## WMS 2023 Summary Programme

### Monday 2nd October 2023

<table>
<thead>
<tr>
<th>Time</th>
<th>Event</th>
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<tbody>
<tr>
<td>08:30-19:00</td>
<td>Pre-Congress Teaching Course (separate registration required)</td>
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### Tuesday 3rd October 2023

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<tr>
<th>Time</th>
<th>Event</th>
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<tbody>
<tr>
<td>08:00-11:00</td>
<td>Pre-Congress Teaching Course (separate registration required)</td>
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<tr>
<td>11:00-16:00</td>
<td>WMS Executive Board Meeting  Meeting Room</td>
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<tr>
<td>15:30-18:00</td>
<td>Registration  Ballroom Foyer, refreshments  PAC Foyer and Exhibit Hall and poster set up  Ballroom Foyer</td>
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<tr>
<td>16:30-17:30</td>
<td>Industry Symposium 1  PAC</td>
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<tr>
<td>18:00-18:45</td>
<td>Opening Ceremony  PAC</td>
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<tr>
<td></td>
<td>INV01: The strength to explore: a review of NASA experience with muscle atrophy in space</td>
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<td>Don Thomas, Ohio Astronaut, USA</td>
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<td>Moderators: Lindsay Alfano &amp; Chris Weihl</td>
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<tr>
<td>18:45-21:00</td>
<td>Networking Reception  Congress Venue (separate registration required)</td>
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### Wednesday 4th October 2023

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<tr>
<th>Time</th>
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<tbody>
<tr>
<td>06:30-19:30</td>
<td>Registration desk open</td>
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<tr>
<td>07:30-08:30</td>
<td>Industry Symposium 3  PAC</td>
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<tr>
<td>09:00-09:15</td>
<td>Congress Welcome  PAC</td>
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<tr>
<td>09:15-09:45</td>
<td>Topic 1: Understanding phenotypic and genetic diversity in neuromuscular disorders 1</td>
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<td>Moderators: Gina Ravenscroft, University of Western Australia, Australia &amp; Marco Savarese, University of Helsinki, Finland</td>
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<td>INV02: High throughput functional assays to improve interpretation of rare variants discovered in Neuromuscular disease genes Monkol Lek, Yale School of Medicine, USA</td>
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<td>INV03: Understanding genetic variants in neuromuscular disorders  Chris Weihl, Washington University in St. Louis, USA</td>
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<tr>
<td>Time</td>
<td>Event</td>
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</table>
| 10:15-10:30  | **O01**: Clinical spectrum and molecular features of asymptomatic and paucisymptomatic DMD mutations  
              Stefan Nicolau, Nationwide Children’s Hospital, USA             |
| 10:30-10:45  | **O02**: Unpacking gene expression profile to the single nuclei level in human muscle Pompe samples  
              Jordi Diaz-Manera, Newcastle University, UK                      |
| 10:45-10:55  | **O03**: In vivo gene therapy for striated muscle laminopathy  
              Mariko Okubo, Institut de Myologie, France                     |
| 10:45-10:55  | **O04**: Myoguide.org: a web-based portal supporting the analysis of MRIs for the diagnosis of neuromuscular patients  
              Carla Bolaño Diaz, John Walton Muscular Research Centre, UK   |
| 10:45-10:55  | **O05**: TDP-43 dependent cryptic exon derived neoepitopes as a novel diagnostic biomarker in muscle biopsies of inclusion body myositis patients  
              Chiseko Ikenaga, Johns Hopkins University School Of Medicine, USA |
| 10:45-10:55  | **O06**: Clinical trial readiness and validation of onsite and remote evaluation in valosin containing protein-associated multisystem proteinopathy: A 24-month longitudinal study  
              Natalie Reash, Nationwide Children’s Hospital, USA             |
| 10:45-11:15  | **Morning refreshments & exhibition**: Exhibit Hall and posters  
              Ballroom                                                        |
| 10:45-11:15  | **Social Media Committee** find out about how to get involved  
              Myology Café, Exhibit Hall                                       |
| 11:15-13:15  | **PAC Topic 1**: Understanding phenotypic and genetic diversity in neuromuscular disorders  
              Gisèle Bonne, Centre de Recherche en Myologie, France & Charlotte Lilien, MDUK Oxford Neuromuscular Centre, UK  |
| 11:15-11:45  | **INV04**: Accounting for phenotypic variability in clinical outcome assessments  
              Lindsay Alfano, Nationwide Children’s Hospital, USA             |
| 11:45-12:15  | **INV05**: Engaging patients from diverse backgrounds in NMD research  
              Gita Ramdharry, UCL Queen Square, Institute of Neurology, UK   |
| 12:15-12:30  | **O03**: In vivo gene therapy for striated muscle laminopathy  
              Mariko Okubo, Institut de Myologie, France                     |
| 12:30-12:45  | **O04**: Myoguide.org: a web-based portal supporting the analysis of MRIs for the diagnosis of neuromuscular patients  
              Carla Bolaño Diaz, John Walton Muscular Research Centre, UK   |
| 12:45-13:15  | **O05**: TDP-43 dependent cryptic exon derived neoepitopes as a novel diagnostic biomarker in muscle biopsies of inclusion body myositis patients  
              Chiseko Ikenaga, Johns Hopkins University School Of Medicine, USA |
| 13:15-14:30  | **Lunch & exhibition**: Exhibit Hall and posters  
              Ballroom                                                        |
| 13:45-14:15  | **New WMS Members Event**  
              Myology Café, Exhibit Hall                                       |
| 14:00-15:30  | **Poster session 1**: Ballroom A-C  
              Refreshments served                                             |
| 15:30-16:00  | **Short Oral Presentations 1**: Ballroom C1  
              Moderator: Tahseen Mozaffar, University of California, USA       |
| 15:30-16:00  | **Short Oral Presentations 2**: Ballroom C2  
              Moderator: Laurent Servais, University of Oxford, UK             |
| 15:30-16:00  | **Short Oral Presentations 3**: Ballroom C3  
              Moderator: Jana Haberlová, Motol University Hospital, Czech Republic |
Thursday 5th October 2023

