

Sept•[15•17] NICE Acropolis FRANCE

mitoNice Presidents:

Véronique Paquis Valerio Carelli

Scientific Committee Chairman: Vincent Procaccio

mitoNice2022.org

PROGRAMME



mitochondrial medicine Meeting ICONICE 2022 2022 Sept-[15-17]

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> 2 <

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> 3 <

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mitoNice 2022



Laurence Tiennot-Herment President of AFM-Téléthon

Because mitochondria are the energy factories for all of our cells, when they malfunction it leads to a variety of diseases, affecting the muscle but also the vision, the brain, the metabolism and the immune system. They make up an entire universe with their own DNA, their biological complexity and their multiple symptoms. Despite this, or because they represent an exciting challenge, it is also a field of research that is rapidly evolving, with the emergence of the first innovative therapies for certain pathologies.

For several decades now, AFM-Telethon has been committed to these rare, or even ultra-rare diseases, supporting several hundreds of research programs, driving the creation of international scientific networks devoted to this very specific field in biology. Since a medical revolution is underway and mitochondrial diseases must be at the forefront of this movement, this year we have decided to organize mitoNice, a major international meeting for mitochondrial medicine, at the same time as our Myology 2022 meeting.

We hope that the communications and discussions that take place here in Nice, between the world's top experts in the field, will help speed up progress to help patients and their families. On their behalf, I would like to thank you for being here for these two days, and wish you a successful mitcoNice 2022 meeting.



> 5 <



Véronique Paquis President of mitoNice 2022

It is a great pleasure to welcome you to the AFM congress, MitoNice2022. Mitochondria are increasingly becoming a major player in Health, Disease and Ageing. These new fields of investigation are illustrated by the sessions of this international congress devoted to immunity, organoids and stem cells as new study models, or neurodegeneration. In the field of mitochondrial diseases, if the search for the responsible gene has long mobilised all energies, innovative therapies are becoming a reality for patients. The session on current clinical trials will highlight the progress made. It will bring together the best experts in the field, reminding us of the importance of interaction between researchers, doctors and patients to achieve an effective therapeutic strategy.

After the COVID crisis, this international congress will be an opportunity for the best international specialists to meet again to discuss these issues. Nice is a city that has inspired many artists but it is also home to the Côte d'Azur University, which is one of France's top ten research-intensive universities (Initiative of Excellence). Between the Alps and the Mediterranean, Nice has all the assets to make these meetings unforgettable scientific and convivial moments.



Valerio Carelli President of mitoNice 2022

I am delighted to see with MitoNice 2022, the mito-community gathering together in person again. Mitochondrial biology and medicine, in over 30 years, invaded any field of human health and pathology, with new topics in rapid expansion such as immunity or stem cell science and new technologies such as multi-omics, single cell analysis and organoids. This meeting, tightly connected with the Myology meeting, will bridge in a transdisciplinary fashion, from basic science to clinical practice, many different communities of scientists, with a special attention to the young generations. We are now entering an exciting transition from gene identification to therapies, including gene editing. A careful overview of the recent cutting-edge discoveries, from preclinical models to clinical trials with patients, will be an opportunity to connect also the patient's community with scientists and clinicians.

Overall, this meeting represents a re-start of personal connections and knowledge, to overcome the difficult times we lived, and we are still facing.

What a better place for a nice meeting than Nice, welcome to MitoNice and enjoy.

AFM-TÉLÉTHON Cure through innovation

> 6 <

Since it was created in 1958, the AFM-Téléthon has been fighting continuously against the disease driven by one objective: to cure neuromuscular diseases. From fundamental research to development of innovative therapies, our association conducts a strategy of innovation which benefits rare diseases at large.

Today, clinical trials are multiplying across the world and with the arrival of the first innovative therapy drugs, a new medicine is emerging. The impossible comes possible! The AFM-Téléthon is more determined than ever to tackle this new era and its challenges, by continuing to favour daring and innovation, therapeutic efficacy and access to the drug for all patients.

To accelerate the development of innovative therapies, the AFM-Téléthon has acquired a unique strike force:

> 3 laboratories at the forefront of innovative therapies, combined within the Biotherapies Institute for Rare Diseases:

- the **Institute of Myology** dedicated to muscles and their diseases, in particular with I-Motion, its children's and adults' clinical trials platform
- Genethon dedicated to gene therapy for rare diseases
- **I-Stem**, dedicated to research on stem cells and their therapeutic applications

> founding member of the Foundation for Rare Diseases and Imagine Institute in Paris

> **200 research and medical projects and young researchers** supported each year through its annual call for proposals

> support to 41 current and upcoming clinical trials for diseases of muscles, vision, brain, heart, skin, liver, blood, etc.

