16th UK Neuromuscular Translational Research Conference
Developing advanced genetic therapies in neuromuscular diseases
UCL GOSH ICH, 30 Guilford Street, London WC1N 1EH
Kennedy Lecture Theatre

Wednesday 29 and Thursday 30 March 2023

PROGRAMME

Day 1 – Wednesday 29th 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: Developments in Gene Therapy technologies

Chairs:

09:45 – 10:15 **The role of NATA in the translational research panorama in the UK**
Professor Nick Lench
Executive Director, Nucleic Acid Therapy Accelerator (NATA)

10:15 – 10:45 **Developing an AAV gene therapy strategy for LGMD2A**
Dr Isabelle Richard
Director of Research, CNRS, France

10:45 – 11:15 Coffee

11:15 – 11:45 **RNA therapeutics for cardiac repair and regeneration**
Dr Mauro Giacca
Head of the School of Cardiovascular and Metabolic Medicine and Sciences
Kings College London

11:45 – 12:00	Platform presentation 1 **Treatment of congenital myasthenia using a novel AAV-DOK7 gene therapy** Dr Judith Cossins University of Oxford
12:00 – 12:15	Platform presentation 2 **Decoding the transcriptome of Duchenne muscular dystrophy to the single nuclei level reveals clinical-genetic correlations** Prof Jordi Diaz-Manera Newcastle University

12:15 – 12:35 ***Sponsored lunchtime symposium (followed by lunch)***
(optional and does not form part of the educational programme)

12:15 – 13:15 Lunch

13:15 – 14:45 **Guided poster sessions**

Session 2: Emerging Advanced Therapies in Neuropathies

Chairs:

14:45 – 15:15 **Genetic therapies for demyelinating CMT neuropathies**
Professor Kleopas A. Kleopa
Center for Neuromuscular Disorders and Department of Neuroscience, The Cyprus Institute of Neurology and Genetics

15:15 – 15:45 **Small molecule therapy for CMT2A due to MFN2 mutations**
Gerald W Dorn II, MD
Philip and Sima K Needleman Professor
Washington University School of Medicine

15:45 – 16:15 **Coffee and posters**

16:15 – 17:00 The fifth Morgan-Hughes Thomas lecture
Solving the undiagnosed neurogenetic diseases
Professor Henry Houlden
UCL Queen Square Institute of Neurology

17:00 – 17:15	Platform presentation 3 **Molecular Diagnosis of Facioscapulohumeral Muscular Dystrophy in low and middle-income countries in the ICGNMD consortium** Dr Richard Lemmers LUMC, Netherlands
17:15 – 17:30	Platform presentation 4 **Pathological variants in TOP3A cause distinct disorders of mitochondrial and nuclear genome stability** Dr Mahmoud Fassad Newcastle University
17:30 – 17:45	Platform presentation 5 **Exploring the therapeutic role of miRNA-X on RNA splicing in Spinal Muscular Atrophy** Mr Parth Patel UCL
17:45 – 18:00	Platform presentation 6 **Mutant allele-specific silencing of SPTLC1 by antisense oligonucleotides to treat Hereditary Sensory Neuropathy Type 1A** Dr Jinhong Meng UCL
18:00 – 18:15	**Late breaking scientific update**
18:15 – 18:45	Poster viewing
18:45	Drinks reception Goodenough College, Mecklenburgh Square, London WC1N 2AB
19:30	Networking dinner Goodenough College, Mecklenburgh Square, London WC1N 2AB

Day 2 – Thursday 30th March

Session 3: Advances in Mitochondrial Therapeutics Research

Chairs:

08:30 – 09:00	**Gene therapy in mouse models of mitochondrial disease** Dr Carlo Viscomi Dept of Biomedical Sciences University of Padova, Italy
09:00 – 09:30	**Generating mouse models of mtDNA disease** Dr Jim Stewart Biosciences Institute & Wellcome Centre for Mitochondrial Research Faculty of Medical Sciences, Newcastle University
09:30 – 10:00	**The role of mitochondrial S-Adenosylmethionine in health and disease** Dr Anna Wredenberg Principal Researcher, Wredenberg lab Karolinska Institutet, Sweden
10:00 – 11:30	Poster guided sessions and coffee (poster session commences at 10:10)
11:30 – 11:50	**MRC – UKRI update** Dr Joanna Latimer, Head of Neurosciences and Mental Health Board. MRC UKRI

11:50 – 12:50	**Poster flash sessions** **Congenital Myasthenic syndrome: a Brazilian cohort study** Dr Pedro Tomaselli USP, Brazil **Genotypic and phenotypic spectrum of ANO5-associated muscle disorders** Dr Elisabetta Ghimenton Newcastle University **Defining the nuclear genetic architecture of a maternally-inherited mitochondrial disorder** Róisín Boggan Newcastle University **Classification of GJB1 variants** Dr Chris Record UCL IoN **MTM1 overexpression prevents and reverts BIN1-related centronuclear myopathy**

Dr Jocelyn Laporte
IGBMC, Illkirch, France

The Prevalence and Mortality Analysis of McArdle Disease and other rare muscle glycogenoses in the UK
Dr Ezgi Deniz Arikan
UCL IoN

King Denborough Syndrome also links to the autosomal recessive STAC3 c.851G>C pathogenic variant in a South African paediatric neuromuscular disease cohort: is it still an entity?
Prof Izelle Smuts
Pretoria, SA

Interrogation of 5`UTR and splicing variants in ICGNMD Neuromuscular patients
Dr Heba Morsy
UCL IoN

Diagnostic yield and genetic insights from whole exome sequencing in a cohort of congenital myopathy/muscular dystrophy patients from the International Centre for Genomic Medicine in Neuromuscular diseases (ICGNMD)
Dr Luke Perry
UCL IoN

12:50 – 13:40 Lunch

Session 3 continued:

13:40 – 14:25 The fifth Victor Dubowitz Lecture
Gene editing in mitochondrial diseases
Dr Michal Minczuk
MRC Mitochondrial Biology Unit
University of Cambridge

14:25 – 14:40 **MDUK update**
Dr Kate Adcock
Director of Research and Innovation
MDUK

14:40 – 14:55	Platform presentation 7 **Mitochondrial DNA loss and mitochondrial dysfunction in liver are reversed by deoxynucleotide administration in mice** Prof Antonella Spinazzola UCL GOSH ICH
14:55 – 15:10	Platform presentation 8 **Clinical, electrophysiological and radiologic profile of Hirayama disease patients from a tertiary care institute in India** Dr Saranya Gomathy AIIMS, India
15:10 – 15:25	Platform presentation 9 **Quantifying Variability in Duchenne Muscular Dystrophy: Centiles by Age for the Rise from Floor Velocity and 10m Walk Run Velocity in Glucocorticoid-steroid Treated Boys** Georgia Stimpson UCL

15:25 – 15:55 Coffee and posters

Session 4: Evolving Applications and Impact of Genome Sequencing

Chairs:

15:55 – 16:25	**ICGNMD Genomic Medicine consortium** Professor M Hanna Director, UCL Institute of Neurology
16:25 – 16:55	**When is a variant in TTN pathogenic?** Dr Marco Savarese Folkhälsan Research Center University of Helsinki, Finland
16:55 – 17:25	**Solve-RD Project** Dr Holm Graessner Centre for Rare Diseases and Institute of Medical Genetics and Applied Genomics University Hospital Tübingen, Germany
17:25 – 17:35	Poster prizes and close