9.00– 10.30 FOCUS – GOING BEYOND ANIMAL EXPERIMENTATION: WHEN AND WHERE**
An overview on the alternative tools to model genetic diseases for both basic and translational studies.
Moderator: Manuela Battaglia
- 9.00 – 9.20 **The Institutes' Director point of view**
 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 **Organoids**
 Giuseppe Testa – Università degli Studi di 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 and MANIFESTO “Salviamo la ricerca Biomedica” and next steps**
 Giuliano Grignaschi – IRCCS-Istituto M. Negri, Milano
- 10.30 – 11.00 *Coffee break*
- 11.00 – 13.00 PLENARY SESSION 5 - EYE DISEASES**
The availability of novel and different approaches to tackle eye diseases will be addressed.
- 11.00– 11.10 Video

- 11.10 - 11.20* **Introduction - Chair: Sandro Banfi**
- 11.20 - 11.40* Alberto Auricchio – TIGEM, Pozzuoli (Napoli)
- 11.40- 12.00* Valeria Marigo – Università Modena Reggio Emilia
- 12.00 - 12.20* **Cone dystrophies and retinal degeneration from protein structures to biological networks**
Daniele Dell’Orco – Università di Verona
- 12.20 - 12.40* **Inhibition of autophagy curtails visual loss in a model of autosomal dominant optic atrophy**
Luca Scorrano – Università di Padova
- 12.40- 12.50* **Flash Talk** – Stefano Zancan, Fondazione Telethon
- 13.00 – 13.30** **LATE BREAKING NEWS**
- 13.30 - 13.45** **CLOSING REMARKS** - by Manuela Battaglia, Head of Research Fondazione Telethon