> an **'innovative biotherapies and rare diseases'** seed fund created with Bpifrance: 7 start-up companies financed

As part of its strategy for 'Cure', AFM-Téléthon relies on its Scientific Council, chaired by Prof Odile Boespflug-Tanguy, geneticist and pediatrician, Inserm research director and Head of the Neuropaediatrics department (Robert Debré Hospital, Paris), comprising 85 international scientists, doctors and experts and a global network of 6,000 experts.

Beyond its annual call for projects, the AFM-Téléthon offers its support to numerous accredited strategic projects. From proof of concept to implementing clinical protocols, specialists in basic and preclinical research, experts in clinical development and regulatory affairs can support you throughout such projects.

→ If you have a project, the AFM-Téléthon can support you.

Contact: spcsafm@afm-telethon.fr

afm-telethon.com

Programme at a glance

	Morning	Noon	Afternoon	Evening
Thursday 15 September			15:00-17:00: Plenary lecture 17:30-18:00: Handover ceremony: From Myology 2022 to mitoNice 18:00-19:00: Symposium	19:00-20:00: Poster Session 1
Friday 16 September	8:30-10:00: Plenary 1 10:30-12:00: Plenary 2	12:00-13:00: Symposium 13:00-14:00: Lunch break and exhibit	14:00-15:30: Plenary 1 16:00-17:30: Plenary 2 17:30-19:00: E-MIT meeting	19:00-20:00: Poster Session 2 20:00: Omega Networking event at the conference center
Saturday 17 September	8:30-10:00: Plenary 1 10:30-12:15: Plenary 2	12:15: Best Young investigator Poster Award 12:30: Concluding remarks		

> 8 <

PROGRAMME

Thursday, September 15th

15:00-17:00: Plenary lecture >>> Mitochondria and muscle // APOLLON AUDITORIUM

Moderators: Olivier Baris (Angers, France), Holger Prokisch (Munich, Germany) & Rita Horvath (Cambridge, UK)

- > 15:00 Mechanisms of mitochondrial myopathy: mosaic stress responses, metabolic remodeling and curable NADdeficiency Anu Suomalainen (Helsinki, Finland)
- > 15:45 Regulation of skeletal muscle bioenergetics through mitochondrial calcium during exercise and aging Jerôme Feige (Lausanne, Switzerland)
- > 16:15 Structural heterogeneity of the human respiratory chain Cristina Ugalde (Madrid, Spain)
- > 16:30 OMA1 mediates local and global stress responses against protein misfolding in CHCHD10 mitochondrial myopathy Derek Narendra (Bethesda, USA)
- > 16:45 Efficacy and genetic safety of deoxyribonucleosides as a therapy for mitochondrial DNA replication defects caused by mutations in genes involved in mtDNA maintenance Javier Ramón (Barcelona, Spain)



17:00-17:30 >>> Coffee break // AGORA 2

17:30-18:00: Plenary lecture >>> Handover ceremony: From Myology 2022 to mitoNice // APOLLON AUDITORIUM

AFM-Téléthon President: Laurence Tiennot-Herment Myology Presidents: Elizabeth McNally (Chicago, USA) & Shahragim Tajbakhsh (Paris, France) mitoNice Presidents: Véronique Paquis (Nice,France) & Valerio Carelli (Bologna, Italy)

18:00-19:00: Symposium >>> Development of deoxynucleoside therapy for thymidine kinase 2 deficiency (TK2d) // APOLLON AUDITORIUM

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Moderator: Michio Hirano (New-York, USA)

- Clinical overview and Natural History Spectrum of TK2d
 Cataring Carona (Palagna, Italy)
- Caterina Garone (Bologna, Italy)
- Pharmacological and Gene Therapy in TK2 mutant mice

Carlos Lopez-Gomez (Malaga, Spain)

- Contributions of a Spanish Network to TK2d diagnosis and therapy *Cristina Dominguez-Gonzales (Madrid, Spain)*
- Expanded Access Deoxynucleos(t)ide TK2d Therapy at a US Site Michio Hirano (New-York, USA)

19:00-20:00: Poster Session 1 // RHODES ROOM

All conferences will take place in the ATHENA AUDITORIUM

Friday, September 16th

8:30-10:00 >>> Plenary 1 >>> Innate immunity and mitochondria

Moderators: Agnes Rotig (Paris, France) & Vincent Procaccio (Angers, France)

- >> 8:30 The integrated stress response in mitochondrial cardiomyopathy Thomas Langer (Cologne, Germany)
- >> 9:00 Immune mediated disease pathogenesis in Leigh syndrome Simon Johnson (Seattle, USA)
- >> 9:30 Nuclear sensing of breaks in mitochondrial DNA enhances immune surveillance Marco Tigano (Philadelphia, USA)
- >> 9:45 Cell lineage-specific distribution of pathogenic mtDNA mutations in the immune system and purify selection during the adaptive immune response Jingdian Zhang (Stockholm, Sweden)

10:00-10:30 >>> Coffee break and Visit of the exhibit hall and poster room // RHODES ROOM

10:30-12:00 >>> Plenary 2 >>> iPS cells and organoids as alternatives to mouse models?

