



# mito confe rence

14<sup>TH</sup> MITOCHONDRIAL  
DISEASE CONFERENCE

**2024**

 **Mitocon**

Insieme per lo studio e la cura  
delle malattie mitocondriali ODV



14<sup>TH</sup> MITOCHONDRIAL  
DISEASE CONFERENCE

**2024**

October 25<sup>th</sup> - 27<sup>th</sup>  
BW Plus Hotel Galileo,  
Via Venezia 30 - Padova

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

Dal 2010 Mitocon organizza il Convegno Nazionale sulle Malattie Mitochondriali per favorire lo scambio di conoscenze ed esperienze tra medici e ricercatori, pazienti, familiari e caregiver, con la partecipazione dei principali esperti italiani e internazionali. Mitocon è il principale punto di riferimento in Italia per le persone affette da malattia mitocondriale e le loro famiglie. Sostiene e promuove la ricerca scientifica per accelerare lo sviluppo di terapie per la cura di queste malattie.



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

or write an email to  
o scrivi una mail a  
[scientific.office@mitocon.it](mailto:scientific.office@mitocon.it)



## SCIENTIFIC PROGRAMME

### DAY 1 | FRIDAY 25<sup>TH</sup> | LUNA ROOM

#### 12:00 - 1:00 pm | Arrivals, Registration & Light lunch

#### 1:15 pm | Welcome to the Mitochondrial Disease 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 / Stories

#### 2:00 pm

##### Introduction:

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

##### Keynote Lecture

**Michal Minczuk**, University of Cambridge

#### 2:45 - 4:15 pm | Session 1 | Basic Science

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

Chair: **Massimo Zeviani**, Institute for Maternal and Child Health IRCCS Burlo Garofolo, Trieste

**2:45** Mitochondrial Control of Innate Immunity and Significance for Primary Mitochondrial Disorders - **Marco Tigano**, Thomas Jefferson University

**3:15** Molecular mechanisms purifying mtDNA: a target for disease prevention - **Patrick Chinnery**, University of Cambridge

**3:45** In Vitro iPSC-Derived Models of mtDNA-Related Disorders

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

#### 4:15 pm | Posters with coffee

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

Chair: **Paola Desideri**, Mitocon

#### 5:00 - 6:30 pm | Session 2 | Natural Histories Studies

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

Chair: **Paola Desideri**, Mitocon

**5:00** Natural history of disease in MELAS Syndrome: result of an international collaboration - **Costanza Lamperti**, IRCCS Foundation "Carlo Besta" Neurological Institute

**5:30** The Natural History of PMM in the light of clinical trial experience

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

**6:00** Advances in POLG - **Omar Hikmat**, Haukeland University Hospital

**6:30 | Closing day 1**

## **DAY 2 | SATURDAY 26<sup>TH</sup> | LUNA ROOM**

**8:30 am | Arrivals and Registrations**

**8:45-10.15 am | Session 3 | Advances in Therapies**

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

Chair: **Piero Santantonio**, Mitocon

**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**, Institute of Neurological Sciences of Bologna

**9:45** OpantimiRs, a class of antagonizing microRNAs specific for Opa1, improve mitochondrial myopathies - **Luca Scorrano**, University of Padova

**10.15 am | Coffee break**

**10.30 -11.30 am | Young Investigator Session**

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

Chair: **Piero Santantonio**, Mitocon

**10:30** Pathological molecular mechanisms underlying COA8 loss of function  
**Suleva Povea-Cabello**, University of Padova

**10:45** Ketone bodies resume mitochondrial respiration in neuronal progenitors from iPSCs of patients with AGC1 deficiency - **Simona Nicole Barile**, CNR, Bari

**11:00** Peptide mimetic molecules as potential therapeutic agents against diseases related to mt-tRNA point mutations - **Maria Gemma Pignataro**, Sapienza University, Roma

**11:15** MicroRNA-181a/b modulation as possible therapeutic strategy for Autosomal Dominant Optic Atrophy - **Rosa Saurino**, Telethon Institute of Genetics and Medicine, Telethon Foundation, Pozzuoli

