Message from the NMSG Chair and Co-Chair:

We are once again very pleased to publish the abstracts and agenda for the annual NMSG meeting that will be held in Orlando, Florida on Sept 22, 23 and 24, 2023. This year’s meeting again will highlight many cutting-edge advances in the field of neuromuscular disease. We also give students, residents, fellows, and junior faculty the opportunity to meet with established leaders in the field, present their projects, and get advice on career advancement in academics.

At the time we write this introduction, over 225 individuals have registered for the meeting. We also have a record number of representatives from industry attending and we are proud to have many of them as sponsors for the three-day event. Five years ago, we established a Shark Tank session in which a NMSG member can pitch their research idea to a panel of neuromuscular sharks. The top project receives a grant to carry out their project. An instantly funded research grant!

This year the planning committee was led by Dr Dave Arnold, who is now at the University of Missouri in Columbia where he is the Executive Director of the NextGen Precision Health Initiative. Dave led an engaged group of NMSG members that put together an outstanding program. In addition, we had great work put in by Ladan Bigdeli, NMSG Student Doctor Editor, from The Ohio State University College of Medicine working with all 92 abstracts submitted.

We and the planning committee would like to thank the sponsors for generously supporting the meeting again including our top sponsors of UCB, Argenx, Catalyst Pharmaceuticals, Fulcrum, Pfizer and Sarepta Therapeutics.

We also want to thank Liz Paulk, the administrative manager of the NMSG. Liz works tirelessly year round with the NMSG leadership, planning committee, members, sponsors, and representatives at the site the meeting is held to make these meetings appear to come off seamlessly. The amount of work involved is enormous. And while Liz and her new assistant, Missy Apel are getting one meeting off the ground, they are already planning for the 2024 and the 2025 meetings. It is an ongoing dynamic process and that is a very good thing.

We are already planning the meeting for 2024 that will be in Tarrytown, New York at the Tarrytown House Estates on September 20-22nd.

We hope that many of the abstracts and presentations are ultimately transitioned to full articles that will be submitted to the RRNMFM Neuromuscular Journal in the upcoming months.

