Patient Forum
Saturday 26 November

Session 1

9:30 am  Mitochondrial Disease: The one stop shop
         Professor Carolyn Sue
         University of Sydney and Royal North Shore Hospital, Sydney

9:50 am  TBA
         Dr Grainne Gorman
         University of Newcastle, Newcastle Upon Tyne, United Kingdom

10:10    MORNING TEA

Session 2

10:40 am Mitochondrial Disease Drug Development: the Saga of KH176
         Professor Jan Smeitink
         Radboud University, Nijmegen, The Netherlands

11:00 am Eeny, meeny, miny, mo… on which supplement/s should we spend the dough?
         Dr Christina Liang
         Royal North Shore Hospital, Sydney, Australia

11:20 am Panel Discussion: Recent advances in mitochondrial medicine

12:00 pm CLOSE
Young Investigators Symposium  
Saturday 26 November

Session 1

8:00 am  
WELCOME

8:05 am  
A successful scientific career  
Professor Eric Shoubridge  
McGill University, Montreal, Canada

8:30 am  
Transcriptome-wide analyses of mitochondrial gene expression in models of disease  
Stefan Siira  
Harry Perkins Institute of Medical Research and University of Western Australia, Perth

8:45 am  
Tim29 is a novel subunit of the human TIM22 translocase and is involved in complex assembly and stability  
Yilin Kang  
University of Melbourne, Melbourne

9:00 am  
Determining the genetic basis of mitochondrial complex I deficiency using massively parallel sequencing  
Shalini Thirukeswaran  
Murdoch Childrens Research Institute and University of Melbourne, Melbourne

9:15 am  
Enhanced Mitochondrial NAD⁺ Biosynthesis Improves Hepatic Energy Metabolism  
Nicholas Bentley  
University of New South Wales, Sydney

9:30 am  
MORNING TEA

Session 2

10:00 am  
Intellectual property and product commercialisation  
Dr Daniel Sieveking  
Spruson and Ferguson Intellectual Property

10:20 am  
Biochemical characterisation of the Mitochondrial Complex I Assembly (MCIA) Complex  
Dr Luke Formosa  
Monash University, Melbourne

10:35 am  
Electron Transport Chain Stress Signals Activation of ATM Kinase to Promote Pro-Survival Signaling and Metabolic Adaptations by a ROS Dependent Mechanism  
Jayne Barbour  
University of New South Wales, Sydney

10:50 am  
Characterizing biochemical effects of Complex I deficiency in human embryonic kidney 293 (HEK293T) cells  
Sophia Frentz  
Melbourne University and Murdoch Childrens Research Institute, Melbourne
11:05 am  In vitro cell models reveal OXPHOS defects in MCAD deficiency  
Sze Chern Lim  
Hudson Institute of Medical Research and Monash University, Melbourne

11:20 am  Panel discussion: Successful career development and career alternatives  
Professor Mike Ryan, Monash University, Melbourne  
Professor Aleksandra Filipovska, Harry Perkins Institute for Medical Research, Perth  
Dr Michael Lazarou, Monash University, Melbourne

12:00 pm  SYMPOSIUM CLOSE AND LUNCH
AussieMit 2016 Conference  
Saturday 26 November

1:00 pm  CONFERENCE OPENING AND WELCOME

Session 1 – Frontiers in Mitochondrial Medicine 1

1:10 pm  Interrogating the Mitochondrial interactome using BioID  
Professor Eric Shoubridge  
McGill University, Montreal, Canada

1:40 pm  Mitochondrial Disease Drug Development: the Saga of KH176  
Professor Jan Smeitink  
Radboud University, Nijmegen, The Netherlands

2:10 pm  The Australian Genomics Health Alliance: What does it have to offer to the Mitochondrial Disorders community?  
Professor John Christodoulou  
Murdoch Childrens Research Institute and Melbourne University, Melbourne

2:40 pm  AFTERNOON TEA

Session 2 – Frontiers in Mitochondrial Medicine 2

3:10 pm  Exercise to improve mitochondrial health  
Professor David Bishop  
Victoria University, Melbourne