**11:30 -1:15 pm | Roundtable with Pharmaceutical Industries and Regulatory Authorities**

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

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

Moderator: **Piero Santantonio**, Mitocon

**11:30** Pharma introduction

**12:30** Roundtable with Pharmaceutical Industries and Regulatory Authorities

Confirmed Panel Members:

- **Romano Del Fiacco**, UCB
- **Chad Glasser**, Ti Sento Therapeutics
- **Magnus Hansson**, Abliva AB
- **Fabiana Saccheri**, Medical Direction Chiesi
- **Jan Smeitink**, Khondrion
- **Magali Taiel**, Gensight Biologics
- **Toni Andreu**, European Infrastructure for Translational Medicine - EATRIS
- **Armando Magrelli**, Italian Medicines Agency AIFA

**1:30 pm | Light Lunch**

**2:30 - 4:00 pm | Session 4 | Exploring digital health technologies and AI in mitochondrial medicine**

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

Chair: **Piero Santantonio**, Mitocon

**2:30** Biosensors and remote monitoring - **Nikita Konstantinovskiy**, Technical University of Munich

**3:00** ePROMs, APPs and patient engagement - **Manuela Lavorato**, University of Pisa

**3:30** Leveraging Artificial Intelligence to support the diagnosis in Primary Mitochondrial Diseases: Are we ready? - **Andrea Bandini**, Sant'Anna School of Advanced Studies, Pisa

**4:00 pm | Coffee break**



**4.10 pm | MitoAwards**

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

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

**4:10** Flash Talk: A redox cyler approach to treat mitochondrial diseases,

**Daniele Bonesso**, University of Padova

**4:20** Flash Talk Cellular quality control and mitochondrial function in peripheral neuropathies, **Paola Zanfardino** University of Bari

**4:30 -6:00 pm | Late Breaking News**

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

Chair: **Piero Santantonio**, Mitocon

**4:30** Generation of 2D and 3D cellular models to deciphering pathological phenotypes associated with a single large deletion of mtDNA

**Chiara Fasano**, IRCCS Foundation "Carlo Besta" Neurological Institute

**4:50** Cell type-targeted mitochondrial transplantation mediated by protein binders rescues cell degeneration - **Temurkhan Ayupov**, Institute of Molecular and Clinical Ophthalmology Basel, Switzerland

**5:10** A novel GPCR agonist induces selective mitophagy in a Complex III Deficiency Model improving bioenergetic function and overall cellular health

**Cristiane Benincá**, David Geffen School of Medicine, University of California

**5:30** Exploiting redox-active molecules against mitochondrial diseases linked to complex III and complex I dysfunction - **Ildiko Szabó**, University of Padova

