24th Annual Neuromuscular Study Group Scientific Meeting

Day 1: Friday, September 22, 2023

6:30am-8:00am  Buffet Breakfast and Check-In

8:00am-8:20am  Welcome and State of the Neuromuscular Study Group
   Dr. Richard Barohn and Prof Michael Hanna

Session I: Motor Neuron and Neuromuscular junction
Moderator - Heidi Fuller, PhD

8:20am-8:40am  Simon Rinaldi, MBChB, PhD University of Oxford
   Unravelling inflammatory neuropathies

8:45am-9:05am  David Hermann, MBBCh University of Rochester Medical Center
   Towards Clinical Trial Readiness for Charcot Marie Tooth Neuropathies

9:10am-9:30am  Riccardo Zuccarino, MD NeMO Centre, Milan
   Respiratory updates in CANVAS

9:35am-9:55am  Stephan Zuchner, MD, PhD, FAAN University of Miami, Miller School of Medicine
   hereditary neuropathies

10:00am-10:15am  Refreshment/Exhibitor Break

10:15am-10:35am  James Howard, MD University of North Carolina at Chapel Hill
   Expanding Therapeutic Options in Myasthenia Gravis

10:40am-11:00am  Jon Katz, MD Sutter Health, California Pacific Medical Center, The Forbes Norris MDA/ALS Research and Treatment Center
   Updates on ALS
**Session II – Flash Presentations Moderator - Dr Michael Pulley, MD, PhD**

**11:05am-11:15am**  Marwah Almadhi, BSc Biomedical Sciences, MBChB Medicine (current)  
University of Manchester  

*Investing to Save: Evaluation of Unplanned Hospital Admissions of Neuromuscular Patients in Greater Manchester, UK.*

**11:17am-11:27am**  Michael Hehir, MD  
University of Vermont  

*Measuring Adverse Event Burden in Myasthenia Gravis: Retrospective Validation of the Adverse Event Unit (AEU) with MGTX Trial Data*

**11:29am-11:39am**  Elie Naddaf, MD  
Mayo Clinic  

*A systematic review and meta-analysis of the placebo*

**11:41am-11:51am**  Shaweta Khosa, MD  
University of Rochester, Center of Health and Technology Outcomes Division (CHet)  

*The Inclusion Body Myositis Health-Index (IBM-HI): Development of a Novel, Disease Specific Patient-Reported Outcome Measure for IBM in Clinical Trials*

**11:53am-12:03pm**  Zhihao He, MS  
University of Florida  

*Relationships of Lower Leg Fat Fraction among antagonistic and synergistic muscles and a potential Fat Fraction threshold for functional performance in Myotonic Dystrophy Type 1*

**12:05pm-12:15pm**  Katy Dodd, MBChB, MRCP  
University of Manchester  

*Mycophenolate is better tolerated than azathioprine in myasthenia gravis*

**12:17pm-12:27pm**  Iwona Skorupinska, MSc, BSc  
University College London  

*A UK experience of symptomatic treatment of myotonia with Lamotrigine*

**12:30am-1:30pm**  Lunch  

*Muscle Study Group Executive Committee Meeting Breakout*

**Session III Platform Presentations – Moderator Kris Kelly, DPT, MS, Edm**

**1:30pm – 1:45pm**  William Macken, MD, PhD  
University College London
Improving diagnostic rates for mitochondrial diseases using enhanced WGS analysis and RNA-seq.

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<tr>
<th>Time</th>
<th>Topic</th>
<th>Speaker(s)</th>
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<tbody>
<tr>
<td>1:50pm-2:05pm</td>
<td>Improving diagnostic rates for mitochondrial diseases using enhanced WGS analysis and RNA-seq.</td>
<td>Vinojini Vivekanandam, MBBS(Hons) University College London, Queen Square Institute of Neurology</td>
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<td>MEND: MExiletine versus lamotrigine in Non-Dystrophic Myotonia</td>
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<td>2:10pm-2:25pm</td>
<td>Scoliosis progression in type II SMA at the time of treatment: a comparative study with untreated patients</td>
<td>Giorgia Coratti, PhD Catholic University of Sacred Heart</td>
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<td>2:30pm-2:45pm</td>
<td>Remote Monitoring and Management of Myasthenia Gravis (REMOTE-MG): A Pilot Feasibility Study</td>
<td>Michael Hehir, MD University of Vermont</td>
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<td>2:50pm-3:00pm</td>
<td>Refreshment/Exhibitor Break</td>
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<td>3:00pm-5:30pm</td>
<td>Young Investigator Session/Evaluator Session Breakout; Coordinator Breakout</td>
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*ALL NEW*

Session Moderators – Dr W. David Arnold, Dr Karen Suetterlin, Dr Katherine Dodd, Dr Heidi Fuller, Prof Valeria Sansone, Marie Wencel

- How to give an effective elevator pitch
- Tips for successful Grant Writing
- Failing well, overcoming rejections, criticisms and changing direction

6:00pm-8:00pm  Abstract poster walk through session
8:00pm-9:00pm  Dinner
9:00pm-11:30pm  Reception

**Day 2: Saturday, September 23, 2023**

7:00am-8:00am  Buffet Breakfast
8:00am-8:15am  Dr Barohn and Prof Hanna Opening

Session V: Biomarkers (new) Moderator Donovan Lott

8:15am-8:35am  Lisa Hobson Webb, MD Duke University
Shear Wave Elastography in Neuromuscular Disease

8:40am-9:00am  Professor Anna Rostedt Punga, MD, PhD Uppsala University, Sweden
Unveiling the hidden regulators: Circulating microRNAs as biomarkers in Myasthenia Gravis

