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

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

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

	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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