

# Mitochondrial Medicine. The GENOMIT initiative

Mitochondrial disorders are now considered the most common forms of inherited human disease. In the recent years, amazing progress has been obtained in terms of pathophysiology, natural history and new treatments options for these diseases.

This conference, based on the experience of the GENOMIT consortium, funded by the European Joint Programme on rare diseases, aims to bring different international groups together, exploring the most recent advances in term of natural histories, artificial intelligence, clinical trial readiness and emerging treatment options in preclinical and clinical models of mitochondrial diseases.

with the unconditional support of

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Blueprint Genetics



**REGISTRATIONS**  
Students - Residents - Post Doc  
E-mit members  
NON E-mit members  
PHARMA and STAKEHOLDERS

until 01/09/2023  
FREE  
FREE €  
100,00 €  
150,00 €

from 02/09/2023  
FREE  
50,00 €  
150,00 €  
250,00 €

Registration link: [genomit.myquadra.it](http://genomit.myquadra.it)

Registrations fees include: Access to the conference, Coffee breaks, congress KIT and certificate of attendance. Dinner not included, VAT included  
The event is for maximum 70 participants and once this number of participants will be reached, registrations will be closed.

#### Course Venues

Aula Magna, Scuola Superiore Sant'Anna  
Piazza Martiri della Libertà 33  
56127 – Pisa – Italy

#### Patronages



**European Reference Network**  
for rare or low prevalence complex diseases

• **Network**  
Neuromuscular Diseases (ERN EURO-NMD)

• **Coordinator**  
Assistance Publique - Hôpitaux de Paris, Hôpital Pitié-Salpêtrière – France



Comune di Pisa



Organising secretariat



First Class - Meetings and Conferences  
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# Mitochondrial Medicine. The GENOMIT initiative

Pisa, November 17 - 18, 2023  
Aula Magna, Scuola Superiore Sant'Anna



# Mitochondrial Medicine. The GENOMIT *initiative*

Pisa, November 17 - 18, 2023

## Scientific Committee:

Prof. Michelangelo Mancuso  
Prof. Gabriele Siciliano  
Prof. Silvestro Micera  
Dr.ssa Giulia Ricci  
Dr. Vincenzo Montano  
Dr. Piervito Lopriore  
Dr.ssa Adriana Meli

## Faculty:

Andrea Bandini  
Valerio Carelli  
Patrick Chinnery  
Fang Fang  
Amel Karaa  
Thomas Klopstock  
Costanza Lamperti

Serena Massucci  
Robert McFarland  
Kei Murayama  
Yi Ng  
Holger Prokisch  
Carlo Viscomi

## November 17<sup>th</sup>

**13.30** Registration

**14.00** Greetings and welcome

**14.30 - 17.30 Session 1. The GENOMIT Journey.**

Chairs: *Holger Prokisch (Munich), Costanza Lamperti (Milan)*

**14.30** The global registry on mitochondrial diseases. Where we stand.  
*Thomas Klopstock (Munich)*

**15.00** Advances in Multi-Omics Diagnostics.  
*Holger Prokisch (Munich)*

**15.30** Advances in natural history studies. Focus on MELAS  
*Costanza Lamperti (Milan)*

**16.00** Advances in natural history studies. Focus on MERRF  
*Yi Ng (Newcastle)*

**16.30** Advances in natural history studies. Focus on LHON and its treatment  
*Valerio Carelli (Bologna)*

**17.00** Advances in natural history studies. Focus on Leigh Syndrome  
*Robert McFarland (Newcastle)*

**17.30** Q&A

**18.00** Closing day 1

## November 18<sup>th</sup>

**08.30 - 10.15 Challenging the future of mitochondrial medicine**

Chairs: *Patrick Chinnery (Cambridge), Michelangelo Mancuso (Pisa)*

**08.30** The role of mtDNA variation in common diseases.  
*Patrick Chinnery (Cambridge)*

**09.00** The role of AI in mitochondrial medicine.  
*Andrea Bandini (Pisa)*

**09.30** The global approach to mitochondrial medicine through GENOMIT  
*Fang Fang (China)*

**10.00** Q&A

**10.15 - 10.30** Break

**10.30 - 12.00 The new era of clinical trial readiness**

Chairs: *Amel Karaa (Boston), Kei Murayama (Tokio)*

**10.30** Exploring new treatments for MELAS. The experience of the Japanese Network  
*Kei Murayama (Tokio)*

**11.00** Selection, outcome measures and endpoints in PMDs clinical trials  
*Amel Karaa (Boston)*

**11.30** Developing Gene therapy for mitochondrial diseases  
*Carlo Viscomi (Padova)*

**12.00 - 13.00 The voice of the industries:**

**13.00** The patients 'perspective.  
*Serena Massucci (MITOCON, Italy)*

**13.30** Q&A

**14.00** Closing the meeting