**5:50 pm | Mito Awards Ceremony**

**6:00 pm | Closing Remarks**

**Carlo Viscomi**, University of Padova

**Marco Marmotta**, Mitocon

**6:15 pm | Closing of the Scientific Conference**

**2:30 - 4:00 pm | Satellite Symposium: Mitochondrial Optic Neuropathies**

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

Moderator: **Paula Morandi**, Mitocon

**2:30** Molecular diagnostics for Mitochondrial Optic Neuropathies

**Leonardo Caporali**, IRCCS Institute of Neurological Sciences, Bologna

**2:45** Pharmacogenomics in LHON - **Anna Maria Ghelli**, University of Bologna

**3:00** Mitochondrial optic neuropathies: clinical manifestations

**Chiara La Morgia**, IRCCS Institute of Neurological Sciences, Bologna

**3:15** Update on natural history and idebenone efficacy in LHON

**Thomas Klopstock**, University of Munich

**3:30** Gene Therapy for Mitochondrial Optic Neuropathies

**Patrick Yu-Wai-Man**, University of Cambridge

**3.45 pm | Q&A**

**3:55 pm | Closing Remarks**

**Valerio Carelli**, IRCCS Institute of Neurological Sciences, Bologna

N°	Title	Presenter	Affiliation	Category
01	Impact of the LHON m.11778A>G mutation on mitochondrial function in hiPSC-derived RGCs: Insights from Isogenic Control Comparisons	Andrea Cavaliere	Foundation IRCCS Neurological Institute "C. Besta", Milano	Young Investigator
02	FASTKD5 mutations cause a mitochondrial RNA processing defect resulting in COX deficiency and Leigh syndrome	Piervito Lopriore	University of Pisa	Young Investigator
03	From yeast to zebrafish: insights into drug treatment for POLG-related disorders	Raquel Branas Casas	University of Padova	Young Investigator
04	A simple organism to address big questions: how <i>Saccharomyces cerevisiae</i> can support mitochondrial medicine	Martina Magistrati	University of Parma	Young Investigator
05	Exploring twinkle-related disorders: diversity, genotype-phenotype interactions, and international perspectives	Zeynep Unluturk	University of Pisa	Young Investigator
06	In vivo analysis of POLG mutants sheds new light on the pathogenesis of POLG-related disorders	Alessandro Zuppardo	University of Padova	Young Investigator
07	Unveiling the role of inflammation in primary mitochondrial diseases	Martina Zoccola	Roma Bambino Gesù Children's Hospital, IRCCS	Young Investigator
08	mtDNA release and inflammation in MELAS fibroblasts: connecting the dots.	Alessandro Rapone	University of Bologna	Young Investigator
09	Generation and characterization of patient-derived induced pluripotent stem cells and neuronal progenitors carrying the m.8344A>G at high heteroplasmy	Sacchetti Giulia	University of Bologna	Young Investigator

N°	Title	Presenter	Affiliation	Category
10	Yeast NDI1 rescues muscular NDUFS3-/- mouse model: a new prospect of flexible gene therapy for complex-I deficiencies.	Daniele Sala	Foundation IRCCS Neurological Institute "C. Besta", Milano	Young Investigator
11	Steady State Visual Evoked Potentials reveal cortical hyperexcitability in people with m3243A>G mutation.	Francesco Turco	University of Pavia	Young Investigator
12	Identification of potential therapeutic compounds for mitochondrial disorders due to mutations in mitochondrial respiratory chain assembly factors by exploiting yeast models	Nicole Giandebiaggi	University of Parma	Young Investigator
13	Mitochondrial encapsulation to enhance mitochondrial transplantation	Marco D'Amato	Foundation IRCCS Neurological Institute "C. Besta", Milano	Young Investigator
14	Neuro-ophthalmological phenotype and correlations with heteroplasmy levels in MELAS and MERRF syndromes.	Giulia Amore	University of Bologna	Young Investigator
15	Phosphodiesterase 5 Inhibitors as a new treatment for Maternally Inherited Leigh Syndrome (MILS)	Giulia Pedrotti	Foundation IRCCS Neurological Institute "C. Besta", Milano	Young Investigator
16	Expanding the FDXR-associated disease phenotype and defining the underlining disease mechanism in patients' fibroblast cell lines carrying two novel mutations variants	Silvia Sabeni	University of Bologna	Young Investigator
17	Understand mitochondrial defects leading to cardiac damage associated with CHCHD10 variants.	Melanie Abou-Ali	Université Côte d'Azur, Inserm	Young Investigator
18	Evaluating novel mitophagy-activating compounds in patient-derived cellular models of mitochondrial dysfunction.	Giacomo Giacchin	University of Padova	Young Investigator
19	Oral tyrosine supplementation: a novel therapeutic approach in two children with YARS2 pathogenic variants	Giulia Ferrera	Foundation IRCCS Neurological Institute "C. Besta", Milano	Young Investigator
20	The Role of PRICKLE3 in Cell Migration and Mitochondrial Metabolism: Implications for Cancer Metastasis	Pavla Kloverova	Masaryk University, Brno, Czechia	Young Investigator
21	Dissecting and targeting the molecular pathogenesis underlying optic atrophy type 12 (OPA12)	Alessandra Rocco	IRCCS San Raffaele Hospital, Milano	Young Investigator
22	Characterization of Mitochondrial Patients at the Padova University Hospital	Sara Volta	University of Padova	Young Investigator

