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**
Professor Michael Hanna
Director, MRC Centre and UCL Institute of Neurology

Session 1: Developments in Gene Therapy technologies

Chairs: Professor Francesco Muntoni (UCL GOSH ICH) and Professor Patrick Chinnery (University of Cambridge)

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 **Preclinical development of a gene therapy for calpainopathy**
Dr Isabelle Richard
Director of Research, CNRS, France

10:45 – 11:15 **Coffee**

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

Platform presentations

Chairs: Professor Francesco Muntoni (UCL GOSH ICH) and Professor Patrick Chinnery (University of Cambridge)

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** Professor Jordi Diaz-Manera Newcastle University

12:15 – 13:15	**Lunch**
13:15 – 14:45	Guided poster sessions

Session 2: Emerging Advanced Therapies in Neuropathies

Chairs: Professor Mary Reilly (UCL IoN) and Professor Rita Horvath (University of Cambridge)

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	**Mitochondrial-targeted small molecule therapy for CMT2A and other neurodegenerative conditions** 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** **Introduced by Professor Michael Hanna** **Solving the undiagnosed neurogenetic diseases** Professor Henry Houlden UCL Queen Square Institute of Neurology

Platform presentations

Chairs (ctd): Professor Mary Reilly and Professor Rita Horvath

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 – 18:30	**Poster viewing**
18:30	**Drinks reception** Goodenough College, Mecklenburgh Square, London WC1N 2AB
19:15	**Networking dinner*** Goodenough College, Mecklenburgh Square, London WC1N 2AB

**for those whose booking includes dinner*

Day 2 – Thursday 30th March

Session 3: Advances in Mitochondrial Therapeutics Research

Chairs: Professor Michael Hanna (UCO IoN) and Professor Robert McFarland (Newcastle University)

08:30 – 09:00 **Experimental gene therapy in mitochondrial disorders**
Dr Carlo Viscomi
Dept of Biomedical Sciences
University of Padova, Italy

09:00 – 09:30 **Generating mouse models of mitochondrial DNA 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)

Session 3 continued:

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

Chairs: Dr Robert Pitceathly (UCL IoN) and Dr Giovanni Baranello (UCL GOSH ICH)

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
Dr 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:

Chairs: Professor Michael Hanna and Professor Robert McFarland

13:40 – 14:25 **The fifth Victor Dubowitz Lecture**
Introduced by Prof Francesco Muntoni
The therapeutic potential of mitochondrial genome engineering
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

Platform presentations

Chairs: Dr Robert Pitceathly and Dr Giovanni Baranello

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 IoN
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:	Professor Volker Straub (Newcastle University) and Professor Henry Houlden (UCL IoN)
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: European Rare Disease genomic analysis and interpretation** 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** Prof Mary Reilly, Prof Michael Hanna, Dr Rob Pitceathly

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