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: New International Centre for Genomic Research in NMD
Chairs:
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:  Kate Bushby – an advocate for translational research in NMD
Chairs:  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

19:30 Drinks reception and Gala dinner
Centre for Life

After dinner speaker: Professor Katie Bushby
Day 2 – Friday 4 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
  UCL GOSH ICH

11:45 – 12:45  **Poster flash sessions**
Chaired by: (tbc)
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 Development and Optimisation of techniques to prevent transmission of mitochondrial DNA disease
Professor Mary Herbert, Newcastle University

14:15 – 14:45 Clinical aspects of prevention of mitochondrial disease
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