



10th UK Neuromuscular Translational Research Conference 2017

ICH, 30 Guilford Street, London WC1N 1EH

Kennedy Lecture Theatre

Wednesday 22 and Thursday 23 March 2017

PROGRAMME

Day 1 – Wednesday 22nd March

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

09:30 – 09:45

Introduction

Prof Michael Hanna

Director, MRC Centre and UCL Institute of Neurology

Session 1: Neuropathies

Chairs: Prof Mary Reilly / Prof Rita Horvath

09:45 – 10:15

Using stem cells and light to restore function to paralysed muscles

Professor Linda Greensmith

Sobell Department of Motor Neuroscience and Movement Disorders

UCL Institute of Neurology

10:15 – 10:45

Application of induced pluripotent stem cell technology for disease modelling and drug discovery in peripheral sensory neurons

Dr James Bilsland

ARUK Drug Discovery Institute, UCL

10:45 – 11:15

Coffee

11:15 – 11:45

Modulation of cytoplasmic dynein and tubulin modification as potential therapeutic targets in SMA-LED

Dr Majid Hafezparast

Neuroscience, School of Life Sciences, University of Sussex

- 11:45 – 12:00 Platform presentation 1
Trk receptor signalling and sensory neuron fate are perturbed in human neuropathy caused by *Gars* mutations
Dr James N. Sleigh
UCL Institute of Neurology
- 12:00 – 12:15 Platform presentation 2
Investigating dysfunctional RNA processing in TDP-43 mouse mutants
Prasanth Sivakumar
UCL Institute of Neurology
- 12:15 – 14:00 **Lunch and guided poster sessions (poster session commences at 12:30)**

Session 2: Genomic Therapies / SMA
Chairs: Prof Francesco Muntoni / Prof Hanns Lochmüller

- 14:00 – 14:30 **The power of genetic modifiers in SMA**
Professor Dr Brunhilde Wirth
Institute of Human Genetics, Cologne
- 14:30 – 15:00 **Giant Axonal Neuropathy: Natural History, Outcome Measures, and Intrathecal AAV9 Mediated Gene Transfer**
Professor Carsten Bonnemann
National Institute of Neurological Disorders and Stroke, USA
- 15:00 – 15:30 **Experimental trials in spinal muscular atrophy**
Dr Richard Finkel
Nemours Children's Hospital, Orlando, USA
- 15:30 – 16:00 **Coffee and posters**
- 16:00 – 16:15 Platform presentation 3
SMN depletion causes defects in endosomal trafficking that impair synaptic function
Dr Maria Dimitriadi
School of life and medical Sciences, University of Hertfordshire
- 16:15 – 16:30 Platform presentation 4
Potential antisense oligonucleotide therapeutic approaches in COL6-related congenital muscular dystrophy
Dr Haiyan Zhou
Institute of Child Health, UCL

- 16:30 – 16:50 **MRC Strategy**
Kathryn Adcock
Head of Neurosciences and Mental Health, Medical Research Council
- 16:50 – 17:45 The fourth Morgan-Hughes Thomas lecture
Membrane fusion in the cell and how it is harnessed for neurotransmission
Professor James E. Rothman
Nobel Laureate
Research Professor UCL / Professor and Chair Cell Biology, Yale
- 17:45 – 18:00 **Robert Meadowcroft**
CEO, Muscular Dystrophy UK
- 18:00 – 18:30 **Poster viewing**
- 19:00 **Drinks reception**
Goodenough College, Mecklenburgh Square, London WC1N 2AB
- 19:30 **Gala dinner**
Goodenough College, Mecklenburgh Square, London WC1N 2AB

To Note: Those displaying posters. To allow for judging and poster sessions, posters may only be put up and taken down during these times:

*Day 1 Weds 22 March
Posters up: 8.45 am to 9.15 am
Posters down: 18:30 – 18.45 pm*

Day 2 – Thursday 23rd March

Session 3: Mitochondrial disease
Chairs: Prof Michael Hanna / Prof Rob Taylor

- 08:30 – 09:00 **Manipulating mtDNA heteroplasmy with designer zinc-finger nuclease technology**
Dr Michal Minczuk
MRC Mitochondrial Biology Unit, Cambridge
- 09:00 – 09:30 **Manipulating mtDNA with TALEN-based nucleases**
Professor Carlos Moraes
Esther Lichtenstein Professor in Neurology
University of Miami Miller School of Medicine
- 09:30 – 10:00 **Clinical trial readiness in children with mitochondrial disease and the development of the International paediatric rating scale**

Dr Saskia Koene
Radboud University, Nijmegen, NL

10:00 – 11:30 **Poster guided sessions and coffee**

11:30 – 11:45 Platform Presentation 5
Using a quantitative quadruple immunofluorescent assay to detect isolated Complex I deficiency.
Tasnim Ahmed
Wellcome Trust Centre for Mitochondrial Research
Newcastle University

11:45 – 12:00 Platform presentation 6
Cause of death in a cohort of mitochondrial patients
Iwona Skorupinska
UCL Institute of Neurology

12:00 – 13:00 The fourth Victor Dubowitz Lecture
**Spell-checking nature:
A new therapeutic approach to neuromuscular disorders**
Professor Ronald Cohn
Professor and Chair, Department of Paediatrics, University of Toronto
Paediatrician-in-Chief, The Hospital for Sick Children

13:00 – 13:30 **Lunch**

13:30 – 14:30 **Poster flash sessions**
Chaired by Prof. Michael Hanna, UCL Institute of Neurology

Session 4: NIHR Rare Diseases Translational Research Collaboration
Chairs: Prof Volker Straub / Prof Patrick Chinnery

14:30 – 15:00 **The NIHR Translational BioResource for Rare and Common Diseases**
Professor Patrick Chinnery Chair NIH RD TRC
University of Cambridge

15:00 – 15:30 **Coffee and posters**

15:30 – 16:00 **Myotonic dystrophy trial readiness**
Professor Hanns Lochmüller
Institute of Genetic Medicine
Newcastle University

16:00 – 16:30 **Skeletal muscle channelopathies: new phenotypes and new genes**
Dr Emma Matthews
UCL Institute of Neurology

- 16:30 – 16:45 Platform 7
A recessive mutation in EXOSC9 causes abnormal RNA metabolism resulting in a novel form of cerebellar hypoplasia/atrophy with early motor neuronopathy
Dr David Burns
John Walton Muscular Dystrophy Research Centre and Wellcome Trust Centre for Mitochondrial Research
Newcastle University
- 16:45 – 17:00 Platform 8
Salbutamol counteracts long-term adverse effects of pyridostigmine on strength and neuromuscular junction structure in myasthenic mice
An Vanhaesebrouck
Nuffield Department of Clinical Neurosciences, University of Oxford
- 17:00 – 17:30 **Poster prizes and close**

To Note: Those displaying posters. To allow for judging and poster sessions, posters may only be put up and taken down during these times:

Day 2 Thurs 23 March

Posters up: evening of 22nd 18.45 pm onwards OR by 8.30 am latest 23 March

Building access is from 8am.

Posters down: 17:00 onwards