Richard J Barohn MD
Chair, NMSG
Columbia, Missouri, USA

Michael Hanna MD
Co-Chair NMSG
London, UK
Table of Contents

1. Pharmacological and Non-Pharmacological

72  #812 Long-Term Safety, Tolerability, and Efficacy of Efgartigimod in Patients With Generalized Myasthenia Gravis: Concluding Analyses From the ADAPT+ Study
73  #813 Long-Term Safety, and Efficacy of Subcutaneous Efgartigimod PH20 in Patients With Generalized Myasthenia Gravis: Interim Results of ADAPT-SC+
74  #798 Clinical-based prediction models for gastrostomy in patients with amyotrophic lateral sclerosis
75  #796 MEND: MExiletine versus lamotrigine in Non-Dystrophic Myotonia
76  #741 Investing to Save: Evaluation of Unplanned Hospital Admissions of Neuromuscular Patients in Greater Manchester, UK.
77  #764 Development of prediction models based on respiratory assessments to determine the need for Non-Invasive Ventilation in patients with Myotonic Dystrophy type 1
78  #746 A UK experience of symptomatic treatment of myotonia with Lamotrigine
79  #750 Safety and efficacy of ataluren in nmDMD patients from Study 041, a phase 3, randomized, double-blind, placebo-controlled trial
80  #752 Ataluren preserves upper limb function in nmDMD patients from Study 041, a phase 3 placebo-controlled trial, and the STRIDE Registry
81  #756 A Phase 1/2 Study of DYNE-251 in Males with DMD Mutations Amenable to Exon 51 Skipping: DELIVER Study Design
82  #757 A Phase 1/2 Randomized, Placebo-Controlled, Multiple Ascending Dose Study (ACHIEVE) of DYNE-101 in Individuals with Myotonic Dystrophy Type 1 (DM1)
83  #780 FREEDOM-DM1: Phase 1 Study to Assess Safety, Tolerability, Pharmacokinetics, and Pharmacodynamics of PGN-EDODM in Adults with Myotonic Dystrophy Type 1 (DM1)
84  #781 CONNECT-EDO51: Nonclinical and Phase 1 Data Support Phase 2 Trial Designs to Continue Evaluating Safety and Efficacy of PGN-EDO51 for Duchenne Muscular Dystrophy (DMD) Amenable to Exon 51 Skipping
85  #792 Phase 3b Extension Study Evaluating Superiority of Daily vs Approved On/Off Oral Edaravone Dosing in Patients With Amyotrophic Lateral Sclerosis
86  #804 Phase 1/2 Study to Evaluate the Safety, Tolerability, Pharmacokinetics, and Pharmacodynamic Effects of AOC 1020 Administered Intravenously to Adult Patients with Facioscapulohumeral Muscular Dystrophy (FORTITUDE) Trial Design
87  #805 Phase 1/2 Trial Evaluating AOC 1044 in Healthy Volunteers and Participants with DMD Mutations Amenable to Exon 44 Skipping (DMD44): EXPLORATION Trial Design
88  #772 Longer Milestone-Free Time in IV Edaravone-Treated vs Non–IV Edaravone-Treated Patients With Amyotrophic Lateral Sclerosis: An Administrative Claims Analysis
89  #773 PGN-EDO51, an Enhanced Delivery Oligonucleotide (EDO) Candidate for the Treatment of Duchenne Muscular Dystrophy (DMD): Positive Results from a Phase 1 Study in Healthy Volunteers
91  #809 Preliminary Results from MLB-01-003; An Open Label Phase 2 Study of BBP-418 in Patients with Limb-girdle Muscular Dystrophy 'Type 21'
92  #815 Empasiprabart (ARGX-117) in Multifocal Motor Neuropathy: Interim Baseline Characteristics of the Phase 2 ARDA Study
93  #825 Design Of A Phase 2, Multicenter, Randomized, Double-Blind, Placebo-Controlled, Parallel-Group Study Of Nipocalimab In Participants With Active Idiopathic Inflammatory Myopathies (Spiere
94  #737 Design of REACH: Phase 3 Randomized, Double-Blind, Placebo-Controlled, 48-Week Study of the Efficacy and Safety of Losmapimod in FSHD
96  #738 Safety and Tolerability of Losmapimod for the Treatment of FSHD
97  #744 A Phase 1/2a, Randomized, Double-Blind, Placebo-Controlled, First-in-patient Study of JM17 To Evaluate safety, Tolerability, Pharmacokinetics, and Pharmacodynamics in Adults with Spinal and Bulbar Muscular Atrophy
98  #806 Rozanolixizumab in Muscle-specific Kinase Autoantibody-positive Myasthenia Gravis: Further Analyses from MycarinG Study
100  #751 Ataluren preserves muscle function in nmDMD patients: a pooled analysis of results from three randomized, double-blind, placebo-controlled trials
101  #739 Therapeutic Play Gym (TPG): A Caregiver-Mediated Exercise System for Children with Neuromuscular Weakness – Feasibility and Extension Study
102  #732 Endosomal Escape Vehicle (EEV™) - Oligonucleotide Conjugates Produce Exon Skipping and Dystrophin Production in Preclinical Models of Duchenne Muscular Dystrophy
103  #842 Matching-adjusted indirect comparison of ravulizumab/efgartigimod in generalized myasthenia gravis: Timepoint challenges
105  #843 Assessing the extent of symptom control provided by ravulizumab or efgartigimod to patients with generalized myasthenia gravis (gMG)
108  #808 Long-term safety, efficacy & self-injection satisfaction with zilucoplan in myasthenia gravis: RAISE-XT interim analysis
### 2. Stratification of NMD and Disease Burden

