



**Update in Neuromuscular Disorders**  
**Tuesday 23 - Friday 26 May 2017**  
***Clinical Neuroscience Lecture Theatre at 33 Queen Square, London WC1N***  
***3BG***

***PROGRAMME***

**Day 1 – Tuesday 23 May**

**09:30 – 10.00 Registration**

**THEME            Duchenne and limb girdle muscular dystrophies**

**10:00 – 10:30 Motor function variability in DMD. Lessons from trajectory analysis project (TAP)**

Prof Eugenio Mercuri, Università Cattolica del Sacro Cuore

**10:30 – 11:15 Dystrophin protein - quality versus quantity - relevance for diagnosis and clinical trials**

Prof Francesco Muntoni, ICH Dubowitz Neuromuscular Centre

**11:15 – 11:30 Case presentations**

**11:30 – 12.00 COFFEE**

**12:00 – 12:30 Limb girdle muscular dystrophy- diagnostic update**

Dr Anna Sarkozy, ICH Dubowitz Neuromuscular Centre

**12:30 – 13:00 Case presentations**

**13.00 – 13.45 LUNCH**

**THEME            Spinal Muscular Atrophy**

**13:45 – 14:15 SMA1. variable phenotypes and impact on management**

Adnan Manzur ICH Dubowitz Neuromuscular Centre

**14:15 – 14:45 Nusinersen treatment in SMA1**

Prof Eugenio Mercuri, Università Cattolica del Sacro Cuore

**14:45 – 15:15 TEA**

**THEME Channelopathies and MHS**

**15:15 – 15:45 Skeletal muscle channelopathies in childhood**  
Dr Pinki Munot, ICH Dubowitz Neuromuscular Centre

**15:45 – 16:15 Case presentations**

**16:15 – 16:45 Malignant Hyperthermia and neuromuscular disorders**  
Dr Heinz Jungbluth, Evelina Childrens Hospital

**16:45 – 17:00 Case presentations**

### **Day 2 – Wednesday 24 May**

**THEME: Congenital myopathies, complications and mimics**

**09:00 – 09:30 Update on structural congenital myopathies**  
Prof Francesco Muntoni ICH Dubowitz Neuromuscular Centre

**09:30 – 10:00 The spectrum of Titinopathies**  
Dr Emily Oates, UCL Institute of Child Health

**10:15 – 10:45 Case presentations**

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

**11:15 – 12:00 Congenital Myasthenic Syndromes**  
Professor Stephanie Robb, Great Ormond Street Hospital

**12:00 – 12:30 Case presentations**

**12:15 – 12:45 Role of EMG in myopathies and their differential diagnosis**  
Dr Matthew Pitt, Great Ormond Street Hospital

**12:45 – 13:00 Case presentations**

**13:00 – 13:45 LUNCH**

<b>THEME</b>	<b>Workshop in respiratory standards of care in SMA 1</b>
<b>13:45 – 14:00</b>	<b>Audit of SMA1 outcomes at GOSH</b> Dr Anwaar Alanizi, Great Ormond Street Hospital
<b>14:00– 14:15</b>	<b>SMA1 respiratory management at GOSH</b> Dr François Abel, Great Ormond Street Hospital
<b>14:15 – 14.30</b>	<b>Symptom care team perspective</b> Dr Finella Craig Louis Dundas Centre, Great Ormond Street Hospital
<b>14:30 – 15.00</b>	<b>Anticipatory respiratory management in SMA1</b> Prof Anita Simonds, Royal Brompton Centre for Sleep
<b>15.00 – 15.30</b>	<b>Panel discussion - Optimal standards of respiratory care- consensus, outcomes, challenges?</b>
<b>15.30 – 16.00</b>	<b>TEA</b>
<b>16:00 – 16:30</b>	<b>CPC discussion</b> - inviting Dr Elizabeth Wraige, Evelina Children’s Hospital
<b>16:30 – 17:00</b>	<b>CPC conclusion</b> – Dr Adnan Manzur, ICH Dubowitz Neuromuscular Centre

### **Day 3 - Thursday 25 May**

- 09:00 – 09:45**     **Approach to clinical trials in neuropathy – what’s right and wrong?**  
Professor Richard Hughes, MRC Centre for Neuromuscular Diseases
- 09:45 – 10:15**     **Inflammatory neuropathies**  
Dr Mike Lunn, MRC Centre for Neuromuscular Diseases
- 10:15 – 11.00**     **Critical Care Neuromyopathy**  
Dr Robin Howard, National Hospital for Neurology and Neurosurgery
- 11:00 – 11:30**     **COFFEE**
- 11:30 – 12:15**     **Inflammatory muscle diseases; diagnosis and management**  
Professor David Hilton-Jones, Nuffield Department of Clinical Neuroscience, Oxford
- 12:15 – 12:45**     **Inclusion body myositis**  
Dr Pedro Machado, MRC Centre for Neuromuscular Diseases
- 12:45 – 13:45**     **LUNCH**
- 13:45 – 14.30**     **Diagnosis and management in muscle channelopathies**  
Prof Mike Hanna, UCL Institute of Neurology
- 14:30 - 15:15**     **Mitochondrial diseases in adults**  
Dr Rob Pitceathly, MRC Centre for Neuromuscular Diseases
- 15:15 – 15:45**     **TEA**
- 15:45 – 16:15**     **Muscular Dystrophies in adults**  
Dr Ros Quinlivan, MRC Centre for Neuromuscular Diseases
- 16:15 – 17.00**     **Axonopathies – what next?**  
Professor Mary Reilly, MRC Centre for Neuromuscular Diseases
- 17.00 – 18.00**     **Myelination and Assembly of Nodes of Ranvier: What’s the Connection?**  
Special guest speaker – Professor Peter Brophy University of Edinburgh
- 18.00- 19.00**     **Drinks and canapies**

**Day 4 – Friday 26 May**

- 09:00 – 09:30**                      **Kennedy’s disease; an update and expanding the phenotype**  
Dr Pietro Fratta, Sobel Department of Motor Neuroscience at the UCL Institute of Neurology
- 10:15 – 10.45**                      **MND – when diagnosis is difficult**  
Dr Katie Sidle, National Hospital for Neurology and Neurosurgery
- 09:30 – 10:15**                      **Update on the early diagnosis of Motor Neurone Disease**  
Dr Nikhil Sharma MRC Unit for Lifelong Health and Ageing, UCL
- 10-45 – 11:15**                      **COFFEE**
- 11: 15 – 12.00**                      **MRI in Muscle Diseases**  
Dr Jasper Morrow, MRC Centre for Neuromuscular Diseases
- 12:00 -12:45**                      **Myotonic Dystrophies**  
Dr Chris Turner, MRC Centre for Neuromuscular Diseases
- 12:45- 13:30**                      **LUNCH**
- 13:30 – 14:15**                      **Neuropathy/myopathy: converging entities**  
Dr Alex Rossor, MRC Centre for Neuromuscular Diseases
- 14:15 – 14:45**                      **Case presentations**  
Dr Andrea Cortese and Dr Emma Matthews, MRC Centre for Neuromuscular Diseases
- 14:45 – 15:00**                      **TEA**
- 15:00 – 15:45**                      **Peripheral Nerve Vasculitis**  
Dr Aisling Carr, MRC Centre for Neuromuscular Diseases
- 15:45 – 16:30**                      **B12 Folate Pathway Neurology**  
Dr Elaine Murphy, Charles Dent Metabolic Unit, National Hospital for Neurology and Neurosurgery