



12th UK Neuromuscular Translational Research Conference

Centre for Life, Newcastle

Thursday 4 and Friday 5 April 2019

PROGRAMME

Day 1 – Thursday 4 April

09:00 – 09:30 **Registration and Coffee**

09:30 – 09:40

Introduction

Professor Michael Hanna

Director, MRC Centre for Neuromuscular Diseases and Institute of Neurology, UCL

Session 1: Chairs:

New International Centre for Genomic Research in NMD

Professor Michael Hanna, Director, MRC Centre for Neuromuscular Diseases and Queen Square Institute of Neurology, UCL

Professor Volker Straub, Director, John Walton Muscular Dystrophy Research Centre and MRC Centre, Newcastle University and Newcastle Hospitals NHS Foundation Trust

Professor Patrick Chinnery, Head of Department of Clinical Neurosciences, University of Cambridge

09:40 – 09:55

Overview and vision for new International Centre

Professor Michael Hanna

09:55 – 10:15

South Africa – Professor Francois van der Westhuizen, North-West University

10:15 – 10:35

Brazil – Professor Wilson Marques, University of São Paulo

10:35 – 10:55

Zambia – Dr Michelle Kvalsund, Michigan State University and University of Zambia

- 10:55 – 11:15 **Turkey** – Dr Yavuz Oktay, Dokuz Eylül University
- 11:15 – 11:35 **India** – Dr K Thangaraj, Centre for Cellular and Molecular Biology, Hyderabad
- 11:35 – 12:00 Coffee**
- 12:00 – 12:30 **Platform presentations (2 x 15 min)**
- PN11 Pentanucleotide repeat expansion causes CANVAS and Late-Onset Sensory Ataxia**
Dr Andrea Cortese
UCL MRC CNMD
- OD21 Clinical presentation of patients with TANGO2 mutations**
Dr Angela Pyle
Wellcome Centre for Mitochondrial Research, Newcastle University
- 12:30 – 12:45 **Late breaking science platform**
- 12:45 – 13:30 **Lunch**
- 13:30 – 14:45 **Poster guided sessions and coffee**
- Session 2:
Chairs:** **Kate Bushby – an advocate for translational research in NMD**
Professor Volker Straub, Newcastle University and **Professor Mary Reilly**, Co-Director MRC CNMD, UCL
- 14:45 – 15:05 **Clinical Services for Neuromuscular Disease Patients**
Professor Sir Doug Turnbull, Newcastle University
- 15:05 – 15:25 **From Gene Discovery to Therapy**
Professor Francesco Muntoni, UCL GOSH ICH
- 15:25 – 15:45 **The FOR DMD trial: Individualized vs standardized treatment**
Professor Robert C Griggs, University of Rochester, NY
- 15:45 – 16:05 **Vision-DMD, developing an alternative to steroids**
Dr Michela Guglieri, Newcastle University
- 16:05 – 16:35 Coffee and posters**
- 16:35 – 16:50 **EURO-NMD, a reference network for neuromuscular diseases**
Dr Teresinha Evangelista, Newcastle University
- 16:50 – 17:05 **The DMD-Hub, increasing trial capacity in the UK**
Emma Heslop, Newcastle University

17:05 – 17:20 **Promoting rare disease policies across Europe**
Victoria Hedley, Newcastle University

17:20 – 17:30 Comfort break

17:30 – 18:15 **4th John Walton Lecture**
What makes Genetics Human?
Professor Sir John Burn, Newcastle University

18:15 **Drinks reception and posters / networking**
Mezzanine level, Centre for Life

19:30 **Gala dinner**
Scotswood, Centre for Life

After dinner speaker: Professor Katie Bushby

Day 2 – Friday 5 April

- Session 3:** **Developing Genetic Therapies**
Chairs: **Professor Dame Kay Davies**, Dr Lee's Professor of Anatomy Oxford Neuromuscular Centre
Professor Francesco Muntoni Director, Dubowitz Neuromuscular Centre, ICL GOSH ICH
- 08:30 – 09:00 **Genome Editing for Duchenne Muscular Dystrophy**
Dr Charles Gersbach, Duke University
- 09:00 – 09:30 **Alternate gene therapy approaches to DMD**
Dr Kevin Flanigan, Nationwide Children's Hospital, Columbus, Ohio
- 09:30 – 10:00 **AAV-mediated Gene Therapy in Neuromuscular Disease: Clinical Immunology Considerations**
Dr Barry Byrne, University of Florida
- 10:00 – 11:15 **Poster guided sessions and coffee**
- 11:15 – 11:45 **Platform presentations (2 x 15 min)**
- MND14 FIREFISH Part 1: Early clinical results following an increase of survival of motor neuron protein (SMN) in infants with Type 1 spinal muscular atrophy (SMA) treated with risdiplam (RG7916)**
Dr Giovanni Baranello
Dubowitz NM Centre, UCL GOSH ICH
- D32 A Human iPS Cell-Derived Artificial Skeletal Muscle for DMD Therapy Development**
Moustafa Khedr
Cell and Developmental Biology Department, UCL
- 11:45 – 12:45 **Poster flash sessions**
Chaired by:
Dr Rob Taylor, Wellcome Centre for Mitochondrial Research, Newcastle University and
Dr Rob Pitceathly, UCL Queen Square Institute of Neurology
- See flash poster presentations list at end of programme*
- 12:45 – 13:45 **Lunch**

Session 4:	Mitochondrial Disease
Chairs:	Professor Bobby McFarland , Wellcome Centre for Mitochondrial Research, Newcastle University Professor Rita Horvath Department of Clinical Neurosciences, University of Cambridge
13:45 – 14:15	Preventing transmission of pathogenic mitochondrial DNA mutations Professor Mary Herbert, Newcastle University
14:15 – 14:45	Mitochondrial donation: predicting who might benefit? Grainne Gorman, Newcastle University
14:45 – 15:15	Coffee and posters
15:15 – 16:15	Platform presentations (4 x 15 min)
	M14 Small molecules counter-select deleterious mitochondrial DNA variants by inhibiting their replication in human cells Antonella Spinazzola UCL Queen Square Institute of Neurology
	NMJ+C06 Salbutamol modulates postsynaptic specialisation at the neuromuscular junction in a mouse model of ColQ myasthenic syndrome Dr Grace McMacken Institute of Genetic Medicine, Newcastle University
	D08 Non-invasive assessment of fibrosis in mouse skeletal and cardiac muscle by contrast enhanced magnetic resonance imaging using EP3533 Dr Alex Murphy, Newcastle University
	MND15 MRI detection of human motor unit fasciculation in Amyotrophic Lateral Sclerosis. Prof Andrew Blamire Institute of Cellular Medicine, Newcastle University
16:15 – 16:30	MDUK update Dr Kate Adcock Director of Research, MDUK
16:30 – 16:45	MRC strategy Dr Louise Jones, Head of Translational Research, Medical Research Council
16:45 – 17:00	Poster prizes and close Professor Michael Hanna and Professor Volker Straub

