

## Mitochondrial Diseases Conference

October 7-8-9, 2022

Holiday Inn Rome – Parco dei Medici

### Summary

Since 2010 Mitocon organizes the Mitochondrial Diseases 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, still little known by the scientific community, 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 the diagnosis to the study of possible treatments.

Since its first edition our conference has contributed to this process by gathering the entire community of Italian doctors and researchers around the association of patients and helping to strengthen international partnerships and collaborations, making the whole community grow together and contributing to sharing knowledge and research progress.

The conference is **hybrid** with both **onsite and online participation** (online participation will be possible only for October 7 and 8). Since previous editions, the scientific sessions will be entirely in English to facilitate the participation of doctors and researchers from all over the world, promoting international exchange and collaboration, crucial to fight together against mitochondrial diseases. 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)

## Scientific Programme Committee

**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.

## Speakers and Moderators

**Anthony Abbruscato**, Senior Director, Clinical Development, Stealth BioTherapeutics

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

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

**Gino Cortopassi**, Professor Molecular Biosciences, University of California

**Lorena Castillo**, Head of the Neuro-Ophthalmology Department at the ICR Ophthalmology Center

**Madhu Davies**, Medical Director, Reneo Pharma Ltd

**Michael Decressac**, Inserm Researcher at Grenoble Institut Neurosciences

**Paola Desideri**, Vice President Mitocon

**Michela Di Nottia**, Researcher Neuroscience, Bambino Gesù Children's Hospital

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

**Magnus Hansson**, Chief Medical Officer, VP preclinical & clinical development, Abliva AB

**Mary Herbert** Professor of Reproductive Biology, Newcastle Fertility Centre International Centre for Life

**Amel Kaara** Amel Karaa, M.D., Massachusetts General Hospital

**Jin- Soo Kim** Director Center for Genome Engineering, Institute for Basic Science, Republic of Korea

**Matthew Klein**, Chief Operating Officer, PTC Therapeutics

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

**Andrea Legati** PhD, The Foundation of the Carlo Besta Neurological Institute, IRCCS · Unit of Medical Genetics and Neurogenetics

**Roan Louw**, Research Director, Human Metabolomics at North-West University, Noordwes-Universiteit

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

**Marco Marmotta** President, Mitocon – Insieme per lo studio e la cura delle malattie mitocondriali

**Robert McFarland** Professor of Paediatric Mitochondrial Medicine and Hon Consultant Paediatric Neurologist, Wellcome Centre Mitochondrial Research, Newcastle University

**Paula Morandi**, Patients' Representative for Mitochondrial Eye Diseases Member of Mitocon, IMP Board Member

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

**Robert Pitceathly** Principal Research Fellow Department of Neuromuscular Diseases UCL Queen Square Institute of Neurology

**Alessandro Prigione**, Associate Professor of Pediatric Metabolic Medicine at the Department of General Pediatrics, Neonatology and Pediatric Cardiology at Heinrich Heine University (HHU) in Düsseldorf, Germany

**Piero Santantonio**, Founder Mitocon

**Luca Scorrano**, Professor of Biochemistry Dept. of Biology, University of Padova and VIMM

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

**Antonella Spinazzola** Professor PI, Neuroscience and Mitochondrial Medicine Brain Research UK Miriam Marks Senior Fellow UCL Queen Square Institute of Neurology, Royal Free Campus, London

**Magali Taiel**, Chief Medical Officer, Gensight-Biologics

Foundation Center for the Study of Mitochondrial Pediatric Diseases, Foundation IRCCS Neurological Institute "C. Besta"

**Livia Tomasso**, Livia Tomasso. Medical Advisor Rare Diseases, EU & Emerging Markets Global Rare Disease Business Unit, Chiesi Group

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

**Philip Yeske**, PhD Science & Alliance Officer, United Mitochondrial Disease Foundation

**Massimo Zeviani** Direttore Scientifico IRCCS Burlo Garofolo, professore di Neurologia, Fisiologia umana, Patologia Generale

## Scientific Programme

**Continuing Medical Education- CME will be provided only for those who attend in presence**

### Plenary room

#### Day 1: Friday, October 7

09.00 - 09.30 AM

#### Arrivals & Registration

09.30 - 10.00 AM

#### Welcome to the Mitochondrial Diseases Conference 2022

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

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

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

10.00 - 10.30 AM

#### Keynote Lecture Manipulating mitochondrial DNA

Introduction: **Carlo Viscomi**

#### *Speaker*

**Mary Herbert**

10:30 – 10:50 AM

## A new era of mitochondrial genome editing

### *Speaker*

**Jin-Soo Kim**

Q&A

11:00-11:30 AM

Coffee Break

11:30 – 1:00 PM

## Session 1: Basic Science

### *Moderators*

*Valeria Tiranti, Piero Santantonio*

### *Speakers*

11:30 **Antonella Spinazzola** - Mitochondrial DNA disorders: mechanisms and potential therapeutic strategies