N°	Title	Presenter	Affiliation	Category
23	Serum biomarkers in Primary Mitochondrial Diseases with different organ involvement. Data from a single center experience	Ignazio Arena	University of Messina	Young Investigator
24	Yeast models of human MTO1 variants confirm two diagnoses of mitochondrial modopathy	Ilaria Notaroberto	University of Parma	Young Investigator
25	The absence of SNF8 in the yeast <i>Saccharomyces cerevisiae</i> leads to mitochondrial dysfunction	Alexandru Ionut	University of Padova	Young Investigator
26	Integrating RNA analysis to improve genetic diagnosis in mitochondrial diseases	Rossella Izzo	Foundation IRCCS Neurological Institute "C. Besta", Milano	Young Investigator
27	Unravelling the role of MPV17	Samantha Corrà	University of Padova	Late Breaking News
28	Urolithin A is a promising treatment for mitochondrial myopathies.	Valeria Balmaceda	University of Padova	Late Breaking News
29	De novo DNMT1L pathogenic variant associated with lethal encephalomyopathy and a literature review	Maria Teresa Bonati	Institute for Maternal and Child Health IRCCS Burlo Garofolo, Trieste	Late Breaking News
30	Yeast-Based Functional Investigation of ELAC2 Variants in Mitochondrial Dysfunction	Camilla Ceccatelli Berti	University of Parma	Late Breaking News
31	Characterization of the BCS1L c.38A>G variant identified in a patient with Björnstad syndrome	Valeria Capaci	Institute for Maternal and Child Health IRCCS Burlo Garofolo, Trieste	Late Breaking News
32	Dissection of Intrinsic apoptosis Pathway in OPA3-KO Model Cell Line	Concetta Valentina Tropeano	IRCCS Istituto delle Scienze Neurologiche di Bologna	Late Breaking News
33	The yeast <i>Saccharomyces cerevisiae</i> as a model to deepen the knowledge about the mitochondrial metallopeptidase PITRM1 in health and disease	Cristina Dallabona	University of Parma	Late Breaking News
34	Role of COQ8A and COQ8B in CoQ synthesis and mitochondrial homeostasis.	Agata Valentino	University of Parma	Late Breaking News
35	A family with autosomic dominant transmission of multiple acyl-coenzyme A (CoA) dehydrogenase deficiency (MADD) disease	Fulvio Celsi	Institute for Maternal and Child Health IRCCS Burlo Garofolo, Trieste	Late Breaking News



N°	Title	Presenter	Affiliation	Category
36	Role of COQ8A and COQ8B in CoQ synthesis and mitochondrial homeostasis.	Kristýna Čunátová	Czech Academy of Sciences, Prague, Czech Republic	Late Breaking News
37	Early initiation of ketogenic diet in a newborn affected by pyruvate dehydrogenase complex deficiency	Niccolò Campagna	Florence University	Late Breaking News
38	Four-Year Results of Bilateral Injection of Lenadogene Nolparvovec Gene Therapy for Leber Hereditary Optic Neuropathy	Magali Taiel	GenSight Biologics, Paris, France	Pharma
39	PRIZM: A Phase 2b randomized, double-blind, placebo-controlled, crossover study evaluating the efficacy and safety of zagociguat in participants with MELAS	Chad Glasser	Tisento Therapeutics	Pharma
40	FALCON: A Randomized, Placebo-Controlled Study of the Efficacy of KL1333 in Adult Patients with Primary Mitochondrial Disease	Magnus Hanson	Abliva	Pharma
41	Employment of Spatial Omics and Quinomics Technologies For the Advancement of Mitochondrial Targeted Therapeutics	Michael A. Kiebish	BPG BIO	Pharma

**DIGITAL  
ABSTRACT BOOK**  
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## PROGRAMMA SCIENTIFICO



**GIORNO 1 | VENERDÌ 25 | SALA LUNA**

### **12.00 - 13.00 | Arrivo, Registrazione & Light lunch**

**13.15** Apertura dei lavori e saluti di benvenuto

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

**Rosario Rizzuto**, Università di Padova

**Costanza Lamperti, Michelangelo Mancuso**, Delegati del Comitato Scientifico Vivere con una Malattia Mitocondriale - Testimonianze di Mito Pazienti e Famiglie