07:00-15:00 Registration desk open

08:00-09:00 Industry Symposium 7 PAC Industry Symposium 8 Exhibit Hall A1

09:30-11:00 PAC Topic 2: Pathobiology of neuromuscular expansion disorders 1
Moderators: Ichizo Nishino, National Institute of Neuroscience, NCNP, Japan & Louise Benarroch, Centre De Recherche En Myologie, France

09:30-10:00 INVO8: RAN translation in C9orf72 ALS/FTD and other repeat opportunities
Laura Ranum, University of Florida, USA

10:00-10:30 INVO9: Novel repeat disorders in muscle disease: the emergence of OPDM Zhaoxia Wang, Peking University First Hospital, China

10:30-10:45 O07: RAN translation of expanded CGG repeat in LR12 may contribute to oculopharyngodistal myopathy, Chengcheng Li, Washington University in St. Louis, USA

10:45-11:00 O08: Single-nucleus RNA sequencing reveals characteristic gene expression in pathologically-specific myofibers in oculopharyngodistal myopathy Nobuyuki Eura, Nara Medical University, Japan

16:15-17:00 PAC Debate: Is the muscle biopsy still indicated?
Moderators: Teerin Liewluck, Mayo Clinic-Rochester, USA & Edoardo Malfatti, Paris Est University/INSERM U955, France

INVO6: Teresinha Evangelista, Institut de Myologie, France

INVO7: Baziel van Engelen, Radboud University Medical Centre, The Netherlands

17:15-18:15 Poster session 2 Ballroom A-C Refreshments served

18:15-18:45 Short Oral Presentations 4 Ballroom C1
P398-P403 Moderator: Anna Sarkozy, Dubowitz Neuromuscular Centre, Great Ormond Street Hospital, UK

Short Oral Presentations 5 Ballroom C2
P168, P281-P285 Moderator: Meredith James, John Walton Muscular Dystrophy Research Centre, UK

Short Oral Presentations 6 Ballroom C3
P169-P172, P308-P309 Moderator: Vishnu Venuugopalan Thampy Yamuna, All India Institute of Medical Sciences, India

19:15-20:15 Industry Symposium 5 PAC Industry Symposium 6 Exhibit Hall A1
11:00-11:30  Morning refreshments & exhibition  Exhibit Hall and posters  Ballroom
11:00-11:30  Guidelines Committee find out about how to get involved  Myology Café, Exhibit Hall

11:30-13:30  PAC  
Topic 2: Pathobiology of neuromuscular expansion disorders 2  
Moderators: Gauthier Remiche, Hopital Erasme, Belgium & Nicol Voermans, Radboud University Medical Center, The Netherlands

