11th UK Neuromuscular Translational Research Conference
Fitzwilliam College, Cambridge

Thursday 19 and Friday 20 April 2018

Day 1 – Thursday 19th April

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: Genetic Therapies
Chairs: Professor Francesco Muntoni and Professor Volker Straub

09:45 – 10:15 Strategies for treating myotonic dystrophy
Dr Charles Thornton
University of Rochester

10:15 – 10:45 Therapeutic advances in hereditary ATTR amyloidosis
Professor Philip Hawkins
UCL Royal Free Hospital

10:45 – 11:15 Gene Delivery Translation: Lessons Learned
Dr Brian Kaspar
Avexis

11:15 – 11:45 **The Joana Domingos Memorial Lecture**
Oligonucleotide therapies for neuromuscular disease
Professor Matthew Wood
University of Oxford

11:45 – 12:30 Lunch
12:30 – 14:00 Guided poster sessions

Session 2: Myostatin Biology and Therapy
Chairs: Professor T Voit and Dr Ros Quinlivan

14:00 – 14:30 Myostatin expression in neuromuscular diseases
Dr Julie Dumonceaux
UCL GOS ICH

14:30 – 15:00 The maintenance of muscle mass through the neutralisation of Myostatin activity in diseases and aging
Professor Ketan Patel
University of Reading

15:00 – 15:30 Coffee and posters

15:30 – 16:00 **Platform presentations – 3 x 10 mins**

15:30 – 15:40 D01 Golodirsen Induces Exon Skipping Leading to Sarcolemmal Dystrophin Expression in Patients With Genetic Mutations Amenable to Exon 53 Skipping
Professor Francesco Muntoni
UCL GOS ICH

15:40 – 15:50 D10 Proteomic evaluation of Pip6a-PMO treatment for Myotonic Dystrophy type 1
Ashling Holland
University of Oxford

15:50 – 16:00 D42 Myostatin is a reliable biomarker for monitoring drug response in DMD
Dr Virginie Mariot
UCL

16:00 – 17:00 Targeting ubiquitin pathways to develop new therapies for neuromuscular disease
Professor Thomas Gillingwater
University of Edinburgh

17:00 – Fitzwilliam Lecture Theatre Building and Grounds Closed to MRC Conference until 07:30 on 20 April (accommodation will be accessible). Poster boards will not be accessible at this time.

19:00 Drinks reception and Gala dinner
Caius College Cambridge
(20 min walk – see porter's lodge at Fitzwilliam College for freephone taxi or public bus directions)

Day 2 – Friday 20th April

Session 3: Translational Challenges in Clinical Trials
Chairs: Professor Mary Reilly and Dr John Thornton

08:30– 09:00 Potential and challenges of using MRI/MRS as a biomarker/endpoint for clinical studies in neuromuscular disease
Dr Pierre Carlier
Institut de Myologie, France

09:00 – 09:30 The changing scenario of molecular genetic diagnostics for neuromuscular diseases
Dr Alessandra Ferlini
University of Ferrara, Italy

09:30 – 10:00 **Platform presentations – 3 x 10 mins**

09:30 – 09:40 PN 009 Plasma neurofilament light chain levels are raised in patients with inherited peripheral neuropathy and correlate with disease severity
Dr Alexander Rossor
UCL ION

09:40 – 09:50 Other 004 Identification of a novel kinase target in DM pathophysiology
Professor David Brook
University of Nottingham

09:50 – 10:00 NMJ+C 004 Testing a novel therapy in a MYO9A-related Congenital Myasthenic Syndrome zebrafish model
Emily O'Connor
Newcastle University

10:00 – 10:30 Coffee and posters

10:30 – 11:30 **Poster flash sessions**
Chaired by Prof. Michael Hanna, UCL Institute of Neurology

11:30 – 12:15 Guest lecture:
The genomic era: molecular genetics as the first line investigation **Professor Lucy Raymond**
University of Cambridge

12:15 – 12:45 Lunch
12:45 – 14:00 Guided poster sessions

Session 4: Advances in Mitochondrial Disorders
Chairs: Professor Patrick Chinnery and Professor Rob Taylor

14:00 – 14:30 Genes, modifiers and other oddities in mitochondrial translation deficiencies

Professor Rita Horvath
Newcastle University

14:30 – 15:00 Molecular diagnostics of Mendelian disorders via RNA sequencing
Dr Holger Prokisch
Institute for Human Genetics, Munich, Germany

15:00 – 15:30 Coffee and posters

15:30 – 16:00 Manipulating mitochondrial ROS production as a therapeutic strategy
Dr Mike Murphy
MRC MBU, University of Cambridge

16:00 – 16:40 **Platform presentations – 4 x 10 mins**

16:00 – 16:10 Mito 018 Understanding the mechanism underpinning the transmission of mtDNA mutations
Mikael Pezet
Cambridge University

16:10 – 16:20 Mito 007 Using zebrafish to model mitochondrial DNA depletion syndromes caused by DGUOK mutations

Juliane Muller
Newcastle University

16:20 – 16:30 Mito 004 Mitochondrial Genetic Diagnostics in Oxford: a 25 year journey of service developments and novel findings

Carl Fratter
Oxford University Hospitals

16:30 – 16:40 Mito 016 Clonally Expanded mtDNA Deletions in Human Skeletal Muscle Originate as a Proliferative Perinuclear Niche
Amy Vincent
Newcastle University

16:40 – 16:55 MDUK update (15 mins)
Robert Meadowcroft
MDUK

16:55 – 17:10 MRC update (15 mins)
Kathryn Adcock
MRC

17:10 – 17:30 **Poster prizes and close**

RCP CPD reference for this event: 117804