**UK Neuromuscular Translational Research Conference 2016**
*Medical Sciences Teaching Centre, Oxford, OX1 3PL*

Tuesday 22 and Wednesday 23 March 2016

## PROGRAMME

## Day 1 – Tuesday 22nd March

**08:45 – 09:15** **Registration and Coffee**

09:15 – 09:30 **Introduction**
Prof. Michael Hanna
UCL Institute of Neurology

**09:30–11:00** **Session 1: Genomic Therapies**
Chairs: Prof. Francesco Muntoni and Prof. Dame Kay Davies

09:30 – 10:00 **Alternate translational initiation of dystrophin: clinical and therapeutic implications**
Prof. Kevin Flanigan, Nationwide Children's Hospital, Ohio
(abstract S01)

10:00 – 10:30 **From pathogenesis to therapy in spinal muscular atrophy**
Prof. Francesco Muntoni, UCL Institute of Child Health
(abstract S02)

10:30 – 11:00 **Antisense targeting of 3'end elements involved in DUX4 mRNA processing is an efficient therapeutic strategy for Facioscapulohumeral Dystrophy: a new gene silencing approach**
Dr. Julie Dumonceaux, Institut de Myologie, Paris
(abstract S03)

11:00 – 11:30 **Coffee**

11:30 – 11:45 Platform presentation 1 (merged)
**Results of North Star Ambulatory Assessments in the Phase 3 Ataluren Confirmatory Trial in Patients with Nonsense Mutation Duchenne Muscular Dystrophy (ACT DMD)**

Prof. Francesco Muntoni, UCL Institute of Child Health
**ACT DMD: Effect of Ataluren on Timed Function Tests in Nonsense Mutation Duchenne Muscular Dystrophy**
Prof. Nathalie Goemans, University Hospitals Leuven, Belgium

(abstract P03 and P04)

11:45 – 12:00 Platform presentation 2
**Cell therapy for muscular dystrophy: lessons learned and a road to efficacy**
Prof. Giulio Cossu, University of Manchester
(abstract P02)

12:00 – 12:15 Platform presentation 3
**Charcot-Marie-Tooth and Centronuclear myopathy induced mechanistic impairment in endocytosis**
Tayyibah Ali, Imperial College London
(abstract P01)

12:15 – 12:30 Platform presentation 4
**Intestinal Pseudo-obstruction in Adult m.3243A>G-Related Mitochondrial Disease: An Under-Recognised and Poorly-Managed Clinical Entity**
Yi Ng, Newcastle University
(abstract P47)

**12:30 – 13:00 Lunch**

**13:00 – 14:00 Poster guided tours session 1 of 3**

**14:00 – 17:30 Session 2: Next generation biomarkers**
Chairs: Prof. Mary Reilly and Prof. Volker Straub

14:00 – 14:30 **Next generation in vivo imaging technologies in DMD - the BIOIMAGE-NMD programme**
Prof. Andy Blamire, Newcastle University
(abstract S04)

14:30 – 15:00 **MRI Biomarker Outcome Measures in Charcot-Marie-Tooth disease and Inclusion Body Myositis**
Dr. John Thornton, UCL Institute of Neurology
(abstract S05)

15:00 –15:30 **Integration of pharmacodynamic biomarkers into a drug regulatory pipeline: Vamorolone/VBP15 in DMD**
Prof. Eric Hoffman, ReveraGen BioPharma, Children's National Medical Center, Washington
(abstract S06)

**15:30 – 16:00** **Coffee**

**16:00 – 17:00** **Poster guided tour session 2 of 3**

17:00 – 17:30 **Session2: next generation biomarkers (ctd…)**
**Qualification of Novel Methodologies European regulatory perspective**
Dr. Maria Isaac, Senior Scientific Officer, EMA
(abstract S07)

17:30 – 17:45 Platform presentation 5
**Human, Fly and Cell models of Riboflavin Transporter Neuronopathy**
Andreea Manole, UCL Institute of Neurology
(abstract P66)

17:45 – 18:00 Platform presentation 6
**Impaired mitochondrial function in neuronal cells harbouring a dominant glycyl-tRNA synthetase mutation**
Veronica Boczonadi, Newcastle University
(abstract P65)

**18:00 – 18:15** **Robert Meadowcroft**
CEO, MDUK

18:15 – 19:00 Drinks

19:30 **Gala Dinner**
Keble College *OX1 3PG (10 mins walk)*
*(Dress code: smart / smart casual)*

## Day 2 – Wednesday 23rd March

08:30 – 09:30 **Poster guided tour session 3 of 3**

09:30 – 10:30 **Poster flash sessions** (see p.14 for list)
Chaired by Prof. Michael Hanna, UCL Institute of Neurology

**10:30 – 12:30** **Session 3: Big Data**
Chairs: Prof. Hanns Lochmuller and Prof. Thomas Voit

10:30 – 11:00 **A Human Phenotype Ontology (HPO)-driven whole-genome analysis framework for effective identification of pathogenic regulatory variants in Mendelian disease**
Prof. Peter Robinson, Charité Hospital Berlin
(abstract S08)

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| 11:00 – 11:30 | **Big data, large sequencing challenges, and the technology behind it**<br>Dr. Ivo Gut, CNAG (National Centre for Genomic Analysis), Barcelona<br>(abstract S09) |
| 11:30 – 12:00 | **Coffee** |
| 12:00 – 12:30 | **Neurology and Neurodegeneration Genomics England Clinical Interpretation Partnership (Neuro-GeCIP)**<br>Prof. Henry Houlden, UCL Institute of Neurology<br>(abstract S10) |
| 12:30 – 12:45 | Platform presentation 7<br>**Clinical and genetic analysis of CLCN1 mutations with dual inheritance pattern**<br>Dr. Emma Matthews, UCL Institute of Neurology<br>(abstract P94) |
| 12:45 – 13:00 | Platform presentation 8<br>**A GFPT1 deficient mouse model of Congenital Myasthenic Syndrome**<br>Yasmin Issop, Newcastle University<br>(abstract P98) |
| 13:00 – 14.00 | **Lunch** |
| 14:00 – 14:15 | Platform presentation 9<br>**Development of a cell-penetrating peptide for the delivery of antisense oligonucleotides to peripheral and CNS tissues of spinal muscular atrophy mice**<br>Suzan M Hammond, University of Oxford<br>(abstract P84) |
| 14:15 – 14:30 | Platform presentation 10<br>**Microvascular defect as potential peripheral target in spinal muscular atrophy**<br>Haiyan Zhou , UCL Institute of Child Health<br>(abstract P85) |
| 14:30 – 15.00 | **MRC Strategic plan**<br>Kathryn Adcock<br>Head of Neurosciences and Mental Health<br>Medical Research Council |
| 15:00 – 16:00 | **John Newsom-Davis Lecture**<br>**The congenital myasthenic syndromes: better treatments through an understanding of disease mechanisms** |

Prof. David Beeson, University of Oxford

**16:00 – 16:30 Poster prizes and close**

*To Note: Those displaying posters. To allow for judging and poster sessions, posters should only be put up and taken down during these times.*

*Day 1 Tuesday 22 March*
*Posters up: 8.45 am to 9.15 am*
*Posters down: 18:15 – 18.45 pm*

*Day 2 Weds 23 March*
*Posters up: evening of 22nd 18.45 pm onwards OR by 8.15 am latest 23 March (first poster session of this day is at 8.30 am). Building access is from 8am.*
*Posters down: 16.30 onwards*