



## UK Neuromuscular Translational Research Conference 2012

Scotswood Suite, International Centre for Life, Times Square, Newcastle upon Tyne, NE1 4EP  
Thursday 22<sup>nd</sup> – Friday 23<sup>rd</sup> March

### Day 1 – Thursday 22<sup>nd</sup> March

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|----------------------|---|
| 09:00 – 10.15        | Registration  |
| 10:15 – 10:30        | Introduction<br>Professor Mike Hanna, UCL Institute of Neurology  |
| <b>10:30 – 12:30</b> | <b>Molecular Mechanisms in Neuromuscular Disease</b><br><b>Chair: Professor Doug Turnbull</b>   |
| 10:30 – 11:00        | Molecular mechanisms and molecular pathophysiology in Duchenne muscular dystrophy<br>Professor Dominic Wells<br>Professor in Translational Medicine, Royal Veterinary College                     |
| 11:00 – 11:30        | Molecular mechanisms of myofibrillar myopathies<br>Professor Dr Dieter Fürst<br>Institute for Cell Biology, University of Bonn  |
| 11:30 – 12:00        | Coffee break  |
| 12:00 – 12:30        | Molecular mechanisms of mitochondrial encephalomyopathies<br>Dr Massimo Zeviani<br>Director, Unit of Molecular Neurogenetics, Fondazione Istituto Neurologico "Carlo Besta", Milan                |
| 12:30 – 12:45        | Platform presentation<br>Identification of new chemical compounds which upregulate utrophin for the therapy of Duchenne muscular dystrophy<br>Dr Rebecca Fairclough<br>University of Oxford       |
| 12:45 – 13:00        | Platform presentation<br>Dysregulated mitophagy and mitochondrial transport in severe dominant optic atrophy due to OPA1 mutations<br>Professor Joanna Poulton<br>John Radcliffe Hospital, Oxford |
| 13:00 – 14:30        | Posters and Lunch, mezzanine  |
| <b>14:30 – 16:30</b> | <b>Translational Pathways</b><br><b>Chair: Professor Mary Reilly</b>  |
| 14:30 – 15:00        | Title TBC<br>Katherine Klinger  |

Genzyme Corporation

- 15:00 – 15:30 Translating discovery into experimental medicine and treatment - the amyloid experience  
Professor Philip Hawkins  
Clinical Director, National Amyloidosis Centre,  
UCL/Royal Free Hospital NHS Foundation Trust
- 15:30 – 15:45 Platform presentation  
High-Dose riboflavin therapy in Brown-Vialetto-Van Laere syndrome: clinical and biochemical improvement  
Dr Reghan Foley  
UCL Institute of Child Health
- 15:45 – 16:00 Platform presentation  
Efficacy of mexiletine in non-dystrophic myotonia: results of an international multi-centred randomised controlled trial  
Dr Dipa Raja Rayan  
UCL Institute of Neurology
- 16:00 – 16:30 TBC
- 16:30 – 17:00 The MRC's Translational Research Strategy  
Declan Mulkeen  
Director, Research Programmes, Medical Research Council
- 17:00 - 17:30 Posters & tea, mezzanine
- 17:30 – 18:30 **The Second John Walton Lecture**  
Introduced by Professors Kate Bushby & Doug Turnbull  
'Overcoming weakness of the flesh'  
Professor Robert C. Griggs, M.D  
Professor of Neurology, Medicine, Pathology, Pediatrics Center for Human Experimental Therapeutics at the University of Rochester School of Medicine and Dentistry, USA
- 18:30 – 19:30 Drinks reception & posters introduced by Robert Meadowcroft, MDC CEO
- 20:00 - 22:45 Gala dinner, Scotswood Suite

**Day 2 – Friday 23<sup>rd</sup> March**

- 09:00 – 10:30 Next generation sequencing**  
**Chair: Professors Francesco Muntoni**
- 09:00 – 09:30 Advancing from targeted resequencing to whole exome sequencing: a perspective on neuromuscular disorders  
Professor Madhuri Hegde  
Associate Professor/Emory Genetics Lab Scientific Director,  
Emory University School of Medicine, USA
- 09:30 – 10:00 Next generation sequencing in neuromuscular disorders  
Professor Henry Houlden  
Professor of Neurology, Department of Molecular Neuroscience,  
UCL Institute of Neurology

- 10:00 – 10:15 Platform presentation  
Exome sequencing in three families with cytoplasmic body myopathy with early respiratory failure  
Dr Gerald Pfeffer  
Newcastle University
- 10:15 – 10:30 **Platform presentation**  
**TBC**
- 10:30- 12:45** **Poster guided tours**  
Mitochondrial disease: Rob Taylor & Mike Hanna  
Peripheral neuropathies: Rita Horvath & Mary Reilly  
Neuromuscular MRI: Tarek Yousry & Andrew Blamire  
Neuromuscular animal models: **Volker Straub** & Sue Brown  
Molecular therapies for DMD: Kate Bushby & Nic Wells  
Congenital myopathies & IBM: David Hilton-Jones & Chris Turner  
Channelopathies & myasthenia gravis: David Beeson & Hanns Lochmüller  
Muscle satellite cells: Kay Davies & Jenny Morgan  
Neuromuscular databases: **Adnan Manzur**
- 12:45 – 13:45 Lunch, mezzanine
- 13:45 – 15:45** **Stem cells**  
**Chair: Professor Dame Kay Davies**
- 13:45 – 14:15 Epicardium-derived cardiac repair  
Professor Paul Riley  
The Oxford Stem Cell Institute, DPAG, University of Oxford
- 14:15 – 14:45 Stem cells in multiple sclerosis and motor neuron disease  
Dr Siddharthan Chandran  
Director of The Euan MacDonald Centre for MND Research & Professor of Neurology, University of Edinburgh
- 14:45 – 15:15 A phase I/II cell therapy trial for Duchenne Muscular Dystrophy  
Dr Giulio Cossu  
UCL Institute of Child Health
- 15:15 – 15:30 Platform presentation  
The satellite cell in male and female, developing and adult mice: evidence for functionally distinct stem cell populations  
Alice Neal  
UCL Institute of Neurology
- 15:30 – 15:45 Platform presentation  
Bmi1 controls satellite cells proliferation and maintenance and plays an important role in muscle regeneration  
Valentina Di Foggia  
QMUL, Barts and the London School of Medicine and Dentistry
- 15:45 – 16:00 Poster prizes and close