17th and 18th April 2024

Day 1 – Wednesday 17th April

09:00 – 09:30 **Registration and Coffee**

09:30 – 09:45 **Welcome and Introduction**

11:00 – 11:30 **Coffee**

Platform presentations

11:30 – 12:45

Platform presentation 1
DOK7-AAV9 gene therapy in a novel mouse model for Congenital Myasthenic Syndrome caused by mutations in CHRND
Dr. Setareh Alabaf
University of Oxford

Platform presentation 2
Antisense oligonucleotide mediated mutant Sptlc1 silencing as a therapeutic strategy for hereditary sensory neuropathy type IA: A proof of concept study in a mouse model
Dr. Jinhong Meng
UCL GOS Institute of Child Health

Platform presentation 3
Differential TfR1 expression patterns impact TfR1 targeting delivery strategies in neuromuscular disease models
Jessica Stoodley

Day 2 – Thursday 18th April

08:15 Coffee

08:30 - 08:45 **Welcome**

Session 2 (ctd): Applications of Exercise Therapy in the era of Gene Therapy in NMD

Chairs: Professor Francesco Muntoni (UCL GOSH ICH) and Dr. Anna Sarkozy (Great Ormond Street Hospital)

08:45 – 09:30 **Role of Physiotherapy Rehabilitation in the Landscape of Genetic Therapies**
Dr. Gita Ramdharry
Queen Square Centre for Neuromuscular Diseases
National Hospital for Neurology & Neurosurgery, UCLH
UCL Queen Square Institute of Neurology

Session 3: Mechanisms and New Advanced Therapeutics in Autoimmune Neuromuscular Disease

Chairs: Professor Michael Hanna (UCL IoN) and Professor Rob Pitceathly (UCL IoN)

09:30– 10:15

Using innovative Data Modelling methods to improve data quality- learning from Adult SMA REACH a real-world data collection study
Sonia Segovia Simon
John Walton Muscular Dystrophy Research Centre, Newcastle University

Interactome analysis of dystrophin isoforms in the mouse brain
Dr. Konstantina Tetorou
Dubowitz Neuromuscular Centre, Institute of Child Health, UCL

Retrospective deep phenotypic and genotypic analysis of UK patients with recessive early onset titinopathy
Dr. SiewMei Yap
Department of Neurology, Royal Free Hospital

Targeting adipogenic differentiation of fibro-adipogenic precursors in Duchenne muscular dystrophy
Priyanka Mehra
John Walton Muscular Dystrophy Research Centre, Newcastle University

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Platform presentations

Chairs: Professor Michael Hanna (UCL IoN) and Professor Rob Pitceathly (UCL IoN)

12:

Session 4: New Genomic Advances in Neuromuscular Disease

Chairs: Professor Rob Taylor (Newcastle University) and Professor Henry Houlden (UCL IoN)

16:00 – 16:35 **The Sixth Morgan-Hughes Thomas lecture**
Introduced by Professor Henry Houlden (UCL IoN)

Improved diagnosis of patients through systematic variant resolution
Professor Conrad (Chris) Weihl
Head of Weihl Lab, University of Washington in St Louis

16:35 – 17:05 **Gene Modifiers in Mitochondrial**

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