Moderators: Valeria Tiranti (Milan, Italy) & Maria Falkenberg (Mölndal, Sweden)

- >> 10:30 Modelling OPA1 Dysfunction in iPSC-derived Retinal Ganglion Cells Joshua Harvey (Cambridge, UK)
- >> 11:00 Drug discovery of mitochondrial diseases using patient-specific brain organoids Alessandro Prigione (Dusseldorf, Germany)
- >> 11:30 PKAN stem cell derived neurons and astrocytes show massive iron accumulation mimicking the human phenotype Sonia Levi (Milan, Italy)
- >> 11:45 Mitochondrial regulation of neural stem cell ageing in progressive multiple sclerosis Rosana-Bristena Ionescu (Cambridge, UK)

12:00-13:00 >>> Symposium >>> Optic neuropathies: From NOHL to DOA

Moderator: Robert Pitceathly (London, UK)

- >> 12:00 Autosomal dominant optic atrophy Guy Lenaers (Angers, France)
- >> 12:30 Leber's hereditary optic atrophies Patrick Yu Wai-Man (Cambridge, UK)

13:00-14:00

Lunch break and Visit of the exhibit hall and poster room // RHODES ROOM

14:00-15:30 >>> Plenary 1 >>> Neurodegeneration and mitochondria

Moderators: Timothy Wai (Paris, France) & Antonio Zorzano (Barcelona, Spain)

- >> 14:00 CHCHD10-related neurodegeneration: from gene to therapeutic approaches Veronique Paquis (Nice, France)
- >> 14:30 Metabolic rewiring precedes OXPHOS dysfunction in a mutant CHCHD10 mouse model Giovanni Manfredi (New York, USA)
- >> 15:00 Mitochondrial dysfunction and Ca2+ dysregulation in COQ8A-Ataxia Purkinje neurons that can be rescued by CoQ10 treatment Hélène Puccio (Lyon, France)
- >> 15:15 Bi-allelic variants in TAMM41 are associated with low muscle cardiolipin levels leading to neonatal mitochondrial disease Robert Taylor (Newcastle, UK)

15:30-16:00 >>> Coffee break and Visit of the exhibit hall and poster room // RHODES ROOM

16:00-17:30 >>> Plenary 2 >>> New models for genotype/phenotype correlation

Moderators: Shamima Rahman (London, UK) & Robert McFarland (Newcastle, UK)

- >> 16:00 Mechanisms and logic of mitochondrial metabolite signaling Edward Chouchani (Boston, USA)
- >> 16:30 Modelling phenotypes of mitochondrial translation defects in vitro and in vivo Rita Horvath (Cambridge, UK)
- >> 17:00 Mitochondrial genome engineering in vivo Michal Minczuk (Cambridge, UK)
- >> 17:15 The first in human clinical application of autologous mesoangioblasts as cell therapy medical product in m.3243A>G mutation carriers Florence Van Tienen (Maastricht, The Nederlands)

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17:30-18:00 >>> Coffee break and Visit of the exhibit hall and poster room // RHODES ROOM

17:30-19:00 >>> Plenary 3 >>> E-MIT meeting: European Mitochondrial Society

Moderators : Valerio Carelli (Bologna, Italy) & Valeria Tiranti (Milan, Italy)

>> **17:30** • Introductory talk Massimo Zeviani (Milan, Italy)

· General discussion & the voice of patient representatives



at the Nice Acropolis Center (Rhodes Room) All conferences will take place in the ATHENA AUDITORIUM

Saturday, September 17th

8:30-10:00 >>> Plenary >>> New treatments for mitochondrial diseases: update on recent and current clinical trials

Moderators: Patrick Chinnery (Cambridge, UK) & Thomas Klopstock (Munich, Germany)

- >> 8:30 Clinical trials landscape in primary mitochondrial diseases in the USA: looking into the future Amel Karaa (Boston, USA)
- » 8:45 Sonlicromanol in primary mitochondrial disease MELAS spectrum disorders Jan Smeitink (Nijmegen, The Netherlands)
- >> 9:00 Mitochondrial disease clinical trials: experience gained at the London highly specialised service for rare mitochondrial disorders Robert Pitcaethly (London, UK)
- >> 9:15 Elamipretide treatment and Barth syndrome: combined data from a phase 2/3 clinical trial and a natural history comparison study *Hilary Vernon (Baltimore, USA)*
- >> 9:30 General discussion between the panel and the audience

10:00-10:30 >>> Coffee break and Visit of the exhibit hall and poster room // RHODES ROOM