<table>
<thead>
<tr>
<th>Paper Number</th>
<th>Title</th>
</tr>
</thead>
<tbody>
<tr>
<td>#740</td>
<td>Compound Muscle Action Potential Amplitude as a Biomarker of Myasthenia Gravis</td>
</tr>
<tr>
<td>#776</td>
<td>Burden of Myasthenia Gravis (MG) Based on Sentiment Analysis of Patients’ Digital Conversations</td>
</tr>
<tr>
<td>#770</td>
<td>Remote Monitoring and Management of Myasthenia Gravis (REMOTE-MG): A Pilot Feasibility Study</td>
</tr>
<tr>
<td>#771</td>
<td>Measuring Adverse Event Burden in Myasthenia Gravis: Retrospective Validation of the Adverse Event Unit (AEU) with MGTX Trial Data</td>
</tr>
<tr>
<td>#727</td>
<td>Treatment Preferences of Patients with Myasthenia Gravis</td>
</tr>
<tr>
<td>#779</td>
<td>Summated Compound Muscle Action Potential Amplitude as a Biomarker of Amyotrophic Lateral Sclerosis</td>
</tr>
<tr>
<td>#783</td>
<td>Factors Influencing Genetic Testing Uptake in Amyotrophic Lateral Sclerosis (ALS) Patients</td>
</tr>
<tr>
<td>#761</td>
<td>Utility of genetic panels for neuromuscular disorders in a tertiary referral center neurology clinic in Central Pennsylvania</td>
</tr>
<tr>
<td>#755</td>
<td>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</td>
</tr>
<tr>
<td>#821</td>
<td>Use Of Gas Scale To Identify Relevant Central Nervous System Domains In Myotonic Dystrophy Type 1: Diagnostic And Outcome Measure Prospective For Clinical Care And Trials</td>
</tr>
<tr>
<td>#789</td>
<td>Natural history of pulmonary function in adult patients with Spinal Muscular Atrophy type 2 and 3</td>
</tr>
<tr>
<td>#803</td>
<td>Scoliosis progression in type II SMA at the time of treatment: a comparative study with untreated patients</td>
</tr>
<tr>
<td>#797</td>
<td>Assessment of Patient-Reported Physical Fatigue in Spinal Muscular Atrophy (SMA): Insights from a Pilot Study</td>
</tr>
<tr>
<td>#745</td>
<td>Relationship of autoantibody status in dermatomyositis patients to response to IVIG treatment. A post-hoc analysis of the ProDerm trial</td>
</tr>
<tr>
<td>#819</td>
<td>Getting Ready For Trials Investigating Dysphagia Diagnostic And Outcome Measures In Myotonic Dystrophy Type 1 (Dm1): A Single-Center Retrospective Longitudinal Study</td>
</tr>
<tr>
<td>#816</td>
<td>Evaluation of ankle reflex and sural sensory nerve action potentials in a large patient cohort with cryptogenic peripheral polyneuropathy</td>
</tr>
<tr>
<td>#774</td>
<td>Development of a Novel, Disease-Specific, Patient-Reported Outcome Measure; the Myotonic Dystrophy Type 2 Health Index (MD2HI)</td>
</tr>
<tr>
<td>#769</td>
<td>Motor Outcomes to Validate Evaluations in Facioscapulohumeral muscular dystrophy (MOVE FSHD): Preliminary Baseline Characteristics</td>
</tr>
<tr>
<td>#820</td>
<td>Examining Recovery from Maximal Exercise Testing in Patients with Neuromuscular Disease</td>
</tr>
<tr>
<td>#791</td>
<td>Improving diagnostic rates for mitochondrial diseases using enhanced WGS analysis and RNA-seq.</td>
</tr>
<tr>
<td>#747</td>
<td>Introducing routine diagnostic Whole Genome Sequencing into the clinic</td>
</tr>
<tr>
<td>#784</td>
<td>Genetic And Epidemiology Characterization Of A Large Cohort Of Patients With Amyotrophic Lateral Sclerosis: Ten Years Of Experience In A Dedicated Neuromuscular Clinic In Italy (The Nemo Clinical Center)</td>
</tr>
<tr>
<td>#785</td>
<td>Magnetic resonance imaging and spectroscopy biomarkers for primary mitochondrial myopathies: preliminary results of a longitudinal study</td>
</tr>
<tr>
<td>#775</td>
<td>The Inclusion Body Myositis Health-Index (IBM-HI): Development of a Novel, Disease Specific Patient-Reported Outcome Measure for IBM in Clinical Trials</td>
</tr>
<tr>
<td>#807</td>
<td>Investigation into the Long-Term Prognosis of Patients with Sporadic Inclusion Body Myositis</td>
</tr>
<tr>
<td>#777</td>
<td>Rasch Analysis of the Patient-Reported Outcomes Measurement Information System (PROMIS) Parent Proxy (PP) Upper Extremity (UE) Item Bank Administered to Caregivers of Patients With Duchenne Muscular Dystrophy at Nationwide Children's Hospital</td>
</tr>
</tbody>
</table>
# Abstracts from the 2023 Neuromuscular Study Group Meeting

## 3. Mechanism of Disease and Less Common Disease Presentations

<table>
<thead>
<tr>
<th>#</th>
<th>Title</th>
</tr>
</thead>
<tbody>
<tr>
<td>#778</td>
<td>Comparison of Functional Ability Between 4–7 Years of Age (YOA) Children With Duchenne Muscular Dystrophy (DMD) to that of Typically Developing Age-Matched Children Using the Patient-Reported Outcomes Measurement Information System (PROMIS) Parent Proxy (PP) Mobility and Upper Extremity (UE) Questionnaires</td>
</tr>
<tr>
<td>#788</td>
<td>Pre- and post-natal outcomes in congenital and childhood onset DM1 - the impact of parental diagnostic delay</td>
</tr>
<tr>
<td>#673</td>
<td>MR Neurography and Quantitative Muscle MRI of Parsonage Turner Syndrome Involving the Long Thoracic Nerve</td>
</tr>
<tr>
<td>#733</td>
<td>Not just liver enzymes: Transaminitis as a marker of Immune Mediated Necrotizing Myopathy</td>
</tr>
</tbody>
</table>