### **14.00 | Introduzione**

**Valeria Tiranti**, Fondazione IRCCS Istituto Neurologico "Carlo Besta"

*Lectio Magistralis: Michal Minczuk, Università di Cambridge*

### **14:45 - 16:15 | Sessione 1 - Ricerca di Base**

*Moderatore: Carlo Viscomi, Università di Padova*

*Moderatore: Massimo Zeviani, IRCCS Materno Infantile Burlo Garofolo, Trieste*

*Moderatore: Paola Desideri, Mitocon*

**14:45** Controllo dell'immunità innata da parte dei mitocondri e implicazioni per le malattie mitocondriali - **Marco Tigano**, Thomas Jefferson University

**15:15** Meccanismi molecolari di selezione del mtDNA: una strategia per la prevenzione delle malattie - **Patrick Chinnery**, Università di Cambridge

**15:45** Modelli in vitro derivati da cellule indotte pluripotenti (iPSCs) per lo studio di malattie associate a mutazioni del mtDNA - **Valeria Tiranti** Fondazione IRCCS Istituto Neurologico "Carlo Besta"

### **16:15 | Sessione: Poster con coffee break**

*Moderatore: Serenella Servidei, Università Cattolica del Sacro Cuore*

*Moderatore: Paola Desideri, Mitocon*

### **17:00 - 18:30 | Sessione 2 - Studi di Storia Naturale**

*Moderatore: Enrico Bertini, Ospedale Pediatrico Bambino Gesù IRCCS*

*Moderatore: Paola Desideri, Mitocon*

**17:00** Storia naturale di malattia nella Sindrome di Melas : risultato di una cooperazione internazionale - **Costanza Lamperti**, Fondazione IRCCS Istituto Neurologico "Carlo Besta"

**17:30** Storia naturale delle PMM alla luce dell'esperienza dei clinical trial

**Serenella Servidei**, Università Cattolica del Sacro Cuore

**18:00** Progressi nella storia naturale della POLG - **Omar Hikmat**, Haukeland University Hospital

**18:30 | Fine dei lavori giorno 1**

## **GIORNO 2 | SABATO 26 | SALA LUNA**

**8:30 | Arrivo e Registrazione**

**8:45 - 10:15 | Sessione 3 - Progressi nelle Terapie**

Moderatore: **Anna Ardisson**, Fondazione IRCCS Istituto Neurologico "Carlo Besta"

Moderatore: **Piero Santantonio**, Mitocon

**8:45** Diagnosi e terapia nella sindrome da deplezione del DNA mitocondriale  
**Caterina Garone**, Università di Bologna

**9:15** Terapia basata su Rapamicina come opzione terapeutica promettente per la MERRF - **Alessandra Maresca**, Università di Bologna

**9:45** OpantimiRs, una serie di antagoMiR specifici per Opa1, migliorano il fenotipo di modelli di miopatie mitocondriali" - **Luca Scorrano**, Università di Padova

**10:15 | Pausa caffè**

**10:30 - 11:30 | Sessione Giovani Ricercatori**

Moderatore: **Valeria Tiranti**, Fondazione IRCCS Istituto Neurologico "Carlo Besta"

Moderatore: **Piero Santantonio**, Mitocon

**10:30** Meccanismi molecolari patologici alla base della perdita della funzione di COA8, **Suleva Povea Cabello**, Università di Padova

**10:45** I corpi chetonici ripristinano la respirazione mitocondriale nei progenitori neuronali derivati da iPSCs di pazienti affetti da deficienza di AGC1- **Simona Nicole Barile**, CNR Bari

**11:00** Composti peptidomimetici e il loro potenziale utilizzo per il trattamento di sindromi causate da mutazioni di tRNA mitocondriali - **Maria Gemma Pignataro**, Università La Sapienza di Roma

**11:15** Modulazione del MicroRNA-181a/b come possibile strategia terapeutica per l'Atrofia Ottica Autosomica Dominante (ADOA)