11:30:12:00  INV10: Genetic discovery and pathomechanism of repeat disorders in neuromuscular diseases: lessons from RFC1  
Henry Houlden, UCL Queen Square, Institute of Neurology, UK

12:00:12:30  INV11: Motor neuron and muscle involvement in SBMA: therapeutic implications  
Kenneth Fischbeck, National Institutes of Health (NIH), USA

12:30:12:45  O09: Bi-allelic variants of FILIP1 cause congenital myopathy, dysmorphism and neurological defects  
Andreas Roos, University Medicine Essen, Germany

12:45:13:00  O10: Universal genomic newborn screening for early, treatable, and severe conditions including 33 genes of NMD: Baby Detect  
Tamara Dangouloff, University Of Liege, Belgium

13:00:13:15  O11: Long-read nanopore sequencing in FSHD patients reveals CpG methylation patterns including methylation gradients in contracted D4Z4 arrays  
Russell Butterfield, University of Utah, USA

13:15:13:30  O12: Muscle imaging in natural history of FSHD: quantitative MRI and ultrasound results compared head-to-head  
Sanne Vincenten, Radboudumc, The Netherlands

13:30-14:45  Lunch & exhibition  Exhibit Hall and posters  Ballroom
13:45-14:45  Career Development Workshop  Ballroom C1 (Lunch available in the room)  
Moderator: Chris Weihl, Washington University in St. Louis, USA  
Panel: Meredith James, John Walton, Muscular Dystrophy Research Centre, UK, Mike Lawlor, Medical College of Wisconsin, USA, Coen Ottenheijm, Amsterdam UMC, The Netherlands and Carmen Paradas, Hospital Virgen del Rocío, Spain

Friday 6th October 2023

06:45-18:00  Registration desk open
07:30-08:30  Interesting Case Discussions  PAC (Refreshments available)  
Cases presented by delegates  
Moderators: Reghan Foley, National Institute of Health, USA and Riyad El-Khoury, Genethon, France
08:30-08:45  Comfort break
08:45-10:00  PAC  
Topic 3: The effect of lifestyle, exercise and nutrition on neuromuscular pathology and outcomes 1  
Moderators: Salman Bhai, UT Southwestern, USA & Jean-Yves Hogrel, Association Institut de Myologie, France
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<tr>
<th>Time</th>
<th>Event</th>
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| 08:45-09:15  | **INV12:** The exposome in neuromuscular disorders  
Eva Feldman, The University of Michigan, USA |
| 09:15-09:45  | **INV13:** Development of a cycle training paradigm to improve exercise capacity and pathophysiology in boys with Duchenne muscular dystrophy,  
Tanja Taivassalo, University of Florida, USA |
| 09:45-10:00  | **O13:** Promoting an active lifestyle; use of an in-home body weight support system to increase exercise dosage for children with neuromuscular disease  
Megan Iammarino, Nationwide Children’s Hospital, USA |
| 10:00-10:30  | **Morning refreshments & exhibition**  
Exhibit Hall and posters |
| 10:00-10:30  | **Myology Developments Across the World and Education Committees**  
find out about how to get involved  
Myology Café, Exhibit Hall |
| 10:30-12:00  | **PAC**  
Topic 3: The effect of lifestyle, exercise and nutrition on neuromuscular pathology and outcomes  
Moderators: Linda Lowes, Nationwide Children’s Hospital, USA & John Vissing, Rigshospitalet, Denmark |
| 10:30-11:00  | **INV14:** Physical activity and exercise are more than medicine for neuromuscular disorders  
Nicole Voet, Radboud University Medical Centre, The Netherlands |
| 11:00-11:15  | **O14:** Experiences with pregnancy and pregnancy-related physiotherapy in women with Charcot-Marie-Tooth disease. A qualitative interview study  
Andreas Rosenberger, National Neuromuscular Centre, Norway |
| 11:15-11:30  | **O15:** Large-scale proteomics profiling of peripheral blood of DM1 patients identifies biomarkers for disease severity and physical activity  
Peter-Bram ’t Hoen, Radboud University Medical Center, The Netherlands |
| 11:30-11:45  | **O16:** New FDX2-loss of function phenotype presenting with blindness and myopathy with potential responsiveness to Co-enzyme Q10 analogs  
Reghan Foley, National Institute of Health, USA |
| 11:45-12:00  | **O17:** 6’-sialyllactose supplementation in GNE myopathy: a pilot and subsequent placebo-controlled study  
Young-eun Park, Pusan National University Hospital, South Korea |
| 12:15-13:15  | **WMS General Assembly/Poster viewing for non-members**  
Exhibit Hall A1 |
| 13:00-14:00  | **Lunch & exhibition**  
Exhibit Hall and posters  
Ballroom |
| 13:30-14:00  | **Sponsor Meeting**  
Meeting Room 10 |
| 14:00-15:00  | **Poster session 3**  
Ballroom A-C Refreshments served |