3:40 pm  The NAD+ metabolome as a regulator of mitochondrial biogenesis and viability  
Dr Lindsay Wu  
University of New South Wales, Sydney

4:10 pm  Using Gene Editing to Generate Human Models of Mitochondrial Disease  
Dr Matthew McKenzie  
Hudson Institute of Medical Research, Melbourne, Australia

4:40 pm  Panel Discussion on Mitochondrial Donation Therapy  
Dr Grainne Gorman  
Associate Professor Ainsley Newson  
Dr Doug Lingard  
Dr Karen Crawley

5:30 pm  WELCOME RECEPTION
AussieMit 2016 Conference  
Sunday 27 November

**Session 1 – Mitochondrial Disease Research**

8:00 am  **Posttranscriptional Regulation of Mitochondrial Gene Expression**  
Professor Eric Shoubridge  
McGill University, Montreal, Canada

8:40 am  **TBA**  
Dr Grainne Gorman  
Newcastle University, Newcastle Upon Tyne, United Kingdom

9:10 am  **Serum metabolomics for mitochondrial disease diagnosis**  
Dr Ryan Davis  
University of Sydney, Sydney

9:30 am  **The complex I assembly factor TMEM126B, is associated with severe complex I deficiency and variable clinical phenotypes**  
Dr Alison Compton  
Murdoch Childrens Research Institute and University of Melbourne, Melbourne

9:45 am  **Can whole genome sequencing end the diagnostic odyssey for paediatric mitochondrial disorders?**  
Dr Lisa Riley  
The Children’s Hospital at Westmead and University of Sydney, Sydney

10:00 am  **MORNING TEA**

**Session 2 – Mitochondria in Neurodegenerative Disease**

10:30 am  **Parkinson’s Disease – how mitochondria contribute to disease pathogenesis**  
Professor Carolyn Sue  
University of Sydney and Royal North Shore Hospital, Sydney

10:55 am  **Mitochondrial contribution to Multiple Sclerosis susceptibility and disease progression**  
Professor Mark Slee  
Flinders University, Adelaide

11:20 am  **Mitochondria in Motor Neuron Disease**  
Dr Peter Crouch  
University of Melbourne, Melbourne

11:45 am  **Disease-associated protein Tau impairs Parkin translocation to damaged mitochondria by binding Parkin and trapping it in the cytosol**  
Nadia Cummins  
University of Queensland, Brisbane

12:00 pm  **Amyloid precursor protein drives down-regulation of mitochondrial oxidative phosphorylation independent of amyloid beta**  
Dr Isabel Lopez Sanchez  
Centre for Eye Research Australia and University of Melbourne, Melbourne
12:15 pm  Energy crisis in Parkinson’s disease: the pathogenic role of ATP13A2 mutations in mitochondrial bioenergetics  
Dr Jin-Sung Park  
Kolling Institute, Royal North Shore Hospital and University of Sydney, Sydney

12:30 pm  LUNCH AND POSTER VIEWING

Session 3 – Mitochondria in Metabolism

2:00 pm  Role of mitochondria in obesity and diabetes  
Associate Professor Nigel Turner  
University of New South Wales, Sydney

2:30 pm  Reining in the powerhouse: restoring mitochondrial function in diabetic kidney disease  
Associate Professor Melinda Coughlan  
Baker IDI Heart & Diabetes Institute, Melbourne

3:00 pm  Lipid-induced stress in oocyte mitochondria changes embryo development and mitochondrial inheritance in offspring  
Associate Professor Rebecca Robker  
University of Adelaide, Adelaide

3:30 pm  AFTERNOON TEA

Session 4 – Tools for Mitochondrial Research

4:00 pm  C. elegans: Approaches to examine mitochondrial dysfunction in metazoans  
Associate Professor Cole Haynes  
University of Massachusetts, Massachusetts, United States of America

