



Neuromuscular Study Group

ANNUAL SCIENTIFIC MEETING

SEPTEMBER 30 - OCTOBER 2
HOTEL REGINA PALACE
STRESA, LAKE MAGGIORE, ITALY

Agenda

THURSDAY, SEPTEMBER 29

6:00 - 7:30 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
Liberty

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

SESSION I: BIG THERAPEUTIC WAVES

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

8:20 - 8:40 a.m. Near term prospects for disease targeted
therapies in 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
Prof Laurent Servais, M.D., Ph.D. | *University of Oxford, MDUK Oxford Neuromuscular Centre*

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
Prof Eduardo Nobile-Orazio, M.D., Ph.D. | *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

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

11:05 - 11:25 a.m. Muscle MRI as a tool for the diagnosis and follow-
up of myopathies
Giorgio Tasca, M.D., Ph.D. | *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*

12:00 - 1:30 p.m.

Neuromuscular Study Group Executive
Committee Meeting | *Rhododendro*

1:30 - 3:00 p.m.

Coordinators/Evaluators Session Breakout
Moderators: Melissa McIntyre, DPT, Marie Wencel, CCRP, Tina Duong, PT, Ph.D., Gita Ramdharry, Ph.D. | *Margherita*

1:30 - 3:00 p.m.

Young Investigator Session Breakout
Moderators: Prof Valeria Sansone & Dr. Emma Cialfoni | *Lalique*

3:00 - 3:15 p.m.

Refreshment / Exhibitor Break | *Azalea*

SHARK TANK SESSION

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

3:15 - 5:00 p.m.

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. | *NEMO Milan*

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
Dr. Richard Barohn and Prof Michael Hanna Lalique

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. Spinal muscular atrophy: hope vs hype; guide to handle patients' expectations
Prof Eugenio Mercuri, M.D., Ph.D. | 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. | Muscle Disease Unit, NIAMS/NIH | 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

Moderator: Prof Valeria Sansone | 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

Full day infant movement analysis: potential for wearable sensors to support early identification and assessment of neuromuscular diseases

1:50 - 2:05 p.m. **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

Prof Laurent Servais, M.D., Ph.D. | University of Oxford, MDUK Oxford Neuromuscular Centre

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

Dr. Stefano Regondi | NeMO Lab Milan

2:45 - 3:00 p.m. Break | *Hotel Regina Palace Lobby*

SESSION VI: ABSTRACT PLATFORM PRESENTATIONS

Moderator: Eli Naddaf, M.D. | 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 - 3:30 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-101 and DYNE-251: Moving from bench to clinic to deliver potentially transformative therapies in DM1 and DMD
Ashish Dugar, Ph.D. MBA | SVP, Global Medical Affairs, Dyne Therapeutics

3:55 - 4:15 p.m. Update on the growing body of evidence for Duchenne muscular dystrophy therapies
Dr. Christian Werner | Executive Director, Global Medical Affairs - Global DMD Lead, PTC Therapeutics

4:20 - 4:40 p.m. Our commitment to NMDs: the present and the near future
Kathryn R. Wagner, M.D., Ph.D. | Global Head Neuromuscular, Roche

4:45 - 5:05 p.m. Following the Science in Rare Neurological Diseases
Cinzia Dorigo, Pharm.D. | Alexion

ROBERT C. GRIGGS ANNUAL NMSG KEYNOTE SPEAKER

Lalique

7:00 p.m. Neuromuscular Diseases — lessons learnt 1980-2020
Shree Pandya, PT, DPT | University of Rochester

7:45 p.m. Meet for transportation to Isola Pescatori for evening activities

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
Dr. Richard Barohn and Prof Michael Hanna
Lalique

SESSION VII: NMSG YOUNG INVESTIGATORS PROJECTS

Moderator: Karissa Gable, M.D. | *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
University of Rochester

8:50 - 9:05 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

ABSTRACT PLATFORM PRESENTATIONS

9:30 - 9:40 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

9:45 - 9:55 a.m. Neuromuscular junction transmission failure is a translationally-relevant mechanism of sarcopenia
W. David Arnold, M.D.
University of Missouri

10:00 - 10:10 a.m. Symptom Onset In Maternally versus Paternally Inherited Myotonic Dystrophy type 2
Paloma Gonzalez-Perez, M.D., Ph.D.
Massachusetts General Hospital

10:15 - 10:25 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

10:30 - 10:45 a.m. Break | *The Regina Palace Lobby*

10:45 - 10:55 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

11:00 - 11:10 a.m. Proposal for the functional assessment of acute inflammatory neuropathy (FAAIN) in Guillain-Barré syndrome
Dra. Zurina Lestayo O'Farrill, Ph.D.

11:15 - 11:25 a.m. Survival Motor Neuron Protein: Addressing Therapeutic Concerns of Sensorimotor Toxicity
Maria Balch, Ph.D.
The Ohio State University

11:30 - 11:40 a.m. Episodic weakness in patients with mitochondrial DNA MT-ATP6 mutations: the Queen Square experience
Chiara Pizzamiglio, M.D., Ph.D. Candidate
QS University College London

ABSTRACT FLASH PRESENTATIONS

11:45 - 11:55 a.m. Risk factors for falls and fracture in myositis: A cross-sectional study of 470 patients
Salman Bhai, M.D. | *UT Southwestern*

11:57 a.m. - 12:07 p.m. Towards digital monitoring of Amyotrophic Lateral Sclerosis (ALS) patients: a deep learning-based application to assess the evolution of dysarthria via the analysis of multimedia data
Michela Coccia, M.D.
The Nemo Clinical Center in Ancona

12:10 - 12:20 p.m. Unmasking anti-HMGCR myopathy: the hurdles of a prompt recognition
Andrea Barp, M.D.
The Nemo Clinical Center in Trento

12:22 - 12:32 p.m. Motor Outcomes to Validate Evaluations in Facioscapulohumeral muscular dystrophy (MOVE FSHD): Protocol for an observational study
Micheala Walker, MPH, CCRP
University of Kansas Medical Center

Closing

12:35 - 1:35 p.m. Lunch | *Liberty Lago*