**P68-P70, VP71, P72-P81:** SMA - outcome measures  
P152-P162, VP163, P164-P166, VP167: Distal myopathies  
P191-P204: Registries  
P252-P258, VP259, P260, P262-P268: Dystrophinopathies  
P368, VP369, P370-P380, VP381, P382: Pompe disease
### Saturday 7th October 2023

<table>
<thead>
<tr>
<th>Time</th>
<th>Event</th>
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<tbody>
<tr>
<td>07:30-15:30</td>
<td>Short Oral Presentations 7</td>
</tr>
<tr>
<td>15:00-15:30</td>
<td>Ballroom C1</td>
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<tr>
<td>15:30-16:30</td>
<td>Poster session 4</td>
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<tr>
<td>16:30-16:45</td>
<td>Short Oral Presentations 10</td>
</tr>
<tr>
<td>18:00-18:30</td>
<td>Transport to Networking Pre-Dinner Drinks (separate registration required)</td>
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<tr>
<td>18:30-19:30</td>
<td>Pre-Networking Dinner Drinks Sharehouse, Downtown Charleston (separate registration required)</td>
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<tr>
<td>19:30-23:00</td>
<td>Networking Dinner The Bus Shed, Downtown Charleston (separate registration required)</td>
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### Summary Programme

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<tbody>
<tr>
<td>07:30-15:30</td>
<td>Short Oral Presentations 7</td>
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<tr>
<td>Ballroom C1</td>
<td>Moderators: Bjarni Udd, Tampere Neuromuscular Center, Finland</td>
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<tr>
<td>15:00-15:30</td>
<td>Short Oral Presentations 8</td>
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<tr>
<td>Ballroom C2</td>
<td>Moderator: Payam Mohassel, Johns Hopkins University, USA</td>
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<tr>
<td>15:30-16:30</td>
<td>Poster session 4</td>
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<tr>
<td>Ballroom A-C</td>
<td>Refreshments served</td>
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<tr>
<td>16:30-16:45</td>
<td>Short Oral Presentations 10</td>
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<tr>
<td>Ballroom C1</td>
<td>Moderator: Tina Duong, Stanford University, USA</td>
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<tr>
<td>18:00-18:30</td>
<td>Transport to Networking Pre-Dinner Drinks (separate registration required)</td>
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<tr>
<td>18:30-19:30</td>
<td>Pre-Networking Dinner Drinks Sharehouse, Downtown Charleston (separate registration required)</td>
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<td>Networking Dinner The Bus Shed, Downtown Charleston (separate registration required)</td>
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**Saturday 7th October 2023**

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<tr>
<td>07:30-15:00</td>
<td>Registration desk open</td>
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<tr>
<td>07:30-09:00</td>
<td>Arrival refreshments PAC Foyer</td>
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<tr>
<td>07:45-08:45</td>
<td>Clinical Trial Updates PAC</td>
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<td>Moderators: Kristl Claeys, Universitaire Ziekenhuizen Leuven, Belgium &amp; Ulrike Schara Schmidt, University of Essen, Germany</td>
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<td>O18: Topline Safety and Efficacy Data Analysis of Phase 1/2 Clinical Trial Evaluating AOC 1001 in Adults with Myotonic Dystrophy Type 1: MARINA™ Nicholas Johnson, Virginia Commonwealth University, USA</td>
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<td>O19: Preliminary Results from MLB-01-003: An Open Label Phase 2 Study of BBP-418 in Patients with Limb-girdle Muscular Dystrophy Type 21/R9 Amy Harper, Virginia Commonwealth University, USA</td>
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<td>O20: Safety and efficacy of intravenous onasemnogene abeparvovec in patients with spinal muscular atrophy: interim findings from the phase 3 SMART study Hugh McMillan, Children’s Hospital of Eastern Ontario, USA</td>
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<td></td>
<td>O21: 104-week efficacy and safety of cipaglucosidase alfa+miglustat in patients with late-onset Pompe disease previously treated with alglucosidase alfa Tahseen Mozaffar, University of California, USA</td>
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09:00-11:00  The Victor Dubowitz Lecture  
*PAC* 
_Moderators: Volker Straub, Newcastle University, UK & Chris Weihl, Washington University in St. Louis, USA_

