2025

Neuromuscular Study Group

26TH ANNUAL SCIENTIFIC MEETING

SEPTEMBER 26-28, 2025
HOTEL REGINA PALACE
STRESA, LAKE MAGGIORE, ITALY

THANKS TO:

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Livestream Director, StreamGuru.net

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University of Copenhagen

Michaela Walker, MPH, CCRP, Coordinator Member
University of Kansas Medical Center

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Welcome

Dear Colleagues,

On behalf of the Neuromuscular Study Group (NMSG), we are delighted to welcome you back to Stresa, Italy for our 26th Annual Scientific Meeting.

For nearly three decades, the NMSG has united researchers from around the world to collaborate on advancing neuromuscular science. This meeting in particular has become a unique opportunity for our community to come together for a vibrant exchange of ideas that reinforces our commitment to progressing innovation in our field. Whether you are presenting a poster or discussing trials between sessions, we encourage you to engage fully with your colleagues and our industry partners as your active participation will greatly strengthen the value of our time together. A few meeting highlights for this year:

- **Young Investigators:** We welcome back this valuable session where emerging talent can connect with and learn from established leaders in the field. Join us at 3:35pm on Friday.
- **Abstract Posters Presentations:** We had an unprecedented number of submissions this year with 163. Please stop by to see their work at 6pm on Friday in the Tiffany room.
- **Industry Partners:** We appreciate the immense industry involvement from Europe and the United States. Please join us for their presentations starting at 1:45pm on Saturday and visit their tables daily in the Gallè room.
- **Neuromuscular Research 2-Year Fellowship Program:** We are proud to continue our partnership with the American Brain Foundation to fund this program. Our current fellow Sophie Rengarajan, M.D., Ph.D., will present her research just before our keynote session on Saturday at 6pm.
- **Shark Tank:** In its seventh year, the Shark Tank session will feature four exciting proposals with the winner receiving a $10,000 grant. New this year: Everyone who attends the session will be able to score the presentations. Join us at 9:45am on Sunday.

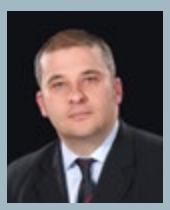
Outside of the scientific program, take time to enjoy the beauty of Stresa. With its elegant promenade, historic villas, and the famed Borromean Islands just offshore, Stresa offers a truly inspiring setting. We hope you will soak in the scenery, savor Italian hospitality and cuisine, and make the most of this charming location during your visit.

We are excited for the continuing opportunity to share breakthroughs, clinical insights, and new ideas that can improve the treatment and care of our patients. Let us continue to embrace the spirit of collaboration as we explore the latest research and inspire one another with our findings.

RICHARD J. BAROHN, M.D.

Chair, Neuromuscular Study Group

Executive Vice Chancellor for Health Affairs; Hugh E. and Sarah D. Stephenson Dean, School of Medicine, University of Missouri



PROF MICHAEL G. HANNA, M.D.

Co-chair, Neuromuscular Study Group

Director, University College London Institute of Neurology

Committees

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University of Kansas Medical Center

Gita Ramdharry, Ph.D.
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General Information

WIFI

The NMSG has a special wifi access for NMSG meeting attendees. This network can be used in all the meeting rooms and also the hotel.

Network name: Neuromuscular Study
Password: NMSG@LakeMaggiore

BREAKFAST

If you are staying at the Regina Palace, breakfast is from 7:00-10:00 a.m. in the Liberty Lago room. If you are staying at another hotel, please check with your front desk for details.

SATURDAY NIGHT GALA

If you signed up to attend the gala dinner, please gather at the back door of the hotel starting after the keynote address ends around 6:45 p.m. for the 10-minute drive to our dinner venue, La Tenuta del Golfo. The transport busses will run between the hotel and the villa all evening with the plated dinner service beginning at 8:00 p.m. The gala ends at 11:30 p.m.

Suggested dress for the evening is business or cocktail attire.

SPEAKERS

Please provide your presentation on a USB drive to Amardeep Gill, our onsite AV expert, as early as possible so your slides can be loaded into our laptop computer. He will be in the Lalique general session room during meeting hours starting on Thursday evening.

ABSTRACT POSTER PRESENTATION

The abstract poster presentation is in the Tiffany Room on Friday, September 26th. You can set up your posters starting on Friday morning. Please be next to your poster during the walk-though session from 6:00-8:00 p.m. and remove it after the session ends.

ABSTRACTS

Scan the QR code to view abstracts.

CONTINUING EDUCATION

NEUROMUSCULAR STUDY GROUP (NMSG)
26TH ANNUAL SCIENTIFIC MEETING
September 26-28, 2025
Stresa, Italy

Joint Accreditation Statement

JOINTLY ACCREDITED PROVIDER™
INTERPROFESSIONAL CONTINUING EDUCATION

In support of improving patient care, this activity has been planned and implemented by Amedco LLC and Neuromuscular Study Group (NMSG). Amedco LLC is jointly accredited by the Accreditation Council for Continuing Medical Education (ACCME), the Accreditation Council for Pharmacy Education (ACPE), and the American Nurses Credentialing Center (ANCC), to provide continuing education for the healthcare team. Amedco Joint Accreditation #4008163.

