

Sunday, June 11

- 15:00 Registration open
- 16:30-17:00 Opening ceremony and welcome from the Euromit co-chairs
- 17:00-18:30 **Session I - Cell biology, genetic and clinical perspective on mitochondria**
- Chair: Jan Smeitink, Radboud University, Nijmegen, Netherlands
- 17:00-17:30 Jodi Nunnari, University of California, Davis, USA: *The behavior of mitochondria*
- 17:30-18:00 Nils-Göran Larsson, Max Planck Institute for Biology of Ageing, Cologne, Germany: *The role of mtDNA copy number in mtDNA mutation disease*
- 18:00-18:30 Patrick Chinnery, University of Cambridge, United Kingdom: *Mitochondrial DNA inheritance and the bottleneck*
- 19:00-22:00 Opening Reception at Früh am Dom

Monday, June 12

- 08:45-10:15 **Session II - Diseases and Diagnosis**
- Chairs: Holger Prokisch, Technical University Munich, Germany  
Marni Falk, The Children's Hospital of Philadelphia, USA
- 08:45-09:15 David Thorburn, University of Melbourne, Australia: *Exome sequencing: how good are we and how can we increase diagnostic yield?*
- 09:15-09:45 Doug Turnbull, Newcastle University, United Kingdom: *New insights into adult mitochondrial disease*
- 09:45-10:00 Metodi Metodiev, Institut Imagine, Paris, France: *Biallelic mutations in the mitochondrial ribosomal protein MRPS2 cause sensorineural hearing loss, lactic acidemia, hypoglycemia and multiple OXPHOS complex deficiencies*
- 10:00-10:15 Charlotte Alston, Newcastle University, United Kingdom: *Biallelic mutations in NDUFA6 establish its role in early-onset mitochondrial complex I deficiency*
- 10:15-10:45 Coffee Break
- 10:45-12:45 **Session III - Mitochondrial DNA: replication, gene expression, and translation**
- Chairs: Jim Stewart, Max Planck Institute for Biology of Ageing, Cologne, Germany  
Maria Sola, Spanish National Research Council, Barcelona, Spain

- 10:45-11:15 **PLoS Genetics Lecture:** Maria Falkenberg, University of Gothenburg, Sweden: *Human mitochondrial DNA replication: deletion formation*
- 11:15-11:45 Aleksandra Filipovska, University of Western Australia, Perth, Australia: *Mitochondrial RNA-binding proteins in health and disease.*
- 11:45-12:15 Zofia Chrzanowska-Lightowlers, Newcastle University, United Kingdom: *Human mitoribosomes – what makes them stop?*
- 12:15-12:30 Sofia Zaganelli, University of Geneva, Switzerland: *The pseudouridine synthase RPUUSD4 plays a pivotal role in mitochondrial gene expression*
- 12:30-12:45 Dusanka Milenkovic, Max-Planck Institute for Biology of Ageing, Cologne, Germany: *The in vivo role of the mitochondrial DNA nuclease MGME1*
- 12:45-13:45 Lunch Break
- 12:45-14:00 *O2k-Workshop on high-resolution respirometry and O2k-Fluorometry*
- 13:45-16:30 **Poster Session A**
- 16:30-17:00 Coffee Break
- 17:00-19:00 **Session IV - Mechanisms of mitochondrial diseases**
- Chairs: Agnès Rötig, Institut Imagine, Paris, France  
Lawrence Bindoff, University of Bergen, Norway
- 17:00-17:30 Valeria Tiranti, Neurological Institute C. Besta, Milan, Italy: *Experimental models in mitochondria related neuromuscular disorders: from pathogenic mechanism to therapeutic approaches*
- 17:30-18:00 Ramon Marti, Vall d’Hebron Research Institute, Barcelona, Spain: *Nucleotide metabolism and mitochondrial DNA maintenance diseases*
- 18:00-18:30 Massimo Zeviani, University of Cambridge, United Kingdom: *New players and mechanisms in mitochondrial biogenesis and disease*
- 18:30-18:45 Romain Cartoni, Boston Children's Hospital and Harvard Medical School, Boston, USA: *The mammalian specific protein Armcx1 regulates mitochondrial transport during axon regeneration*
- 18:45-19:00 Ana Peñas, FIB Hospital 12 de Octubre, Madrid, Spain: *Functional involvement of the actin-binding protein gelsolin in respiratory chain dysfunction: A putative biomarker for Oxphos disorders*

