

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
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- 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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