11:50 **Michela Di Nottia** - Free cytosolic-mitochondrial DNA triggers a potent type-I Interferon response in Kearns–Sayre patients counteracted by mofetil mycophenolate

12:10 **Andrea Legati** - Advantages of short and long-read NGS in the genetic analysis of the mitochondrial DNA

12:30 **Luca Scorrano** - A screening for compounds that ameliorate mitochondrial function in ADOA

Q&A

1:00- 2:30 PM - Lunch

2:30 – 4:15 PM

## Session 2: Leigh syndrome road map

### *Moderators*

*Philip Yeske, Piero Santantonio*

### *Speakers*

2:30 Introduction: **Massimo Zeviani**

2:50 TO BE CONFIRMED)

3:10 **Alessandro Prigione** - Induced pluripotent stem cells (iPSC)-driven drug repositioning for Leigh syndrome

3:25 **Roan Louw** - Metabolic investigation of Leigh Syndrome – the search for novel treatment options

3:40 **Gino Cortopassi** - Fumarates rescue multiple mouse models of Mitochondrial disease': Leigh, Friedreich's, LHON

3:55 **Phil Yeske** - Patient-Led Research- An Update on the Global Leigh Syndrome Roadmap Project

Q&A

4:15 – 4:45PM Coffee Break

4:45 - 5:45 PM

## Session 3: Exploring and managing PMD phenotypes

### *Moderators*

*Michelangelo Mancuso, Paola Desideri*

### **Speakers**

4:45 **Robert McFarland** - Management of stroke like episodes

5:10 **Amel Kaara** - Approaching a patient with possible PMD: an existential crisis?

Q&A

5:45 - 6:15 PM

## Posters and Mitoldeas Award

### *Moderators*

*Costanza Lamperti, Paola Desideri*

6.30 PM **End of day 1**

**Simultaneous translation will be provided during the scientific sessions**

Mitocon - Insieme per lo studio e la cura delle Malattie Mitocondriali Odv

Organizzazione di Volontariato iscritta al Registro della Regione Lazio – Sezione II n° 705 del 8/9/2008

CF: 97488070588 Sede legale: Via Francesco Benaglia,13 00153 Roma - Tel. 06 66991 333/334 - Contact center pazienti: 340 7569156

Sito internet: [www.mitocon.it](http://www.mitocon.it) E-mail: [info@mitocon.it](mailto:info@mitocon.it) PEC: [mitocononlus@pec.it](mailto:mitocononlus@pec.it)

## Day 2: Saturday October 8

8:30 -9:00

### Arrivals & Registration

9:00 -9:20 AM

### Session 4: Progress in clinical research for new treatment options for LHON: an exciting landscape in front of Patients and Clinicians

#### **Moderators**

*Valerio Carelli, Paula Morandi*

#### **Speaker**

**Lorena Castillo**

9:20 -10:30 AM

### Session 5: Challenges and pitfalls in clinical trials design – presentations and round table discussion

#### **Moderators**

*Enrico Bertini, Paula Morandi*

#### **Speakers**

9:20 **Magali Taniel** - Gene Therapy Development in Leber Hereditary Optic Neuropathy

9:30 **Livia Tomasso**, Medical Advisor Rare Diseases, EU & Emerging Markets Global Rare Disease Business Unit, Chiesi Group

9:40 **Madhu Davies**, Medical Director, Reneo Pharma Ltd

9:50 **Magnus Hansson**, Chief Medical Officer, VP preclinical & clinical development, Abliva

10:00 **Anthony Abruscato**, Senior Director, Clinical Development, Stealth BioTherapeutics

10:10 **Matthew Klein**, Chief Operating Officer, PTC Therapeutics

10:20 **TO BE CONFIRMED**

Q&A



10:30 -11:00 Coffee Break

11:00- 11:30

## **Posters and Mitoldeas Awards 2022**

### **Moderator**

*Costanza Lamperti, Paola Desideri*

11:30- 12:15 PM

## **Session 6: Breaking news**

### **Moderator**

*Olimpia Musumeci, Piero Santantonio*

### **Speakers**

11:30 Breaking News

11:45 Breaking News

12:00 Breaking News

Q&A

12:30 – 1:30 PM

## **Session 7: COVID-19 and Long COVID in PMD**

### **Moderator**

*Serenella Servidei, Paola Desideri*

### **Speakers**

12:30 **Robert Pitceathly** - The European Mito COVID Experience.

12:50 **Massimiliano Filosto** – Long Covid manifestations in Neuromuscular and mitochondrial diseases.

Q&A

1:30 Closing of the scientific part of the congress

