



## UK Neuromuscular Translational Research Conference 2011

Kennedy Lecture Theatre, UCL Institute of Child Health, Guildford Street, London WC1N 1EH  
Tuesday 29<sup>th</sup> – Wednesday 30<sup>th</sup> March

### Day 1 – Tuesday 29<sup>th</sup> March

- 09:00 – 10.15 Registration
- 10:15 – 10:30 Introduction  
Professor Mike Hanna, UCL Institute of Neurology
- 10:30 – 12:30 Translational Research in Human Mitochondrial Diseases  
Jointly Sponsored session MRC Centre for Neuromuscular Diseases  
and MRC Mitochondrial Biology Unit Cambridge**
- Chairs: Professor John Walker, MRC Mitochondrial Biology Unit  
Cambridge, and Professor Patrick Chinnery, Newcastle University**
- 10:30 – 11:00 Prevention of the transmission of mitochondrial diseases  
Professor Doug Turnbull, Newcastle University
- 11:00 – 11:30 Developing therapies for human mitochondrial diseases  
Dr Michael Murphy, MRC Mitochondrial Biology Unit, Cambridge
- 11:30 – 12:00 Disturbance of calcium regulation and mitochondrial turnover as targets for  
therapy in mitochondrial disease  
Professor Michael Duchen, University College London
- 12:00 – 12:15 Platform presentation  
The MRC Centre for Translational Research in Neuromuscular Disease:  
Mitochondrial Disease Patient Cohort Study UK  
Dr Rob Pitceathly, UCL Institute of Neurology
- 12:15 – 12:30 Platform presentation  
Dominant and recessive *RRM2B* mutations cause familial PEO and multiple  
mtDNA deletions in muscle  
Dr Grainne Gorman, Newcastle University
- 12:30 – 13:30 **The Second Victor Dubowitz Lecture  
Introduced by Professor Francesco Muntoni**  
2011 State of the art in treatments for muscular dystrophy  
Professor Thomas Voit, Institute of Myology, Paris
- 13:30-14:30 Posters and Lunch

- 14:30 – 16:30**      **Peripheral Neuropathy: Bench to Bedside**
- Chairs: Professor Mary Reilly, UCL Institute of Neurology, and Professor Kristjan Jessen, University College London**
- 14:30 – 15:00      New molecular targets in hereditary neuropathies  
Professor Vincent Timmerman, University of Antwerp
- 15:00 – 15:30      Clinical trials in peripheral neuropathies – where have we got?  
Professor Richard Hughes, Cochrane Neuromuscular Disease Group
- 15:30 – 16:00      Animal models' contribution to the pathogenesis and treatment of inherited neuropathies  
Dr Michael Sereda, Max Planck Institute for Experimental Medicine, Göttingen
- 16:00 – 16:15      Platform presentation  
Neuregulin-1 is required for axoglial signalling following peripheral nerve injury to ensure normal re-myelination and functional recovery  
Florence Fricker, King's College London
- 16:15 – 16:30      Platform presentation  
Characterisation of novel mutations within heat shock protein 27 causing motor axonopathies  
Amy Innes, UCL Institute of Neurology
- 16:30 – 17:00      Professor Sir John Tooke  
Vice Provost (Health), Head of the UCL School of Life & Medical Sciences and Head of the UCL Medical School
- 17:00 - 17:30      Posters  
Tea                      Balcony
- 17:30 – 18:30      **The Second Morgan-Hughes-Thomas Lecture**  
**Introduced by Professor Mary Reilly**  
Motor Neuropathies - bench to bedside  
Professor Robert Brown, University of Massachusetts Medical School
- 18:30 – 19:30      Drinks reception posters introduced by Robert Meadowcroft, MDC CEO
- 20:00 - 22:45      Gala dinner – Great Hall, BMA House, Tavistock Square

**Day 2 – Wednesday 30<sup>th</sup> March**

- 09:00 – 11:00**      **New MRI Applications in Neuromuscular Disease**  
**A diagnostic and outcome measure tool in NM Disease?**
- Chairs: Professor Tarek Yousry and Professor Volker Straub**
- 09:00 – 09:30      Application of MRI in paediatric muscle disease  
Professor Eugenio Mercuri, Catholic University, Rome

- 09:30 – 10:00 Quantitative magnetic resonance imaging of neuromuscular diseases in adults  
Dr John Thornton, National Hospital for Neurology and Neurosurgery
- 10:00 – 10:30 Quantitative MRI in FSHD and DMD  
Dr Hermien Kan, Leiden University Medical Centre
- 10:30 – 11:00 MRI in LGMD2I  
Dr Tracey Willis, Newcastle University
- 11:00 – 11:15 Platform presentation  
Skeletal Muscle MRI-Determined Fat Fraction and Myometric Strength in Inclusion Body Myositis and Charcot-Marie-Tooth Disease Type 1A  
Dr Chris Sinclair, UCL Institute of Neurology
- 11:15 – 11:30 Platform presentation  
Magnetic resonance imaging in the non-dystrophic myotonias  
Dr Jasper Morrow, UCL Institute of Neurology
- 11:30- 13:15** **Poster guided tours**  
Mitochondrial disease: Ian Holt  
Peripheral neuropathies: Vincent Timmerman & Richard Hughes  
Neuromuscular MRI: Paul Matthews  
Neuromuscular animal models: Francesco Muntoni & Kay Davies  
Molecular therapies for DMD: Matthew Wood  
Congenital/myofibrillar myopathies & IBM: David Hilton-Jones & Janice Holton  
Muscle channelopathies & myasthenia gravis: David Beeson  
Muscle satellite cells: Jenny Morgan  
Neuromuscular databases: Adnan Manzur
- 13:15 – 14:00 Lunch
- 14:00 – 16:00** **Lessons from mouse models of muscular dystrophy and spinal muscular atrophy**  
  
**Chairs: Professor Francesco Muntoni, UCL Institute of Child Health, and Professor Dame Kay Davies, University of Oxford**
- 14:00 – 14:30 Outcome measures in the mdx mouse  
Professor Dominic Wells, Royal Veterinary College
- 14:30 – 15:00 Mouse models of SMA: implications for the timing and delivery of therapy  
Professor Kevin Talbot, University of Oxford
- 15:00 – 15:30 Novel insight in muscle and brain involvement in dystroglycanopathies  
Dr Susan Brown, Royal Veterinary College
- 15:30 – 16:00 TDP-43 mouse models – lessons for neuromuscular disease  
Dr Peter Joyce, MRC Mammalian Genetics Unit, Harwell
- 16:00 – 16:15 Poster prizes and close