MITOCHONDRIA IN HUMAN DISEASE

September 16-18, Stockholm, Sweden

JIM Journal of Internal Medicine
Founded in 1863

Karolinska Institutet
Karolinska Institutet Anno 1810

Welcome

We welcome you to the conference **"Mitochondria in Human Disease"**. We look forward to this exciting meeting which includes international participants working in many different areas of mitochondrial biology, from human disease to basic biochemistry. Since the discovery of the first pathogenic mutations in human mtDNA in 1988, the disease field has developed at an accelerating pace. Today we know of >250 pathogenic variants in mtDNA and mutations in >300 nuclear genes have been shown to cause mitochondrial disease. Whole genome and exome sequencing are now routinely used in the diagnosis of mitochondrial disease and the identified mutations have given profound new insights into disease syndromes and biochemical pathways controlling mitochondrial function. This remarkable progress in diagnosis has been paralleled by completely novel insights into the function and organization of mitochondria. Technological advances, such as metabolomics, proteomics, cryo-electron microscopy for high resolution structures of protein complexes, electron cryo-tomography to study mitochondrial morphology and super-resolution microscopy of cells and tissues, have together with advanced biochemistry and genetics profoundly changed our insights into the biology of mitochondria. We have brought scientists representing all of these exciting areas together to stimulate transdisciplinary interactions.

We hope that you will enjoy the conference and thank you all for coming.

Nils-Göran Larsson and Anna Wedell

Professor Nils-Göran Larsson
Department of Medical Biochemistry and Biophysics (MBB), Karolinska Institutet

Professor Anna Wedell
Department of Molecular Medicine and Surgery (MMK), Karolinska Institutet

Programme

DAY 1, SEPTEMBER 16, 2019

12:00-13:30 **Lunch and registration**

13:45-14:00 **Welcome and introductory remarks**
Nils-Göran Larsson and Anna Wedell, Karolinska Institutet
Bo Angelin, Journal of Internal Medicine

Session 1: MECHANISMS IN MITOCHONDRIAL DISEASE
Chair: Nils-Göran Larsson, Karolinska Institutet, Stockholm, Sweden

14:00-14:30 **Mitochondrial disease phenotypes in children**
Shamima Rahman, Great Ormond Street Hospital, London, UK

14:30-15:00 **Mitochondrial RNA processing in disease**
Aleksandra Filipovska, University of Western Australia, Perth, Australia

15:00-15:15 **Poster talk: dNTP levels determine the frequency and identity of ribonucleotides in mitochondrial DNA**
Paulina H Wanrooij, Department of Medical Biochemistry and Biophysics, Umeå University, Umeå, Sweden

15:15-15:30 **Poster talk: Cardiac specific deletion of MTP18 induces heart failure in mice**
T Wai, Mitochondrial Biology Group, Institut Pasteur CNRS UMR 3691, Paris, France

15:30-16:00 **Coffee break**

16:00-16:30 **Mitochondrial disease and optic atrophy**
Valerio Carelli, University of Bologna, Italy

16:30-17:00 **Malfunction of the mitochondrial replisome and mtDNA mutations**
Maria Falkenberg, Göteborgs Universitet, Sweden

17:00-17:30 **Principles of human mtDNA inheritance**
Patrick Chinnery, University of Cambridge, UK

17:30- 19:30 **Dinner**

Session 2: REGULATION OF MITOCHONDRIAL FUNCTION
Chair: Anna Wedell, Karolinska Institutet, Stockholm, Sweden

19:30-20:00 **Mitochondrial import of S-adenosyl methionine and the mitochondrial methylproteome**
Anna Wredenberg, Karolinska Institutet, Sweden

20:00-20:30 **Physiological principles of mitophagy and implications for Parkinson's disease**
Ian Ganley, University of Dundee, UK

