



**Update in Neuromuscular Disorders**  
**Tuesday 7 – Friday 10 May 2019**  
***Clinical Neuroscience Lecture Theatre at 33 Queen Square, London WC1N***  
***3BG***

***DRAFT PROGRAMME***

**Day 1 – Tuesday**

**09:30 – 10.00 Registration**

**THEME: Duchenne muscular dystrophy: State of the art in translational medicine**

**10:00 – 10:45 Update on translational research in DMD**  
Prof Francesco Muntoni ICH Dubowitz Neuromuscular Centre

**10:45 – 11:30 Opportunities and challenges in AAV gene therapy for DMD**  
Prof Thomas Voit, NIHR GOSH UCL Biomedical Research Centre

**11:30 – 12.00 COFFEE**

***Theme: Update on Emery-Dreifuss muscular dystrophy syndromes***

**12:00 – 12:30 The Emery-Dreifuss muscular dystrophy syndromes**

**12:30 – 13:00 Case presentations**

**13.00 – 13.45 LUNCH**

**Theme Limb girdle muscular dystrophy and treatable mimics!**

**14:00 – 14:30 Diagnostic approach to limb girdle muscular dystrophy**  
Dr Anna Sarkozy, ICH Dubowitz Neuromuscular Centre

**14:30 – 15:00 Update on the spectrum of childhood myositis**

**15:00 – 15:30 Case presentations**

**15:00 - 15:30 TEA**

**Theme: Endocrine management of corticosteroid treated Duchenne muscular dystrophy**

**15:30 – 15:45 Glucocorticosteroids in DMD: Endocrine focus on standards of care**

Dr Adnan Manzur, ICH Dubowitz Neuromuscular Centre

**15:45 – 16:15 Management of emergencies in long term corticosteroid treated DMD**

**16:15 – 16.45 Bone health management in corticosteroid treated DMD**

**16:45 – 17:00 Discussion forum - endocrine management in corticosteroid treated DMD**

Facilitated by Dr Adnan Manzur

### **Day 2 – Wednesday**

**Theme Myopathies and skeletal channelopathies update**

**09:00 – 09:30 Update on the structural congenital myopathies**

**09:30 – 10:00 Paediatric channelopathies: diverse presentations and new treatments**

**10:15 – 10:45 Illustrative Case presentations**

**10:45 - 11:15 COFFEE**

**Theme: Spinal muscular atrophy (SMA5q)**

**11:15 – 11:45 Nutritional aspects in SMA**

**11:45 – 12:15 Translational research in SMA**

**12:15 – 12:45 Case presentations**

**12:45 – 13:00 Discussion forum on aspects of SMA management**

**13:00 – 14:00 LUNCH**

**Theme: Metabolic aspects in neuromuscular clinical practice**

**14:00 – 14:45 Update on treatable metabolic neuromuscular disorders**

**14:45 – 15:15 Myalgias, cramps and rhabdomyolysis**

Dr Ros Quinlivan, MRC Centre for Neuromuscular Diseases

**15:15 – 15.45 TEA**

**Theme: Clinical Pathological Conference (CPC)**

15.45 – 16:15 **CPC discussion**

16:15 – 17:00 **CPC conclusion**

17 :00 – 18:00 ***You are invited to join us for a drinks reception in the foyer***

***outside 33 Queen Square Lecture Theatre***

### **Day 3 - Thursday 9 May**

- 09:00 – 09:45**      **Diagnosis of inherited neuropathies in next generation sequencing (NGS) area: opportunities and challenges**  
Professor Mary Reilly, Queen Square (QS) Centre for Neuromuscular Diseases
- 09:45 – 10:15**      **Case presentations**  
Dr Menelaos Pipis and Dr Carolynne Doherty, QS Centre for Neuromuscular Diseases
- 10:15 – 11.00**      **Cerebellar ataxia, neuropathy, vestibular areflexia syndrome (CANVAS)- new repeat expansion disease**
- 11:00 – 11:30**      **COFFEE**
- 11:30 – 12:15**      **Genomics England and diagnosis of neuromuscular disease**
- 12:15 – 12:45**      **Whole exome studies in muscle disease**
- 12:45 – 13:45**      **LUNCH**
- 13:45 – 14.30**      **Toxic neuropathies**
- 14:30 - 15:15**      **Mononeuropathies: Approach, diagnosis and management**  
Dr Alex Rossor, QS Centre for Neuromuscular Diseases
- 15:15 – 15:45**      **TEA**
- 15:45 – 16:15**      **Adult mitochondrial disease – a practical approach**  
Dr Rob Pitceathly, QS Centre for Neuromuscular Diseases
- 16:15 – 17.00**      **Inflammatory myopathies – diagnosis and management**

**Day 4 – Friday 10 May**

- 09:00 – 09:45**                      **Channelopathies: Diagnosis and treatment**  
Professor Mike Hanna, Director, QS Institute of Neurology
- 09:45 – 10:15**                      **When is neurophysiology useful in muscle disease?**  
Dr Doreen Fialho, National Hospital for Neurology and  
Neurosurgery
- 10:15 – 10.45**                      **Adult limb girdle dystrophies – diagnosis and management**  
Dr Andria Merrison, Director, Bristol MND Centre, North Bristol  
NHS Trust
- 10-45 – 11:15**                      **COFFEE**
- 11: 15 – 12.00**                      **Nodal and paranodal antibodies in CIDP**  
Dr Simon Rinaldi, Nuffield Department of Clinical Neurosciences,  
Oxford
- 12:00 -12:45**                      **Paraproteins and neuropathy**
- 12:45- 13:30**                      **LUNCH**
- 13:30 – 14:15**                      **Genetic landscape of Motor Neurone Disease**  
Dr Pietro Fratta, QS Centre for Neuromuscular Diseases
- 14:15 – 14:45**                      **Treatment development of Motor Neurone Disease**
- 14:45 – 15:00**                      **TEA**
- 15:00 – 15:45**                      **Exercise and neuromuscular disease – what advice is right?**
- 15:45 – 16:30**                      **Pregnancy and neuromuscular diseases**  
Dr Matilde Laura, QS Centre for Neuromuscular Diseases