

Event programme

15th UK Annual Neuromuscular Translational Research Conference

Online event

26th and 27th April 2022

(international colleagues pls note time zone is UTC+01:00)

Day 1 – Tuesday 26th April 2022

Poster viewing: Prior to the event official start time, of 13:00 hrs, 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 – 13:15

Introduction
Professor Michael Hanna
Director, ICGNMD and UCL Institute of Neurology

Session 1:

Session Chairs:

Professor Mary Reilly, UCL and **Professor Volker Straub**, Newcastle University

13:15 – 13:45

Development and Assessment of Novel biomarkers for Charcot-Marie-Tooth Disease
Professor John Svaren
Director, IDD Models Core
University of Wisconsin-Madison

13:45 – 14:15

Update on methods development in rare disease diagnostics for Mendelian myopathies
Dr Vijay Ganesh
Broad Institute

14:15 – 14:35

Comfort Break and sponsor slides

14:35 – 15:05

The relevance of Smart Technologies for patient reported outcomes/remote assessments
Dr Elin Haf Davies
CEO, Aparito

15:05 – 16:15

Poster session chairs:

Flash poster sessions 1 hour 10 min
Dr Anna Sarkozy, UCL and **Professor Rita Horvath**, University of Cambridge

9 x flash posters of 5 min + 2 min questions each. 3 slides max.

Loss of function mutations in DNAJB4 cause a myopathy with early respiratory failure
Dr Ana Topf
Newcastle University

REN001, a novel peroxisome proliferator-activated receptor delta (PPAR δ) agonist, improves gait in patients with primary mitochondrial myopathy after 12 weeks
Dr Lisa Alcock
Newcastle University

A Cross-sectional Analysis Of Spinal MRI Findings In UK hATTR
Dr Luke O' Donnell
UCL IoN

Progressive Transcriptional Phenotypes of a Human Stem Cell Model of Spinal Bulbar Muscular Atrophy (SBMA)
Dr Helen Devine
Newcastle University

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Whole-body fasciculation detection in Amyotrophic Lateral Sclerosis (ALS) using motor unit

MRI (MUMRI)

Dr Linda Heskamp

Newcastle University

Functional motor performance in young steroid-naïve boys with Duchenne muscular dystrophy compared to healthy controls

Jasmine Hoskens

University of Leuven

Impact of levels of mobility on participation in people with neuromuscular conditions

Karen Wong

Newcastle University

An update on the Brazilian cohort of Neuromuscular Disorders enrolled in the ICGNMD study

Dr Pedro Tomaselli

University of São Paulo

Novel DNM1L variants impair mitochondrial dynamics through divergent mechanisms

Newcastle University

Dr Monika Olahova

Newcastle University

16:15 – 16:35

Comfort break and sponsor slides

Session chairs:

Professor Rob Taylor, Newcastle University and **Professor Henry Houlden**, UCL

16:35 – 17:05

Leveraging AAV biology and directed evolution for developing tissue-targeted viral gene delivery vehicles

Sharif Tabebordbar, Ph.D

Kate Therapeutics

17:05 – 17:35

Rare disease research in Europe and beyond – why the UK must continue to collaborate

Victoria Hedley

Rare Disease Policy Manager

Newcastle University

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Day 2 – Weds 27th April 2022

13:00 – 13:10

Welcome day 2
Professor Michael Hanna

13:10 – 14:10

Session chairs:

Platform presentations 1 hour
Dr Rob Pitceathly, UCL and **Professor Bobby McFarland**, Newcastle University

4 x platform presentation of 10 min talk + 3 min questions

Biallelic variants in ARHGAP19 cause mixed demyelinating and axonal polyneuropathy
Natalia Dominik
UCL IoN

Analysis of muscle magnetic resonance imaging of a large cohort of chronic motor neuropathy/neuronopathy patients reveals characteristic features useful for diagnosis
Professor Jordi Diaz-Manera
Newcastle University

DOK7-AAV improved acetylcholine deficiency CMS in a mouse model and enhanced the effectiveness of Pyridostigmine treatment
Dr Yin Dong
University of Oxford

rAAV-mediated TCAP Gene Replacement Therapy: A Promising Therapeutic Approach for LGMDR7
Dr Liubov Gushchina
Nationwide Children's Hospital

14:10 – 14:20

MDUK update
Dr Kate Adcock
Director of Research and Innovation, MDUK

14:20 – 14:40

Comfort break and sponsor posters

Session 2

Session Chairs:

Professor Patrick Chinnery, University of Cambridge and **Professor Francesco Muntoni**, UCL

14:40 – 15:10

Systems biochemistry approaches to exploring mitochondrial protein function and disease pathogenesis
Dr David J. Pagliarini
BJC Investigator and the Hugo F. and Ina C. Urbauer Professor of Cell Biology & Physiology, Biochemistry & Molecular Biophysics, and Genetics. Washington University in St. Louis School of Medicine

15:10 – 15:40

Application of omics-technologies in congenital myopathies
Dr Jim Dowling
Senior Scientist, Genetics & Genome Biology, Hospital for Sick Children (Sick Kids) Toronto/ University of Toronto

15:40 – 15:55

Comfort break and sponsor posters

15:55 – 16:25

Implementing the UK Rare Diseases Framework
Dr Louise Wood CBE
Director: Science, Research & Evidence, Deputy CEO - National Institute for Health Research
Department of Health and Social Care

16:25 – 16:55

Therapeutic Approaches for FSHD
Professor Scott Harper
Center for Gene Therapy, Nationwide Children's Hospital // Ohio State University College of Medicine

16:55

Prizes and close
Professor Michael Hanna and Professor Mary Reilly

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