20:30-22:00 **Poster session and drinks – (Odd poster numbers)**

DAY 2, SEPTEMBER 17, 2019

Session 3: THE ARCHITECTURE OF MITOCHONDRIA
Chair: Nils-Göran Larsson, Karolinska Institutet, Sweden

09:00-09:30 **Protein complexes organizing mitochondrial compartments**
Klaus Pfanner, University of Freiburg, Germany

09:30-09:45 **Poster talk: Mutations in the mitochondrial RNA polymerase POLRMT are associated with a spectrum of clinical presentations**
Bradley Peter, Department of Medical Biochemistry and Cell Biology, University of Gothenburg, Sweden

DAY 2, SEPTEMBER 17, 2019

09:45-10:00 **Poster talk: Pathways and players in respirasome biosynthesis**
Cristina Ugalde, Instituto de Investigación, Hospital Universitario 12 de Octubre, 28041 Madrid, Spain

Centro de Investigación Biomédica en Red de Enfermedades Raras (CIBERER), U723, Madrid, Spain

10:00-10:30 **Structures of respiratory chain complexes and supercomplexes**
Judy Hirst, Mitochondrial Biology Unit, MRC, Cambridge

10:30-11:00 **Coffee break**

11:00-11:30 **Superresolution microscopy of mitochondrial organization and dynamics**
Stefan Jakobs, Max Planck Institute for Biophysical Chemistry, Göttingen, Germany

11:30-12:00 **Electron cryo-tomography of protein complexes in mitochondrial membranes**
Werner Kühlbrandt, Max Planck Institute for Biophysics, Frankfurt, Germany

12:00-13:00 **Lunch**

13:30-17:30 **Excursion with guided tour of the Nobel museum and visit of the old town "Gamla Stan" in Stockholm.**

17:30-19:30 **Dinner**

DAY 2, SEPTEMBER 17, 2019

Session 4: MITOCHONDRIAL TRANSLATION
Chair: Anna Wredenberg, Karolinska Institutet, Stockholm, Sweden

19:30-20:00 **Coordination of mitochondrial translation and respiratory chain assembly**
Martin Ott, Stockholm University, Sweden

20:00-20:15 **Poster talk: A novel mitochondrial assembly factor RTN4IP1 has an essential role in the final stages of Complex I assembly**
Monika Oláhová, Wellcome Centre for Mitochondrial Research, Newcastle University, Newcastle upon Tyne, UK

20:15-20:30 **Poster talk: Dinucleotide degradation by REXO2 is required to maintain promoter specificity in mammalian mitochondria**
Thomas J Nicholls, Department of Medical Biochemistry and Cell Biology, University of Gothenburg, Gothenburg, Sweden,

Present address: Wellcome Centre for Mitochondrial Research, Newcastle University, Newcastle upon Tyne, UK.

20:30-21:00 **Biogenesis of the mammalian mitochondrial ribosome**
Joanna Rorbach, Karolinska Institutet, Sweden

21:00-22:30 **Poster session and drinks (Even poster numbers)**

DAY 3, SEPTEMBER 18, 2019

Session 5: TREATMENT OF MITOCHONDRIAL DYSFUNCTION
Chair: Claes Gustafsson, Göteborgs Universitet, Göteborg, Sweden

09:00-09:30 **Gene therapy by enzymatic editing of mtDNA in the mouse**
Carlos Moraes, University of Miami, Miami, USA

09:30-10:00 **Cellular responses in mitochondrial disease**
Anu Suomalainen, University of Helsinki, Finland

10:00-10:15 **Poster talk: Reversible anaplerosis induced by mitochondrial dysfunction reveals metabolic plasticity in degenerating neurons**
Elisa Motori, Department of Mitochondrial Biology, Max Planck Institute for Biology of Ageing, 50931 Cologne, Germany

10:15-10:30 **Poster talk: Niacin supplementation alleviates disease symptoms in patients with mitochondrial myopathy**
Eija Pirinen, Research Program for Clinical and Molecular Metabolism, Faculty of Medicine, University of Helsinki, Helsinki, Finland