**Rosa Saurino**, Tigem

**11:30 - 13:15 | Tavola Rotonda con l'Industria Farmaceutica e le Autorità Regolatorie**

Moderatori:

**Valerio Carelli**, Università di Bologna

**Michelangelo Mancuso**, Università di Pisa

**Piero Santantonio**, Mitocon

**11:30** Presentazione Industria Farmaceutica

**12:30** Tavola Rotonda con:

- **Romano Del Fiacco**, UCB
- **Chad Glasser**, Ti Sento Therapeutics
- **Magnus Hanson**, Abliva AB
- **Fabiana Saccheri**, Chiesi Farmaceutici
- **Jan Smeitink**, Khondrion
- **Magali Taiel**, Gensight Biologics
- **Toni Andreu**, EATRIS
- **Armando Magrelli**, AIFA

**13:15 | Pausa Pranzo**

**14:30 - 16:00 | Sessione 4 - Tecnologie Digitali e ruolo dell'intelligenza artificiale nella medicina mitocondriale**

Moderatore: **Massimiliano Filosto**, Università di Brescia

Moderatore: **Piero Santantonio**, Mitocon

**14:30** Biosensori e monitoraggio da remoto - **Nikita Konstantinovskiy**, Università Tecnica di Monaco

**15:00** ePROMs, APPs e coinvolgimento del paziente - **Manuela Lavorato**, Università di Pisa

**15:30** Sfruttare l'Intelligenza artificiale per supportare la diagnosi delle malattie mitocondriali: siamo pronti? - **Andrea Bandini**, Scuola Superiore Sant'Anna, Pisa

**16:00 | Pausa caffè**

### 16:10 - 16:20 | MitoAward

Moderatore: **Serenella Servidei**, Università Cattolica del Sacro Cuore

Moderatore: **Carlo Viscomi**, Università di Padova

**16:10** Flash Talk: Un approccio basato su ciclatori redox per curare le malattie mitocondriali - **Daniele Bonesso**, Università di Padova

**16:20** Flash Talk: Controllo della qualità cellulare e della funzione mitocondriale nelle neuropatie periferiche - **Paola Zanfardino**, Università di Bari

### 16:30 - 18:00 | Le ultime novità dalla ricerca sulle malattie mitocondriali

Moderatore: **Olimpia Musumeci**, Università di Messina

Moderatore: **Piero Santantonio**, Mitocon

**16:30** Generazione di modelli cellulari in 2D e 3D per lo studio dei fenotipi patologici associati a delezione del DNA mitocondriale - **Chiara Fasano**, Fondazione IRCCS Istituto Neurologico Carlo Besta, Milano

**16:50** Trapianto mitocondriale mirato a specifici tipi cellulari mediato da leganti proteici per il recupero della degenerazione cellulare - **Temurkhan Ayupov**, Istituto di Oftalmologia Molecolare e Clinica, Basel, Svizzera

**17:10** Un nuovo agonista GPCR stimola la mitofagia selettiva in un modello di carenza del Complesso III, migliorando la funzione energetica e la salute cellulare - **Cristiane Benincá**, David Geffen School of Medicine, Università della California

**17:30** Utilizzo di molecole redox-attive contro le malattie mitocondriali legate a disfunzioni dei Complessi III e I - **Ildiko Szabò**, Università di Padova

### 17:50 - 18:00 | Premiazione Mito Awards

### 18:00 - 18:15 | Osservazioni conclusive

**Carlo Viscomi**, Università di Padova

**Marco Marmotta**, Mitocon

### 18:15 | Chiusura dei lavori



## GIORNO 2 | SABATO 26 | SALA GANIMEDE

### 14:30 - 16:00 | Simposio sulle Neuropatie Ottiche Mitocondriali

Moderatore: **Valerio Carelli**, IRCCS Istituto delle Scienze Neurologiche, Bologna

Moderatore: **Paula Morandi**, Mitocon

**14:30** Diagnostica Molecolare per le Neuropatie Ottiche Mitocondriali - **Leonardo Caporali**, IRCCS Istituto delle Scienze Neurologiche, Bologna