10:30-12:15 >>> Flash session with hot topics

Moderators: Giovanni Manfredi (New-York, USA) & Anu Suomalainen (Helsinki, Finland)

- >> 10:30 Adapting DNA editing enzymes to treat mitochondrial disease Carlos Moraes (Miami, USA)
- >> 10:45 Inhibition of mtDNA transcription has beneficial effects on metabolism Nils Larsson (Stockholm, Sweden)
- >> 11:00 About neural stem cell trafficking and secretion of functional mitochondria via extracellular vesicles Stefano Pluchino (Cambridge, Uk)
- >> 11:15 Targeting metabolism to purge mutant mitochondrial DNAs Antonella Spinazzola (London, UK)
- >> 11:30 Genetic variants affecting NQO1 protein levels impact on efficacy of idebenone treatment in Leber's hereditary optic neuropathy Valerio Carelli (Bologna, Italy)
- >> 11:45 Neuroglobin overexpression in cerebellar neurons of Harlequin mice improves mitochondrial robustness and reduces ataxic behavior Marisol Corral-Debrinski (Paris, France)
- >> 12:00 Double administration of self-complementary AAV9NDUFS4 prevents Leigh disease in Ndufs4-/- mice Carlo Viscomi (Padova, Italy)

12:15 >>> Best Young investigator Poster Award

12:30 >>> Concluding remarks

Speakers: Valerio Carelli (Bologna, Italy) & Valeria Tiranti (Milan, Italy), Vincent Procaccio (Angers, France), Véronique Paquis (Nice, France), Laurence Tiennot-Herment (President of AFM-Téléthon)

> 12 <

Discover Nice, Capital of the French Riviera

On the Mediterranean coast, only a few kilometres from Italy, Nice has all the charm of a southern city. Even before you land on the Nice tarmac, you will be breathless by the view that awaits you through the window. No wonder Nice Côte d'Azur International Airport has been on the podium of the most beautiful approaches in the world for several years in a row! In addition to the extraordinary monochrome of the Mediterranean Sea, the panorama of the mountains (snow-covered in winter) is also awe-inspiring.

Capital of the French Riviera, Nice is the perfect embodiment of the art of living. The blue of the Mediterranean, the marvellous light that can only be found here, breakfast on a sunny terrace, a quiet stroll through the narrow streets of the old town, lunch on a hotel terrace, an afternoon of shopping in the city's prestigious boutiques, a dip in the sea, an aperitif on the Cours Saleya, a classical music concert at the Opera, a gala evening at the Casino, a candlelit dinner in a Michelin-starred restaurant...Nice is all of these things and more!

Old Nice is, in itself, a festival of sensations! Narrow streets, yellow and ochre facades, craftsmen's shops and small boutiques, restaurants and lively squares make up the soul of this old quarter, which is alive day and night, not forgetting, of course, the Cours Saleya market, classified as one of the exceptional markets in France! From the top of the listed site of the Colline du Château, you can enjoy a remarkable view of the Baie des Anges, the old town and the port, from sunrise to sunset.

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> 13 <



POSTERS

	=	P32	P31	P30	P29
	⊒	471 440	425	424 401	400 381
P28/ Clinical trials in mitochondrial diseases and	-	470 441	439 426	423 402	399 382
From Nº381 to Nº394	2	469 442	438 427	422 403	398 383
		468 443	437 428	421 404	397 384
P29/ Innate immunity and mitochondria		466 445	435 430	419 406	395 386
From N°395 to N°400	465 446 434 431 418 407 394 387				
P20 / iPSC and organoids: alternatives to mouse models	2	464 447	433 432	417 408	393 388
From Nº401 to Nº408	1	463 448	P31	416 409	392 389
		462 449		415 410	391 390
P31/ Muscle and mitochondria (overlap with Myology)	_	461 450		414 411 °	P28
From Nº409 to Nº439	<u>i</u>	460 451	P32	413 412	
D22 / Nourodogonarction and mitachandria		409 402	472	P31	
From N $^{\circ}440$ to N $^{\circ}474$		457 454	474 473		
		456 455			
		► P32	/		
			<u> </u>		

> 14 <

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A STREET



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Nice Acropolis Convention Centre

1 esplanade John Fitzgerald Kennedy 06000 Nice

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• From the airport: Bus n° 98 - Stop: Cathédrale Vieille-Ville, and the tramway direction Pasteur - Stop: Acropolis.	• From the SNCF Station: The tramway at the stop Thiers direction Pasteur - Stop: Acropolis.	 Palais des Congrès: 43°42'21.4"N 7°16'52.1"E Parkings near the Nice Acropolis: Parking Acropolis [<i>Parking Jean Bouin</i>] Parking Promenade des Arts [<i>Parking Promenade des Arts</i>]



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