

## Mitochondrial Disease Conference

October 25<sup>th</sup> - 27<sup>th</sup> , 2024

**BW Plus Hotel Galileo, Via Venezia 30 Padova**



### Welcome to the Mitochondrial Disease Conference 2024

On behalf of Mitocon and its Scientific Programme Committee, we are pleased to announce the organization of the 14<sup>th</sup> Mitochondrial Disease Conference, which will be held on October 25-27, 2024 at the Best Western Hotel Galileo Padova.

As usual, the event will consist of two parts: the scientific meeting on 25<sup>th</sup> and 26<sup>th</sup> October and the patient and family meeting on Sunday 27 October.

During the Scientific Conference, the participation of patients and families will also be encouraged.

### Summary

Since 2010 **Mitocon** has organized the **Mitochondrial Disease Conference** to encourage the exchange of knowledge and experience between doctors and researchers, patients, family members and caregivers, with the participation of the leading Italian and international experts.

**Mitochondrial diseases** are very complex disorders with multi-system manifestations and different ages of onset, ranging from infancy to adulthood. In recent years, giant steps have been taken in the study of the basic mechanisms involved in these diseases and we have finally moved from diagnosis to the study of possible treatments. We are also witnessing an acceleration in the field of mitochondrial medicine, thanks to the availability of new technologies, new therapeutic approaches and possibly clinical trials to improve the lives of our patients.

The Mito Conference 2024 aims to provide a comprehensive overview of the latest advances, challenges, and potential opportunities in the field of mitochondrial research. By covering diverse topics ranging from basic science to mitochondrial biology, with a focus on therapies and the role of artificial intelligence in mitochondrial medicine.

We also stimulate the participation of young scientists and clinicians who will form the next generation of researchers in the field of mitochondrial medicine, The Mitocon scientific conference is entirely in English to facilitate the participation of doctors and researchers from all over the world. **A simultaneous translation into Italian will be provided.**

**Mitocon** is the Italian leading non-profit mitochondrial disease patients organization striving to advance research towards finding a cure. Encouraging collaboration and the exchange of knowledge within the scientific community is our priority.

Get in touch with us on [mitocon.it](http://mitocon.it)

**Join us at the 14th Mitochondrial Disease Conference. Together, let's light the way to a cure for all mitochondrial diseases.**

**We look forward to welcoming you to this remarkable event!**

## Scientific Programme Committee

**Anna Ardisson**, Department of Pediatric Neuroscience, IRCCS Foundation Neurological Institute "Carlo Besta, Milan

**Enrico Bertini**, Neuromuscular and Neurodegenerative Disorders, Bambino Gesù Children's Research Hospital, Rome.

**Valerio Carelli**, IRCCS Institute of Neurological Sciences (ISNB), Bellaria Hospital, Bologna/University of Bologna.

**Massimiliano Filosto**, NeMO Clinical Center-Brescia for Neuromuscular Diseases; Department of Clinical and Experimental Sciences, University of Brescia.

**Costanza Lamperti**, Division of Medical Genetics and Neurogenetics, Mariani Foundation Center for the Study of Mitochondrial Pediatric Diseases, Foundation IRCCS Neurological Institute "C. Besta", Milan.

**Michelangelo Mancuso**, AOUP, Neurological Institute, University of Pisa.

**Olimpia Musumeci**, AOU Policlinico "G.Martino", University of Messina.

**Serenella Servidei**, Neurology Institute, Catholic University of the Sacred Heart, Rome.

**Valeria Tiranti**, Division of Medical Genetics and Neurogenetics, Mariani Foundation Center for the Study of Mitochondrial Pediatric Diseases, Foundation IRCCS Neurological Institute "C. Besta", Milan.

**Carlo Fiore Viscomi**, Department of Biomedical Sciences, University of Padova.

## Scientific Programme

### Friday 25<sup>th</sup> – Luna Room

12.00 –13.00

#### Arrivals, Registration & Light lunch

13.15

#### Welcome to the Mitochondrial Diseases Conference 2024

**Marco Marmotta**, President, Mitocon – Insieme per lo Studio e la Cura delle Malattie Mitocondriali ODV

**Rosario Rizzuto**, University of Padova

**Costanza Lamperti, Michelangelo Mancuso**, Scientific Committee Delegates

**What Does Mitochondrial Disease Mean to Me** – Mito Patients and Families ' /Video Stories

14.00

#### Introduction:

Chair: **Valeria Tiranti**, IRCCS Foundation “Carlo Besta” Neurological Institute

#### Keynote Lecture

**Michal Minczuk**, University of Cambridge, UK

14:45 – 16:15

#### Session 1 - Basic Science

Chair: **Carlo Viscomi**, University of Padova

Chair: **Paola Desideri**, Mitocon

#### Speakers

14:45 **Mitochondrial Control of Innate Immunity and Significance for Primary Mitochondrial Disorders** - **Marco Tigano**, Thomas Jefferson University

15:15 **Patrick Chinnery**, University of Cambridge

15:45 **In Vitro iPSC-Derived Models of mtDNA-Related Disorders** - **Valeria Tiranti**, IRCCS Foundation “Carlo Besta” Neurological Institute

#### 16:15 Posters with coffee

Chair: **Serenella Servidei**, Catholic University of the Sacred Heart

Chair: **Paola Desideri**, Mitocon

### 17:00 – 18:30 Session 2

#### Natural Histories Studies

Chair: **Enrico Bertini**, Bambino Gesù Children's Research Hospital IRCCS

Chair: **Paola Desideri**, Mitocon

### *Speakers*

17:00 **Advances in MELAS** - **Costanza Lamperti**, IRCCS Foundation "Carlo Besta" Neurological Institute