## Tuesday, June 13

- 09:00-10:30 **Session V - Mitochondria biochemistry and diseases**

- Chairs: Jan Riemer, University of Cologne, Germany  
Cristina Ugalde, FIB Hospital 12 de Octubre, Madrid, Spain
- 09:00-09:30 Uli Brandt, Radboud University of Nijmegen, Netherlands: *The mechanism and regulation of the forward and reverse reaction of mitochondrial complex I*
- 09:30-10:00 Eric Shoubbridge, McGill University, Montreal, Canada: *Exploring the mitochondrial-ER interactome*
- 10:00-10:15 Karolina Szczepanowska, University of Cologne, Germany: *CLPP is a modulator of OXPHOS biogenesis*
- 10:15-10:30 Abhishek Aich, University Medical Center Göttingen, Germany: *At the crossroads of cytochrome c oxidase assembly*
- 10:30-11:00 Coffee Break
- 11:00-12:30 **Session VI - Mitochondrial communications**
- Chairs: Mike Ryan, Monash University, Melbourne, Australia  
Guy Lenaers, University of Angers, France
- 11:00-11:30 Luca Scorrano, University of Padua, Italy: *A localized autophagic filter depletes axons from damaged mitochondria*
- 11:30-12:00 Navdeep Chandel, Northwestern University, Chicago, USA: *Mitochondria as signaling organelles*
- 12:00-12:15 Atan Gross, Weizmann Institute of Science, Rehovot, Israel: *The role of mitochondrial MTCH2 in health and disease*
- 12:15-12:30 David Pla Martín, University of Cologne, Germany: *CLUH inhibits mTORC1 and degradation of its target mRNAs by mediating stress granule assembly during starvation*
- 12:30-13.30 Lunch Break
- 13.30-16:30 **Poster Session B**
- 16:30-17:00 Coffee Break
- 17:00-19:15 **Session VII - Mitochondrial and stress responses**
- Chairs: Elena Rugarli, University of Cologne, Germany  
Brendan Battersby, University of Helsinki, Finland
- 17:00-17:45 **EMBO Keynote Lecture:** Anu Suomalainen Wartiovaara, University of Helsinki, Finland: *Integrated stress response in mitochondrial disease*
- 17:45-18:15 Thomas Langer, University of Cologne, Cologne, Germany: *Mitochondrial phospholipid metabolism and cardiomyopathies*

- 18:15-18:30 Sarah Pearce, University of Cambridge, United Kingdom: *Deadenylation specifies a human mitochondrial repair pathway for non-coding RNAs*
- 18:30-18:45 Sebastien Herzig, Salk Institute for Biological Studies, La Jolla, USA: *Regulation of mitochondrial dynamics by the energy sensor AMPK*
- 18:45-19:00 Alessia Angelin, The Children's Hospital of Philadelphia, USA: *FOXP3 increases oxidative phosphorylation and NAD oxidation, adapting regulatory T cells to low glucose high lactate environments*

### Wednesday, June 14

#### 09:00-12:45 **Session VIII - Therapeutic strategies for mitochondrial diseases - part 1**

Chairs: Michal Minczuk, University of Cambridge, United Kingdom  
Howy Jacobs, University of Helsinki, Finland

- 09:00-09:30 Carlos Moraes, University of Miami, USA: *Modulating mtDNA heteroplasmy in vivo with gene editing enzymes*
- 09:30-10:00 José Antonio Enriquez - Spanish National Centre for Cardiovascular Research, Madrid, Spain: *Are there good and bad mtDNA haplotypes?*
- 10:00-10:15 Emanuelle Sarzi, Institute for Neurosciences, Montpellier, France: *Intravitreal injection of a human AAV2- OPA1 vector prevents optic nerve alteration and retinal ganglion cell degeneration in an Opa1 +/- mouse model*
- 10:15-10:30 Jukka Kallijarvi, Folkhälsan Research Center, Helsinki, Finland: *Rescue of premature lethality and late-onset organ manifestations by alternative oxidase in a mouse model of respiratory chain complex III deficiency*
- 10:30-11:00 Coffee Break
- 11:00-11:30 Valerio Carelli, University of Bologna, Italy: *Leber`s hereditary optic neuropathy: current and evolving treatments*
- 11:30-12:00 Shoukhrat Mitalipov, Oregon Health & Science University, Portland, USA: *Somatic and germline mtDNA mutations*
- 12:00-12:30 Rong Tian, University of Washington, USA: *Elevation of intracellular NAD<sup>+</sup> levels as a therapy for mitochondria dysfunction*
- 12:30-12:45 Claudia Catarino, Ludwig-Maximilians-University, Munich, Germany: *Idebenone is effective and well tolerated in leber`s hereditary optic neuropathy (LHON): results of a 3-year expanded-access program*
- 12:45-13:45 Lunch Break

- 13.45-16:30 **Poster Session C**
- 16:30-17:00 Coffee Break
- 17:00-18:00 Patients' viewpoint on mitochondrial diseases  
Shanalle Hamilton  
Rachael Wells
- 18:00-18:15 Announcement of Travel Grants
- 19:00 EUROMIT Party in Flora

Thursday, June 15

08:45-10:45 **Session IX - Therapeutic strategies for mitochondrial diseases - part 2**

Chairs: Ann Saada, Hadassah Medical Center, Jerusalem, Israel  
Thomas Klopstock, Ludwig-Maximilians-University, Munich, Germany

09:00-09:30 Michio Hirano, Columbia University, New York, USA: *Development of a pharmacological therapy for thymidine kinase 2 deficiency*

09:30-10:00 Jan Smeitink, Radboud University Nijmegen, Netherlands: *The redox modulating small molecule KH176: from drawing table towards phase 2 clinical trial*

10:00-10:30 Mary Herbert, Newcastle University, United Kingdom: *IVF-based technologies to prevent germline transmission of mitochondrial DNA disease*

10:30-11:00 Coffee Break

11:00-12:00 **Session X - Late breaking news**

Chair: Aleksandra Trifunovic, University of Cologne, Germany

11:00-11:15 Laura Kremer, Helmholtz Center, Munich, Germany: *Genetic diagnosis of mendelian disorders using RNA sequencing*

11:15-11:30 Olesia Ignatenko, University of Helsinki, Finland: *Astrocyte vs. neuronal mitochondrial DNA dysfunction reveals strikingly different phenotypes unraveling specialized role of mitochondria in astrocytes*

11:30-11:45 Pierre Rustin, INSERM UMR1141 and Université Paris 7, France: *Mitochondria are physiologically maintained at close to 50°C*

11:45-12:00 **Concluding Remarks**