

THURSDAY, SEPTEMBER 29

6:00 - 7:00 p.m. Check-In | *Hotel Regina Palace Lobby*

Welcome Reception | *Front Terrace*

DAY 1: FRIDAY, SEPTEMBER 30

7:00 - 8:00 a.m. Check-In | *Hotel Regina Palace Lobby*

Buffet Breakfast for
Hotel Regina Palace Guests
Hotel Regina Palace Lobby

8:00 - 8:20 a.m. Welcome and State of the
Neuromuscular Study Group
Lalique

**Dr. Richard Barohn and
Prof Michael Hanna**

SESSION I: BIG THERAPEUTIC WAVES

Moderator: James Lilleker, MBChB, Ph.D. | *Lalique*

8:20 - 8:40 a.m. FSHD
Jeffrey Statland, M.D.
University of Kansas Medical Center

8:45 - 9:05 a.m. Duchenne Muscular Dystrophy:
Learning From Our Failures –
a Clinical Trialists View
Laurent Servais | *University of Oxford*

9:10 - 9:30 a.m. The Evolving Therapeutic Options
for Pompe Disease
Mazen Dimachkie, M.D.
University of Kansas Medical Center

9:35 - 9:55 a.m. Update on treatment in CIDP and MMN
Edoardo Nobile-Orazio
*Milan University, IRCCS Humanitas
Research Institute*

10:00 -
10:15 a.m. Refreshment / Exhibitor Break
Azalea

10:15 -
10:35 a.m. Therapeutic Promise in Myotonic
Dystrophy Type 1
Nick Johnson M.D., Msci, FAAN
Virginia Commonwealth University

10:40 -
11:00 a.m. Muscle Dysfunction in Myotonia
Congenita
Mark Rich, M.D., Ph.D.
Wright State University

SESSION II: N&M IMAGING

Lalique

11:05 - 11:25 a.m. Muscle MRI as a tool for the diagnosis
and follow-up of myopathies
Georgio Tasca | *Fondazione Policlinico
Universitario A. Gemelli IRCCS*

11:30 - 11:50 a.m. Contribution of nerve and muscle
ultrasound in diagnosis and
management of peripheral
nerve diseases
Luca Padua, M.D., Ph.D.
*Fondazione Policlinico Universitario
A. Gemelli IRCCS*

12:00 - 1:30 p.m. Lunch | *Liberty Lago*

Neuromuscular Study Group Executive
Committee Meeting | *Rododendro*

3:15 - 4:45 p.m. Coordinators/Evaluators Session
Breakout | *Margherita*

3:15 - 4:45 p.m. Young Investigator Session Breakout
**Moderators: Prof Valeria Sansone &
Dr. Emma Ciafaloni** | *Lalique*

3:00 - 3:15 p.m. Refreshment / Exhibitor Break
Azalea

SHARK TANK SESSION

Moderator: Dr. Aziz Shaibani, M.D., FACP, FAAN, FANA
**Sharks: Tony Amato, M.D., Tahseen Mozaffar, M.D.,
Emma Ciafaloni, M.D.**
Lalique

Improving the Study of Falls in Muscle Disease
Kristina M. Kelly, PT, DPT, MS, EdM, NCS
The Ohio State University

Quantifying Idiopathic Inflammatory Myopathy Associated
Cancer Risk via Comprehensive Phenotyping of a
Large UK-Wide Cohort
Alexander Oldroyd, MBChB, Ph.D., MRCP, MSc
University of Manchester

Salivary Melatonin: Can this Detect Patients in Myotonic
Dystrophy Type 1 (DM1) with Disrupted Sleep Wake Cycle
and EDS?
Giovanni Colacicco, M.D.