**14:45** Farmacogenomica nella LHON - **Anna Maria Ghelli**, Università di Bologna

**15:00** Neuropatie Ottiche Mitocondriali: Manifestazioni Cliniche  
**Chiara La Morgia**, Università di Bologna

**15:15** Aggiornamento sulla storia naturale e l'efficacia dell'idebenone nella LHON - **Thomas Klopstock**, Università di Monaco

**15:30** Neuropatie Ottiche Mitocondriali: manifestazioni cliniche - **Patrick Yu-Wai-Man**, Università di Cambridge

**15:45** Domande e Risposte

**15:55** Osservazioni conclusive, **Valerio Carelli**



## PROGRAMMA PAZIENTI & FAMIGLIE



**GIORNO 3 | DOMENICA 27 | SALA EUROPA**

**9:15**

Arrivi e Registrazione Ospiti

**9:25**

**Apertura dei Lavori e Saluto di Benvenuto**

Marco Marmotta, Presidente Mitocon

**9:30 - 10:00**

**Convegno Nazionale sulle Malattie Mitocondriali: dalla ricerca di base agli approcci terapeutici, quali sono le principali novità per i pazienti**

Serenella Servidei, Policlinico Universitario A. Gemelli di Roma

**10:00 - 11:00**

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

Marco Marmotta, Presidente Mitocon

Paola Desideri e Piero Santantonio,  
Referenti Ricerca Scientifica

Cristina Rebagliati, Referente Pazienti Pediatrici

Marzia Giulia Camera, Referente Pazienti Adulti

Paula Morandi, Referente per le Persone  
con Malattia Mitocondriale della Vista

Marco Cicchelli, Rapporti con i Volontari e il Territorio

Fabrizio Farnetani, Rapporti Istituzionali

Mauro Maniero, Campagne di Raccolta fondi

**11:00** Coffee Break

**11:30 - 12:00**

**Tavola Rotonda | Tecniche di sostituzione del DNA mitocondriale**

Moderatore: Raffaele Abbatista

Daniela Zuccarello, Ospedale San Bortolo Vicenza

Piero Santantonio, Mitocon

Ignazio Zullo, Senatore, 10ª Commissione permanente (Affari sociali, sanità, lavoro pubblico e privato, previdenza sociale)

Ilenia Malavasi, Deputato, XII Commissione Affari Sociali

**12:00 - 12:30**

## Sanità Partecipata nella Regione Veneto

Moderatore: Paola Desideri

**Michele Tessarin**, Direttore Sanitario AOU Padova

**Elena Pegoraro**, Università di Padova

**Teresa Petrangolini**, Direttore Patients Advocacy Lab di Altems, Università Cattolica del Sacro Cuore Milano

**Mauro Maniero**, Referente Mitocon per la Sanità Partecipata nella Regione Veneto

**Fabrizio Farnetani**, Coordinatore del Gruppo Malattie Rare nella Regione Lazio

**12:30 - 13:00**

### La gestione dei pazienti mitocondriali in emergenza-urgenza

Moderatore: Piero Santantonio, Mitocon

Francesco Summaria, Ospedale Sant'Eugenio Roma

**13:00 - 13:30**

### Diagnosi e cura del dolore complesso nei pazienti mitocondriali pediatrici

Moderatore: Francesca Latella

Lorenzo Moscaritolo, AOU Città della Salute Torino

## CONSULENZE CON GLI ESPERTI PER PAZIENTI E FAMIGLIE

**SABATO 26 OTTOBRE, ORE 18:30**

**Enrico Bertini**, Ospedale Pediatrico Bambino Gesù, Roma

**Chiara La Morgia**, IRCCS Istituto di Scienze Neurologiche, Bologna

**Serenella Servidei**, Università Cattolica del Sacro Cuore, Roma

**Marzia Giulia Camera**, Biologa Nutrizionista Mitocon

**DOMENICA 27 OTTOBRE ORE 9:00**

**Daniela Zuccarello**, Ospedale San Bortolo Vicenza



## SCIENTIFIC PROGRAMME COMMITTEE & FACULTY



### SCIENTIFIC PROGRAMME COMMITTEE

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