Event programme
14th UK Annual Neuromuscular Translational Research Conference
New therapies in Neuromuscular Diseases
Online event
25th and 26th March 2021

(International colleagues pls note all times given are in GMT – Greenwich Mean Time)

Day 1 – Thursday 25 March 2021

Poster viewing: Prior to the event official start time, of 13:00 hr, research posters will be available in the morning for viewing online. Links will be provided to all registered delegates to view the posters.

13:00–13:10 Introduction and Housekeeping
Professor Michael Hanna
Director, CNMD, ICGNMD and UCL Institute of Neurology

13:10–13:25 Welcome from UCL President and Provost
Dr Michael Spence AC

New therapy developments in neuromuscular diseases - I
Session Chairs: Professor Francesco Muntoni and Professor Volker Straub

13:25–13:55 RNA antisense therapeutics for neuromuscular and neurological conditions
Professor Adrian R Krainer
Cold Spring Harbor Laboratory, NY, USA

13:55–14:25 New therapies in ALS
Professor Dame Pamela Shaw
Sheffield Institute for Translational Neuroscience (SITraN)

14:25–14:40 Comfort Break and sponsor slides

14:40–15:10 Window for optimal therapeutic intervention in SMA
Dr Charlotte Sumner
Johns Hopkins University, Baltimore, USA

15:10–16:10 Flash poster sessions
Chairs: Professor Mary Reilly (CNMD and ICGNMD, UCL UCL) and Professor Rita Horvath (Dept of clinical neurosciences, University of Cambridge)

Comparing age-dependent changes in heteroplasmy of an mtDNA point-mutation between mitotic and post-mitotic tissues using a mouse model of mitochondrial disease
Dr Tiago Bernardino Gomes
Wellcome Trust Centre for Mitochondrial Research, Newcastle University

Investigation of Compounds Inducing Alpha Tubulin Acetylation by Drug Repurposing Approach
Ms Özge Çetin
Keele University

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Pre-operative exercise and pyrexia as modifying factors in Malignant Hyperthermia
Dr Heinz Jungbluth
IoPPN, King’s College London

Combining the power of patients and mouse models to identify biomarkers in CMT
Mr Matthew Jennings
Department of Clinical Neurosciences,
Cambridge University

Therapeutic potential of reducing Krüppel-like factor 15 (Klf15) activity in Spinal Muscular Atrophy (SMA)
Ms Emma Sutton
Keele University

Impact of Teleneurology Neuromuscular Clinic on the accessibility of care for patients with Inherited Neuromuscular disorders during COVID-19 pandemic
Dr Vishnu Venugopalan Y
All India Institute of Medical Sciences, New Delhi, India

Clinical and genetic spectrum of a large cohort of delta-sarcoglycan muscular dystrophy
Dr Jorge Alonso-Perez
Hospital de la Santa Creu i Sant Pau, Universitat Autònoma de Barcelona, Barcelona, Spain

Making sense of missense variants in TTN-related congenital myopathies
Dr Martin Rees
Randall Centre for Cell and Molecular Biophysics, King’s College London

16:10–16:25 Comfort break and sponsor slides

16:25–16:40 Update from Oxford MDUK Neuromuscular Centre
Professor Dame Kay Davies
University of Oxford

16:40–17:25 The fifth Morgan-Hughes Thomas lecture introduced by Professor Michael Hanna
The origins of mtDNA mutations: implications for neuromuscular disease
Professor Patrick Chinnery
University of Cambridge, UK

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Day 2 – Friday 26th March

New therapy developments in neuromuscular diseases – II
Session Chairs: Professor Mary Reilly and Professor Bobby McFarland

13:00–13:10 Welcome and housekeeping day 2
Professor Mary Reilly
CNMD and ICGNMD, UCL

13:10–13:40 Modifying Muscular Dystrophy
Elizabeth McNally, M.D., Ph.D.
Northwestern University Feinberg School of Medicine
Chicago, USA

13:40–14:10 Giant Axonal Neuropathy:
From Outcome Measures to First-in-Human AAV Mediated Intrathecal Gene Transfer – Interim Analysis and Lessons Learned
Carsten Bönnemann, M.D.
NIH, USA

14:10–14:25 Comfort break and sponsor posters

14:25–15:35 Platform presentations
Chairs: Professor Rob Taylor (Wellcome Trust Centre for Mitochondrial Research) and Dr Rob Pitceathly (UCL IoN)

Recessive variants in orphan disease gene ATG7 cause neuromuscular disorders through impaired autophagy
Dr Jack Collier
Wellcome Centre for Mitochondrial Research, Newcastle University

Molecular mechanism of muscle atrophy in SBMA
Dr Mitra Forouhan
University of Oxford, UK

Increased startle responses to threat in Duchenne muscular dystrophy: A biomarker of brain dystrophin deficiency?
Dr Kate Maresh
UCL GOS Institute of Child Health

Investigating nucleoside supplementation in rrm2b deficient zebrafish model of mtDNA depletion
Mr Benjamin Munro
Department of Clinical Neuroscience, University of Cambridge

15:35-15:45 MDUK update
Dr Kate Adcock
Director of Research and Innovation, Muscular Dystrophy UK

15:45–16:25 The fifth Victor Dubowitz Lecture introduced by Professor Francesco Muntoni, UCL GOSH ICH
Spinal Muscular Atrophy: About a lethal disorder that has been disappearing for 3 years (in Southern Belgium)
Professor Laurent Servais, University of Oxford

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Prizes and close

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