Exploring the Experience of Pain for People with
Facioscapulohumeral Muscular Dystrophy and the effects of
Physiotherapy/Vojta Therapy
Jose Manuel Sanz Mengibar, Ph.D. Physiotherapy
University College London Hospital

Predictive models in spinal muscular atrophy treated
patients using machine learning
Giorgia Coratti, Ph.D.
Fondazione Policlinico Universitario Agostino Gemelli IRCCS

Therapeutic Dietary Intervention Attenuates Weight Gain
From Corticosteroid Use in Boys with DMD
Grace Warner Gough, MS, RDN, CD | *University of Utah*

6:30 - 8:00 p.m. Reception and poster walk through
session | *Azalea*

8:00 - 9:00 p.m. Dinner & Shark Tank Award
Announcement | *New Liberty*

9:00 - 11:30 p.m. Reception | *Bar Regina Palace*

DAY 2: SATURDAY, OCTOBER 1

7:00 - 8:15 a.m. Buffet Breakfast for Hotel Regina Palace Guests

8:15 - 8:30 a.m. Opening | *Lalique*
Dr. Richard Barohn and Prof Michael Hanna

SESSION III: MOTOR NEURON DISEASE CHALLENGES

Moderator: Dr. Senda Adjroud-Driss | *Lalique*

8:30 - 8:50 a.m. Respiratory management of patients with neuromuscular disease in ALS
Lisa Wolfe, M.D.
Northwestern University

8:55 - 9:15 a.m. Update on Clinical Trials for ALS
Angela Genge M.D., FRCP(c)
Montreal Neurological Institute and Hospital

9:20 - 9:40 a.m. Clinical Trials Landscaping in ALS
James Berry, M.D., MPH
Massachusetts General Hospital

9:45 - 10:05 a.m. SMA
Prof Eugenio Mercuri
Catholic University

10:10 - 10:30 a.m. Refreshments/Exhibitor Break
Hotel Regina Palace Lobby

SESSION IV: STRATIFICATION OF NM DISEASES

Moderator: Dr. Salman Bhai | *Lalique*

10:30 - 10:50 a.m. Seronegative Myasthenia Gravis
Vern Juel, M.D.
Duke University School of Medicine

10:55 - 11:15 a.m. Early detection of peripheral neuropathy in hATTR
Chafic Karam, M.D.
University of Pennsylvania

11:20 - 11:40 a.m. Updates in the therapeutic landscape of myositis
Julie Paik, M.D., MHS
Johns Hopkins University School of Medicine

11:45 a.m. - 12:05 p.m. Deconvoluting phenotypic complexity in myositis: from syndromes to diseases
Iago Pinal Fernandez, M.D., Ph.D., Ph.D.
Johns Hopkins Neurology Department

12:10 - 12:30 p.m. Myositis Autoantibodies: What's new?
Lisa Christopher Stine, M.D., MPH
Johns Hopkins University

12:30 - 1:30 p.m. Lunch
Liberty Lago

SESSION V: TECHNOLOGY AND DIGITAL OUTCOMES IN NMD

Lalique

1:30 - 1:45 p.m. Video capture and machine learning to assess hand myotonia and other functional timed tests
Tina Duong, PT, Ph.D.
Stanford University

1:50 - 2:05 p.m. Full day infant movement analysis: potential for wearable sensors to support early identification and assessment of neuromuscular diseases
Beth Smith PT, DPT, Ph.D.
Children's Hospital Los Angeles and University of Southern California

2:10 - 2:25 p.m. First regulatory qualification of a digital outcome in DMD: How can SV95c change the course of clinical developments
Laurent Servais | *University of Oxford*

2:30 - 2:45 p.m. Technology and digital outcomes in Neuromuscular Disorders: be to tech experience at the NeMO Lab
Stefano Regondi | *NeMO LAB*

2:45 - 3:00 p.m. Break | *Galle*

SESSION VI: ABSTRACT FLASH PRESENTATIONS

Lalique

3:00 - 3:15 p.m. Effect of different corticosteroid dosing regimens on clinical outcomes in boys with Duchenne muscular dystrophy: a randomized clinical trial
Robert Griggs, M.D. | *University of Rochester*

3:15 - 3:25 p.m. Caveolae-Associated Protein (cavin)-4 autoantibodies in immune mediated rippling muscle disease (iRMD)
Divyanshu Dubey, M.D., M.B.B.S. | *Mayo Clinic*