10:30-11:00 **Coffee break**

11:00-11:30 **Nuclear effects caused by mutated cytochrome c oxidase**
Ann Saada, Hadassah Medical Center, Jerusalem, Israel

11:30-12:00 **Strategies for treatment of mitochondrial disease**
Massimo Zeviani, Mitochondrial Biology Unit, MRC, Cambridge

12:00-12:30 **Reproductive options for women with mitochondrial DNA mutations**
Doug Turnbull, University of Newcastle, Newcastle, UK

12:30-14:00 **Lunch and departure**

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P1 - Mitochondrial impairment precedes lysosomal deficiency in a Parkinson's disease model of LRRI
Andreas Aufschnaiter, *Verena Kohler, Corvin Walter, F.-Nora Vögtle, Martin Ott, Sabrina Büttner*

P2 - Levels of TFAM expression affect mitochondrial function in vivo
Nina A Bonekamp, *Min Jiang, Elisa Motori, Chan Bae Park, Nils-Göran Larsson*

P3 - Reconstituting mitochondrial translation in vitro
Anneli Borg, *Diana Khananisho, Martin Ott*

P4 - Characterization of GTP-binding proteins and their role in mitochondrial ribosome assembly
M Cipullo, *S Pearce, J Rorbach*

P5 - First Recessive Variant in HECW2 Gene Causes Neurodevelopmental Disorder with Hypotonia, Seizures, and Absent Language (NDHSAL))
Francisco Javier Cotrina-Vinagre, *Laura Hernández-Sánchez, María Elena Rodríguez-García, Marcello Bellusci, Ana Martínez de Aragón, Patricia Carnicero-Rodríguez, Eduardo López-Laso, Elena Martín-Hernández, Francisco Martínez-Azorín*

P6 - Analysis of pathogenic mtDNA mutations in Serbian patients with MELAS
P.G.A Dawod, *B Rovcanin, M Brankovic, A Marjanovic, M Jankovic, I Novakovic, F.I Abdel Motaleb, J Jancic, V Kostic*

P7 - SSBP1 mutations cause a complex optic atrophy spectrum disorder with mitochondrial DNA depletion
Valentina Del Dotto, *Farid Ullah, Ivano Di Meo, Pamela Magini, Mirjana Gusic, Alessandra Maresca, Leonardo Caporali, Flavia Palombo, Francesca Tagliavini, Evan Harris Baugh, Bertil Macao, Zsolt Szilagyi, Camille Peron, Margaret A Gustafson, Chiara La Morgia, Piero Barboni, Michele Carbonelli, Maria Lucia Valentino, Rocco Liguori, Vandana Shashi, Jennifer Sullivan, Shashi Nagaraj, Mays El-Dairi, Alessandro Iannaccone, Ioana Cutcutache, Enrico Bertini, Rosalba Carrozzo, Francesco Emma, Francesca Diomedi-Camassei, Claudia Zanna, Martin Armstrong, Matthew Page, Nicholas Stong, Sylvia Boesch, Robert Kopajtich, Saskia Wortmann, Wolfgang Sperl, Erica E Davis, William C Copeland, Marco Seri, Maria Falkenberg, Holger Prokisch, Nicholas Katsanis, Valeria Tiranti, Tommaso Pippucci, Valerio Carelli*

P8 - Searching for the causes of cerebellar ataxia in mitochondrial diseases: insights from the Harlequin mouse.
M Fernández de la Torre, *C Fiuza-Luces, S Laine-Menéndez, V Bermejo-Gómez, M Morán*

P9 - Complex III is essential for complete assembly and maturation of complex I in human mitochondria
Erika Fernandez-Vizarra, *Margherita Protasoni, Rafael Perez-Perez, Teresa Lobo-Jarne, Michael E Harbour, Shujing Ding, Ana Peñas, Francisca Diaz, Carlos T Moraes, Ian M Fearnley, Massimo Zeviani, Cristina Ugalde*