## 4. Academic Registries and Patient Organizations

<table>
<thead>
<tr>
<th>#</th>
<th>Title</th>
</tr>
</thead>
<tbody>
<tr>
<td>#810</td>
<td>Nematodes deficient in TANGO2 homologs exhibit strong neuromuscular phenotype suggestive of bioenergetic dysfunction</td>
</tr>
<tr>
<td>#766</td>
<td>Amyotrophic Lateral Sclerosis and Spinocerebellar Ataxia Type 2: A Familial Case Report</td>
</tr>
<tr>
<td>#786</td>
<td>Characterisation of TDP-43 cryptic splicing</td>
</tr>
<tr>
<td>#763</td>
<td>Evaluation of the Role of Glial Factors in the Pathogenesis of Spinal Muscular Atrophy</td>
</tr>
<tr>
<td>#762</td>
<td>Proteomic characterisation of molecular pathways involved in Type III Spinal Muscular Atrophy</td>
</tr>
<tr>
<td>#724</td>
<td>Atypical presentation of chronic inflammatory demyelinating polyneuropathy</td>
</tr>
<tr>
<td>#760</td>
<td>Critical illness polyneuropathy/myopathy are associated with exposure to respiratory illness during critical care stays</td>
</tr>
<tr>
<td>#720</td>
<td>Review of Acute Rhabdomyolysis in Genetic Disorders vs Unaccustomed Exercise</td>
</tr>
<tr>
<td>#814</td>
<td>Late-onset autophagic vacuolar myopathy with sarcolemmal features.</td>
</tr>
<tr>
<td>#743</td>
<td>Hemodynamic response to exercise and mechanisms of exercise intolerance in patients with Myositis</td>
</tr>
<tr>
<td>#709</td>
<td>Myasthenic Syndrome Due to Tubular Aggregate Myopathy: A Case Report</td>
</tr>
<tr>
<td>#666</td>
<td>Late onset CMT2A can be a diagnostic challenge when presenting with vague sensory symptoms</td>
</tr>
<tr>
<td>#730</td>
<td>A stable human Schwann cell model of Charcot-Marie-Tooth disease type 1A</td>
</tr>
<tr>
<td>#736</td>
<td>Defining paretic neuromuscular pathophysiology in a mouse model of spinal cord injury</td>
</tr>
<tr>
<td>#765</td>
<td>Paramyotonia congenita in Zambia: A case report with broader implications for rare disease capacity building in sub-Saharan Africa</td>
</tr>
<tr>
<td>#817</td>
<td>Novel Mutations in the PLEC Gene: A Case of Epidermolysis Bullosa Simplex with Muscular Dystrophy</td>
</tr>
</tbody>
</table>

## 4. Academic Registries and Patient Organizations

<table>
<thead>
<tr>
<th>#</th>
<th>Title</th>
</tr>
</thead>
<tbody>
<tr>
<td>#753</td>
<td>Age at loss of ambulation in patients with DMD from the STRIDE Registry and the CINRG Natural History Study: a matched cohort analysis</td>
</tr>
<tr>
<td>#754</td>
<td>Pulmonary function in patients with Duchenne muscular dystrophy from the STRIDE Registry and CINRG Natural History Study: a matched cohort analysis</td>
</tr>
<tr>
<td>#794</td>
<td>MGBase: The launch of an international electronic database for patients with Myasthenia Gravis</td>
</tr>
<tr>
<td>#799</td>
<td>Patients in the Pompe Registry Who Switched From Alglucosidase Alfa to Avalglucosidase Alfa: Real-world experience</td>
</tr>
<tr>
<td>#841</td>
<td>Change In Concomitant Therapies For Generalized Myasthenia Gravis In Patients Receiving Eculizumab: A Retrospective Analysis Of Registry Data</td>
</tr>
<tr>
<td>#815</td>
<td>Introduction of the Peripheral Neuropathy Research Registry</td>
</tr>
<tr>
<td>#811</td>
<td>Comparison of Nerve Conduction Studies: Prediabetes, Type 2 Diabetes, Metabolic Syndrome and Cryptogenic Sensory Neuropathy</td>
</tr>
<tr>
<td>#802</td>
<td>Prevalence of Neuropathies and amyotrophic lateral sclerosis among adults in the United States: A cross-sectional study using the All of Us Research Program Database</td>
</tr>
</tbody>
</table>