9:05am-9:25am  Robert Bowser, PhD Barrow Neurological Institute
Neurofilament as a biomarker for everything

9:30am-9:50am  Eric Wang, PhD University of Florida
Alternative splicing DM1

9:55am-10:15am  Glenn Walter, PhD University of Florida
MRI as a biomarker in MD

10:15am-10:30am  Refreshments/Exhibitor Break

Session VI: Aging Moderator – W. David Arnold

10:30am -10:50am  Thomas Wishart, BSc., M.B.A., PhD University of Edinburgh
proteomic analysis of differentially vulnerable synaptic populations to identify regulators of stability

10:55am - 11:15am  Matthew Birkbeck, PhD Newcastle University/Newcastle Upon Tyne Hospitals NHS Foundation Trust
Changes in Motor Unit MRI with age/sarcopenia

11:20am-11:40am  Roger Fielding, PhD Tufts University
Clinical aspects of sarcopenia

11:45am-1:00pm  Lunch

Session VII: Exercise – Moderator Miguel Chuquilin, MD
1:00pm-1:20pm  Elisa Duchesne, PHT, PhD  Université du Québec à Chicoutimi. Effect of exercise on functional outcomes and disease pathophysiology in DM1

1:25pm-1:45pm  Jackie Montes, PT, EdD, NCS  Columbia University Irving Medical Center  
*Exercise in SMA: More than just an intervention*

1:50pm-2:10pm  Tanja Taivassalo, PhD  University of Florida  
has multiple ongoing projects on exercise in DMD, impacts on functional outcomes and disease biomarkers

2:15pm – 2:35pm  Gita Ramdharry, PhD  University College London  
*Exercise, rehabilitation and physical activity in Charcot-Marie-Tooth disease*

2:40pm-3:10pm  Refreshments/Exhibitor Break

*Session VIII – Moderator*  W. David Arnold, MD

3:10pm-3:30pm  UCB Update

3:35pm-3:55pm  Argenx Update

4:00pm-4:20pm  Sarepta Therapeutics Update

4:25pm-4:45pm  Fulcrum Therapeutics Update

4:50pm-5:10pm  Catalyst Pharmaceuticals Update

5:15pm-5:35pm  Pfizer Update

7:30pm – 8:30pm  Robert C. Griggs Annual NMSG Keynote Speaker  
*The Circle of Translation*

Professor Mary M Reilly  
UCL Queen Square Institute of Neurology

8:30pm-9:30pm  Dinner

9:30pm-11:30pm  Evening Reception

**Day 3: Sunday, September 24, 2023**

7:00am-8:00am  Breakfast

8:00am-8:10am  Dr Barohn and Prof Hanna opening

*Session IX – NMSG Young investigators projects*

*Moderator* - Karen Sueterlin, MBBS, MRCP, PhD
8:10am-8:25am  2022 NMSG Fellow Tyler Rehbein, MD University of Rochester

Neuromuscular ultrasound as a biomarker to improve clinical trial readiness in Charcot Marie Tooth Neuropathies

8:30am-8:45am  2023 NMSG Fellow Dr Natalie Katz, MD, PhD Duke University

Development of novel imaging biomarkers for use in pediatric facioscapulohumeral muscular dystrophy

8:50am-9:05am  2022 Shark Tank Award update Alex Oldroyd MBChB, PhD, MSc, MRCP University of Manchester

Quantifying Idiopathic Inflammatory Myopathy-Associated Cancer Risk via Comprehensive Phenotyping of a Large UK-Wide Cohort

9:10am-9:25am  2022 Shark Tank Award update Giorgia Coratti, PhD Pediatric Neurology and Centro Clinico Nemo Rome

Predictive models in spinal muscular atrophy treated patients using machine learning

9:30am-10:45am  Shark Tank Session

Moderator: Aziz Shaibani, MD, FACP, FAAN, FANA
Nerve and Muscle Center of Texas

Shark Panel: Mazen Dimachkie, MD, Amanda Guidon, MD and Laurie Gutmann, MD

Presentations:
Determination of the unique genotypic/phenotypic characteristics of South East Asia FSHD1 patients in Singapore Dr Zhiyong Chen, UCL Queen Square Centre

Perceived Fatigability Tracker: Improving Assessment to Enhance Spinal Muscular Atrophy (SMA) Patient Outcomes Rafael Rodriguez-Torres DPT, Columbia University Irving Medical Center

Markers of respiratory progression in Duchenne Muscular Dystrophy – systemic inflammation and diaphragm thickness and motion (“DMD-InflUS”) Federica Trucco MD, PhD, University of Milan

LITEBulb SMA - Lingual Intervention of Tongue Exercises for BULBar SMA Carla Zingariello, DO University of Florida College of Medicine, Gainesville, FL
10:45am-11:00am Refreshments/Exhibitor Break

*Session X Emerging Phenotypes - Moderator* Dr. Katy Dodd, MBChB MRCP

11:00am-11:20am Aziz Shaibani, MD, FACP, FAAN, FANA Nerve and Muscle Center of Texas
*Neuromuscular implications of placebo research*

11:25am-11:40am James Lilleker, MBChB, PhD University of Manchester
Adult SMA

11:45am-12:05pm Amanda Guidon, MD, MPH Mass General Hospital/Harvard
*Neuromuscular complications of cancer immunotherapy*

12:10pm-12:30pm Katherine Mathews, MD University of Iowa Carver College of Medicine
Peds FSHD

12:35pm-12:55PM Valeria Sansone, MD, PhD University of Milan
*CNS and cognitive in SMA across the lifespan*

1:00pm-2:00pm Lunch

Closing