17:30 **The Natural History of PMM in the light of clinical trial experience** - **Serenella Servidei**, Catholic University of the Sacred Heart

18.00 **Advances in POLG** - **Omar Hikmat**, Haukeland University Hospital

18:30 Closing day 1

## **Day 2 Saturday 26<sup>th</sup> October – Luna Room**

**8:30 Arrivals and Registrations**

**8:45-10.15 Session 3**

### **Advances in Therapies**

Chair: **Anna Ardissona**, IRCCS Foundation "Carlo Besta" Neurological Institute

Chair: **Piero Santantonio**, Mitocon

### *Speakers*

8:45 **Diagnosis and Treatment in Mitochondrial DNA Depletion Syndrome** - **Caterina Garone**, University of Bologna

9:15 **Rapamycin-based therapy as a promising therapeutic option for MERRF** - **Alessandra Maresca**, University of Bologna

9:45 **AntagoMiRs restoring the levels of the master regulator of mitochondrial shape Opa1 ameliorate mitochondrial myopathies** - **Luca Scorrano**, Veneto Institute of Molecular Medicine

10.15 Coffee break

### **10.30 -11.30 Young Investigator Session**

Chair: **Valeria Tiranti**, IRCCS Foundation "Carlo Besta" Neurological Institute

Chair: **Piero Santantonio**, Mitocon

10:30 Talk 1\*

10:45 Talk 2\*

11:00 Talk 3\*

11:15 Talk 4\*

11:30 -13:30

### **Roundtable with Pharmaceutical Industries and Regulatory Authorities**

Chair: **Michelangelo Mancuso**, University of Pisa

Chair: **Valerio Carelli**, University of Bologna

11:30 Pharma introduction

12:20 Round Table with pharmaceutical industries and regulatory authorities

Confirmed Panel Members:

- **Toni Andreu**-EATRIS

13:30 Light Lunch

### **14.30 – 15.30 Session 4**

#### **Exploring digital health technologies and AI in mitochondrial medicine**

Chair: **Massimiliano Filosto**, University of Brescia

Chair: **Piero Santantonio**, Mitocon

#### **Speakers**

14.30 **Biosensors and remote monitoring** (TBC)

14.50 **ePROMs, APPs and patient engagement** - **Manuela Lavorato**, University of Pisa

15.10 **Applying machine learning to define phenotypes and to predict clinical course. Are we ready?** - **Andrea Bandini**, Sant'Anna School of Advanced Studies, Pisa

15.30 Coffee break

### **16.00 MitoAward**

Chair: **Serenella Servidei**, Catholic University of the Sacred Heart

Chair: **Carlo Viscomi**, University of Padova

16:00 Talk1\*

16:10 Talk2\*

16:20 Talk 3\*

### **16:30 -17:30**

#### **LATE BREAKING NEWS**

Chair: **Olimpia Musumeci**, University of Messina

Chair: **Piero Santantonio**, Mitocon

16:30 Talk 1

16:50 Talk 2

17:10 Talk 3

17:30-18:00

### **Closing Lecture**

**An overview on the state of Mitochondrial Research in Italy, Massimo Zeviani** -  
IRCCS materno infantile Burlo Garofolo

**18:00 Closing Remarks**

**18:30 Consulenze con gli esperti per pazienti e famiglie**

## **Day 2 Saturday 26<sup>th</sup> - Ganimede Room**

### **14:30 -16.15 Satellite Symposium: Mitochondrial Optic Neuropathies**

Chair: **Valerio Carelli**, University of Bologna

Moderator: **Paula Morandi**, Mitocon

### **Speakers**

14:30 **Molecular diagnostics for Mitochondrial Optic Neuropathies** - **Leonardo Caporali**, IRCCS Institute of Neurological Sciences, Bologna

14:45 **Pharmacogenomics in LHON** - **Anna Maria Ghelli**, University of Bologna

15:00 **Patrick Yu-Wai-Man**, University of Cambridge

15:15 **Gene therapy in Mitochondrial Optic Neuropathies** - **Chiara La Morgia**,  
University of Bologna

15:30 Coffee break in the plenary room

15.45 **What we have learned from clinical trials in Mitochondrial Optic Neuropathies and new upcoming trials** - **Thomas Klopstock**, University of Munich

16.00 **Q&A**

16:15 **Valerio Carelli**

Closing Remarks

\*Talks to be selected by call for abstract submission

## **Convention Pazienti e Famiglie Programma**

**Domenica 27 Ottobre – Sala Europa**

9.30 –9:45

### **Arrivi e Registrazione Ospiti**

9:45

### **Saluto di Benvenuto**

Marco Marmotta, President Mitocon

Piero Santantonio, Mitocon Founder

10:00 – 11:00

### **Tavola Rotonda: Tecniche di sostituzione del DNA mitocondriale: dal percorso normativo al progetto pilota.**

Intervengono:

Daniela Zuccarello, Azienda Ospedale Università di Padova

Piero Santantonio, Fondatore Mitocon

Istituzioni (TBC)

11:30 – 12:00

### **Sanità Partecipata nella Regione Veneto**

Elena Pegoraro

12:00 – 13:00

### **Mitocon: una rete di persone a supporto dei pazienti e della ricerca scientifica**

Marco Marmotta, Presidente Mitocon