P10 - Loss of Mitofusin 2 in adult dopaminergic neurons causes a premature activation of surrounding microglial cells.
Roberta Filograna, *Seungmin Lee, Oleg Shupliakov, Lars Olson, Nils-Göran Larsson*

P11 - Using the TFAM-FLAG mice to study the modulation of mtDNA expression in vivo.
Rodolfo García-Villegas, *Nina A Bonekamp, Henrik Spår, Nils-Göran Larsson*

P12 - Novel variants in genes of the mitoribosomal large subunit cause disorders of mitochondrial translation
Ruth Glasgow, *Monika Oláhová, Malak Alghamdi, Rose-Mary Boustany, Rolla Shbarou, Vinod Misra, Mitchell Cunningham, Saskia Wortmann, Holger Prokisch, Robert McFarland, Robert Taylor*

P13 - C6orf203 is an RNA-binding protein involved in mitochondrial protein synthesis
Shreekara Gopalakrishna, *Sarah F Pearce, Adam M Dinan, Florian A Schober, Miriam Cipullo, Henrik Spåhr, Anas Khawaja, Camilla Maffezzini, Christoph Freyer, Anna Wredenberg, Ilian Atanassov, Andrew E Firth, Joanna Rorbach*

P14 - Deep sequencing of mitochondrial DNA and functional characterization of a novel POLG mutation in a patient with autosomal recessive progressive external ophthalmoplegia
Carola Hedberg-Oldfors, *Bertil Macao, Swaraj Basu, Jay P Uhler, Direnis Erdinc, Bradley Peter, Christopher Lindberg, Erik Larsson, Maria Falkenberg, Anders Oldfors*

P15 - Structural basis of mitochondrial transcription
Hauke S Hillen, *Yaroslav I Morozov, Azadeh Sarfallah, Andrey V Parshin, Karen Agaronyan, James J Graber, Aleksandar Chernev, Kathrin Schwinghammer, Henning Urlaub, Dmitry Temiakov, Patrick Cramer*

P16 - More than mitochondrial fusion: Role of Mitofusin 2 in cellular stress response
Selver Altin, ***Mariana Joaquim,*** *Mafalda Escobar-Henriques*

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P19 - Three rare LHON pathogenic substitutions in patients with low mutation load
Tatiana D Krylova, *Natalia L Sheremet, Vyacheslav Yu Tabakov, Konstantin G Lyamzaev, Yulia S Itkis, Polina G Tsygankova, Natalia L Andreeva, Maria S Shmelkova, Tatiana A Nevinitsyna, Vitaly V Kadyshev, Ekaterina Yu Zakharova*

P20 - Impact of Apoptosis Inducing Factor deficiency on Harlequin mouse heart
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P21 - DNMT1 mutations leading to neurodegeneration paradoxically reflect on mitochondrial metabolism
Alessandra Maresca, *Valentina Del Dotto, Mariantonietta Capristo, Emanuela Scimonelli, Francesca Tagliavini, Luca Morandi, Leonardo Caporali, Susan Mohamed, Marina Roberti, Letizia Scandiffio, Mirko Zaffagnini, Jacopo Rossi, Martina Cappelletti, Francesco Musiani, Manuela Contin, Roberto Riva, Rocco Liguori, Fabio Pizza, Chiara La Morgia, Elena Antelmi, Paola Loguercio Polosa, Emmanuel Mignot, Claudia Zanna, Giuseppe Plazzi, Valerio Carelli*

P22 - De novo serine synthesis is protective in mitochondrial myopathy
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Bradley Peter, *Monika Oláhová, Hector Diaz, Zsolt Szilagyi, Maria Falkenberg, Claes Gustafsson, Robert W Taylor*

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Majda Mehmedovic, *Géraldine Farge, Henrik Spåhr, Maria Falkenberg*

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