SPONSOR UPDATES

3:30 - 3:50 p.m. *Dyne*

3:55 - 4:15 p.m. *PTC*

4:20 - 4:40 p.m. *Roche*

4:45 - 5:05 p.m. *Alexion*

7:00 p.m. Meet for transportation to Isola Pescatori for evening activities

7:30 - 8:30 p.m. Robert C. Griggs Annual MSG Keynote Speaker: Neuromuscular Diseases - lessons learnt 1980-2020
Shree Pandya, PT, DPT
University of Rochester

8:30 p.m. Dinner

Evening reception on Borromean Islands following dinner

DAY 3: SUNDAY, OCTOBER 2

7:00 - 8:00 a.m. Buffet Breakfast for Hotel Regina Palace Guests

8:00 - 8:10 a.m. Opening | *Lalique*
Dr. Richard Barohn and Prof Michael Hanna

SESSION VII: NMSG YOUNG INVESTIGATORS PROJECTS

Lalique

8:10 - 8:25 a.m. Post-translational Modifications of DUX4
Renatta Knox, M.D., Ph.D., NMSG Fellow
Washington University School of Medicine

8:30 - 8:45 a.m. Neuromuscular ultrasound as a biomarker to improve clinical trial readiness in Charcot-Marie-Tooth Neuropathies
Tyler Rehbein, M.D., NMSG Fellow

8:50 - 9:10 a.m. 2021 Shark Tank Award Update: Circulating Myeloid Profile in Myasthenia Gravis
Katy Dodd, MBChB MRCP, Ph.D. Candidate
Manchester Centre for Clinical Neuroscience

9:10 - 9:25 a.m. 2021 Shark Tank Grant Award Update: Therapeutic Play Gym: Feasibility of a caregiver-mediated exercise system for infants and young children with severe neuromuscular weakness
Jenna Linn-Lammers, MSR/PT, CNT, PCS
University of Florida

10:45 - 11:00 a.m. Break | *The Regina Palace Lobby*

ABSTRACT PLATFORM PRESENTATIONS

9:27 - 9:37 a.m. A comparison of in silico predictive tools to robust in vivo functional characterisation of CLCN1 genetic variants in skeletal muscle channelopathies
Vino Vivekanandam, MBBS
QS University College London

9:39 - 9:49 a.m. Neuromuscular junction transmission failure is a translationally-relevant mechanism of sarcopenia
W. David Arnold, M.D.
University of Missouri

9:52 - 9:54 a.m. Symptom Onset In Maternally versus Paternally Inherited Myotonic Dystrophy type 2
Paloma Gonzalez-Perez, M.D., Ph.D.
Massachusetts General Hospital

9:56 - 10:06 a.m. Clinical trial readiness and validation of onsite and remote evaluation in valosin containing protein-associated multisystem proteinopathy
Megan Iammarino, DPT
Nationwide Children's Hospital

10:08 - 10:18 a.m. Bridging the preclinical-clinical gap: reverse translation of muscle velocity recovery cycles allows in vivo assessment of skeletal muscle excitability in mice and humans
Karen Suetterlin, MBBS, MRCP, Ph.D.
Newcastle University

10:20 - 10:30 a.m. Proposal for the functional assessment of acute inflammatory neuropathy (FAAIN) in Guillain-Barré syndrome
Dra. Zurina Lestayo O'Farrill, Ph.D.

10:32 - 10:42 a.m. Survival Motor Neuron Protein: Addressing Therapeutic Concerns of Sensorimotor Toxicity
Maria Balch, Ph.D.
The Ohio State University

10:44 - 10:54 a.m. Episodic weakness in patients with mitochondrial DNA MT-ATP6 mutations: the Queen Square experience
Chiara Pizzamiglio, Ph.D. Student

ABSTRACT FLASH PRESENTATIONS

11:00 - 11:20 a.m. **Salmon Bhai**

11:25 - 11:40 a.m. **Michela Coccia**

11:45 a.m. - 12:05 p.m. **Andrea Barp**

12:10 - 12:30 p.m. **Micheala Walker**

Closing

Lunch | *Liberty Lago*