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
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
<table>
<thead>
<tr>
<th>Time</th>
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| 13:45 – 14:00 | Audit of SMA1 outcomes at GOSH  
Dr Anwaar Alanizi, Great Ormond Street Hospital |
| 14:00 – 14:15 | SMA1 respiratory management at GOSH  
Dr François Abel, Great Ormond Street Hospital |
| 14:15 – 14:30 | Symptom care team perspective  
Dr Finella Craig Louis Dundas Centre, Great Ormond Street Hospital |
| 14:30 – 15:00 | Anticipatory respiratory management in SMA1  
Prof Anita Simonds, Royal Brompton Centre for Sleep |
| 15:00 – 15:30 | Panel discussion - Optimal standards of respiratory care - consensus, outcomes, challenges? |
| 15:30 – 16.00 | TEA |
| 16:00 – 16:30 | CPC discussion - inviting Dr Elizabeth Wraige, Evelina Children’s Hospital |
| 16:30 – 17:00 | CPC conclusion – 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

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