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*