4:30 pm  Using cellular and mouse models to help solve mitochondrial disease  
Professor David Thorburn  
Murdoch Childrens Research Institute, University of Melbourne and Victorian Clinical Genetic Services, Melbourne

5:00  TBA

7:00 pm  CONFERENCE DINNER – La Grillade
AussieMit Conference  
Monday 28 November

Session 1 – Mitochondrial Stress and Cell Survival

8:00 am  Mitochondrial UPR-mediated adaptations  
Associate Professor Cole Haynes  
University of Massachusetts, Massachusetts, USA

8:40 am  Peroxiredoxins as markers of mitochondrial oxidative stress  
Professor Mark Hampton  
University of Otago, Christchurch, New Zealand

9:10 am  Mitophagy in response to cellular stress  
Dr Michael Lazarou  
Monash University, Melbourne

9:30 am  Lattice light sheet live cell imaging of apoptotic mitochondria  
Dr Kate McArthur  
Walter and Eliza Hall Institute of Medical Research and University of Melbourne, Melbourne

9:45 am  MORNING TEA

Session 2 – Mitochondrial Biology

10:15 am  New assembly factors for mitochondrial complex I  
Professor Mike Ryan  
Monash University, Melbourne

10:45 am  Hierarchical RNA Processing Is Required for Mitochondrial Ribosome Assembly  
Professor Aleksandra Filipovska  
Harry Perkins Institute of Medical Research and University of Western Australia, Perth

11:15 am  Mitochondrial Epigenetics  
Dr Steve Zuryn  
University of Queensland, Brisbane

11:35 am  Coupling of gene-editing with large-scale proteomics reveals accessory subunits to be integral for assembly and function of human mitochondrial complex I  
Dr David Stroud  
Monash University, Melbourne

11:50 am  LUNCH

Session 3 – Selected Abstracts

1:00 pm  Deletions in the ATAD3 gene cluster cause cerebellar developmental defects  
Dr Ann Frazier  
Murdoch Childrens Research Institute and University of Melbourne, Melbourne
1:15 pm  Using massively parallel sequencing to determine the genetic basis of Leigh Syndrome, the most common paediatric mitochondrial disorder
Nicole Lake
Murdoch Childrens Research Institute and University of Melbourne, Melbourne

1:30 pm  Loss of an unusual RNA-binding protein, TACO1, causes late-onset mitochondrial dysfunction
Associate Professor Oliver Rackham
Harry Perkins Institute of Medical Research and University of Western Australia, Perth

1:45 pm  A novel dominant-negative mutation in the DNM1L gene encoding the mitochondrial fission mediator causes epileptic encephalopathy
Dr Laura Osellame
Monash University, Melbourne

2:00 pm  Identification of biallelic mutations in IARS as the known causative gene for Weak Calf Syndrome, which cause infantile mitochondrial hepatopathy
Dr Masaru Shimura
Chiba Children's Hospital, Chiba, Japan

2:15 pm  Frataxin-deficiency in the heart results in an impaired Nrf2 response: A dual mechanism mediated via up-regulation of Keap1 and GSK3β axis
Dr Michael Huang
University of Sydney, Sydney

2:30 pm  AFTERNOON TEA

3:00 pm  Session 4 – Cross Debates

3:00 pm  Debate 1: Stem cells; tool or treatment
Dr Matthew McKenzie
Hudson Institute of Medical Research, Melbourne
versus
TBA

3:20 pm  Debate 2: Whole Exome Sequencing vs Whole Genome Sequencing for mitochondrial research
Professor David Thorburn
Murdoch Childrens Research Institute, University of Melbourne and Victorian Clinical Genetic Services, Melbourne
versus
Dr Mark Cowley
Kinghorn Centre for Clinical Genomics and Garvan Institute, Sydney

3:40 pm  Debate 3: Mice vs Men for mitochondrial research
Professor Eric Shoubridge
McGill University, Montreal, Canada
versus
Dr Grainne Gorman
Newcastle University, Newcastle Upon Tyne, UK

4:00 pm  AWARD CEREMONY AND CONFERENCE CLOSE