09:00-09:30  RNA-targeted therapy for ALS  
_Tim Miller, Washington University, USA_

**Poster Highlights  
PAC**  
_Moderators: Alan Beggs, Boston Childrens Hospital / Harvard Medical School, USA & Svetlana Gorokhova, National Institute of Health, USA_

**O22:** P81 Gastrointestinal assessment in Spinal Muscular Atrophy (SMA): the experience of SMA healthcare professionals in France  
_Marta Gomez Garcia, APHP Raymond Poincare University Hospital, Child Neurology and Paediatric ICU Department Pediatrique, France_

**O23:** P161 Natural history of distal and myofibrillar myopathies assessed by clinical and technological outcome measures (Dista-Myo): baseline results  
_Giorgio Tasca, Newcastle University, UK_

**O24:** P266 Gene expression profiles and spatial localisation of dystrophin isoforms in developing and adult human brain  
_Francesco Catapano, University College London, UK_

**O25:** P325 A comparative single nuclei transcriptomics approach to evaluating the terminally differentiated lymphocytes in autoimmune Myositis  
_Francia Victoria De Los Reyes, National Center of Neurology and Psychiatry (NCNP), Japan_

**O26:** P350 Clinical characteristics and therapeutic response of patients with adult-onset Multiple Acyl-CoA-Dehydrogenase Deficiency (MADD)  
_Sofie Sunebo, Linköping University Hospital, Sweden_

**O27:** P425 Inhibition of TGFβ signaling pathway as a therapeutic approach in collagen VI-related muscular dystrophy  
_Hailey Hearn, Johns Hopkins University, USA_

11:00-11:30  Morning refreshments  
_PAC Foyer and posters Ballroom_

11:30-13:30  Late Breaking News  
*_PAC*_  
_Moderator: Lindsay Wallace, Nationwide Children’s Hospital, USA and Michele Yang, Children’s Hospital Colorado, USA_

**LBO01:** Impaired iron-sulfur cluster assembly due to biallelic variants in CIAO1 leads to a novel muscle disease  
_Rotem Or Bach, National Institute of Health, USA_

**LBO02:** Ablation of the Carboxyterminal end of MAMDC2 causesa distinctmuscular dystrophy  
_Carmen, Paradas, Hospital Virgen del Rocio, Spain_

**LBO03:** A novel class of Tubulinopathies - Mutations in TUBA4A cause primary skeletal muscle disorders  
_Mridul Johari, Harry Perkins Institute of Medical Research - UWA, Australia_

**LBO04:** CGG repeat expansion in LRP12 causes both amyotrophic lateral sclerosis and oculopharyngodistal myopathy type 1  
_Takashi Kurashige, Nho Kure Medical Center and Chugoku Cancer Center, Japan_

**LBO05:** Proteomic serum profiling identifies ITIH3 as a new biomarker for Myasthenia gravis disease activity  
_Tobias Ruck, Heinrich Heine University Düsseldorf, Germany_

**LBO06:** Functional improvements and decreased aggregate burden in TgT57I Mice following AAVrh74.tMCK. hBAG3 gene therapy  
_Burcak Ozes, Nationwide Children's Hospital, USA_

**LBO07:** RNA-based CRISPRoff silencing to target DUX4 in Facioscapulohumeral muscular dystrophy  
_Junjie He, Center For IPS Cell Research And Application (CIRA), Kyoto University, Japan_

**LBO08:** Identification of AAV variants with enhanced skeletal muscle and muscle stem cell transduction  
_Melissa Spencer, UCLA David Geffen School of Medicine, USA_

**LBO09:** scAAV9.U7-ACCA treatment of DMD exon 2 duplication leads to significant dystrophin expression and evidence of clinical benefit, particularly following treatment as an infant  
_Kevin Flanigan, The Research Institute at Nationwide Children’s Hospital, USA_
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<tbody>
<tr>
<td>13:30-14:30</td>
<td>Homeward lunch</td>
<td>PAC Foyer</td>
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<tr>
<td>13:30-15:00</td>
<td>NMD Board Meeting</td>
<td>Meeting room 6 &amp; 7 (separate registration required)</td>
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