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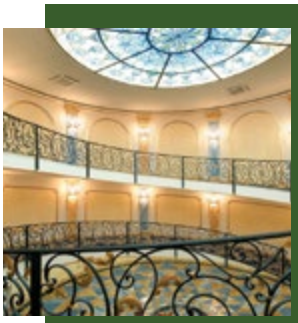
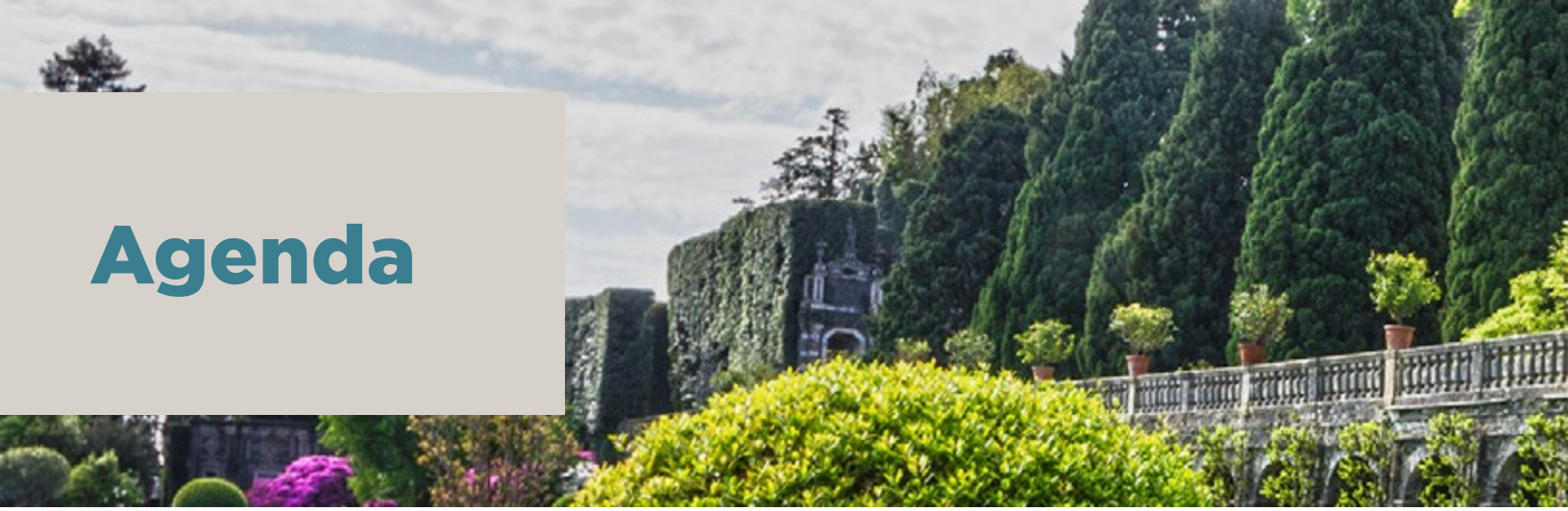


GROUP PICTURE

Don't miss the group picture in the hotel rotunda at 8:05 p.m. on Friday night!



Agenda

DAY 1: FRIDAY, SEPTEMBER 26

7:00 a.m. Breakfast at your hotel

WELCOME
Lalique

8:00 a.m. Welcome
Richard J. Barohn, M.D. & Prof Michael Hanna

8:15 a.m. Opening: Scientific Meeting Summary
Valeria Sansone, M.D., Ph.D. and Emma Ciafaloni, M.D., Planning Chairs

SESSION I: GENE BASED THERAPEUTICS
Moderators: Sophie Rengarajan, M.D., Ph.D., & Prof John Vissing, M.D.
Lalique

8:25 a.m. Management of Select Adverse Events Following Delandistrogene Moxeparvovec Gene Therapy for Patients with Duchenne Muscular Dystrophy
Craig Zaidman, M.D.
WashU Medicine

8:50 a.m. Hepatotoxicity and its management after gene therapy
Prof Anil Dhawan, M.D., FRCPCH
King's College Hospital, London

9:15 a.m. Newborn Screening for Pediatric Neuromuscular Disorders
Bo Hoon Lee, M.D.
University of Rochester

9:40 a.m. Building an infrastructure for safe and efficient gene therapy administration
Emma Ciafaloni, M.D.
University of Rochester

10:05 a.m. Refreshment/Exhibitor Break | *Gallè*

SESSION II: PLATFORM PRESENTATIONS
Moderators: Gita Ramdharry, Ph.D., & James Lilleker, MBChB, MRCP, Ph.D.
Lalique

10:20 a.m. Side-by-side evaluation of systemic AAV8, 9 and rh74 transduction in human muscle
Dongsheng Duan, Ph.D.
University of Missouri

10:40 a.m. The Role of Epigenetic Regulation of DMPK in Congenital Myotonic Dystrophy
Julia Hartman, M.D./Ph.D. Candidate
Virginia Commonwealth University

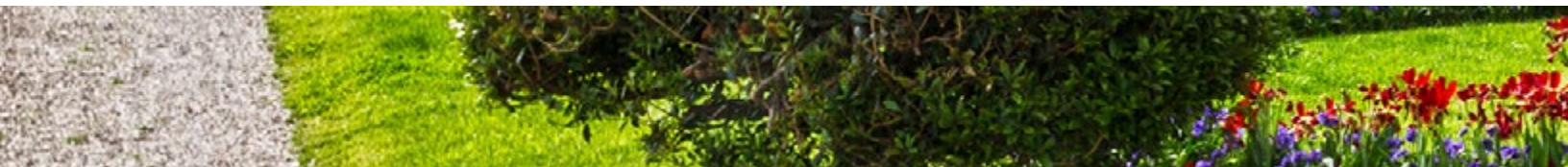
11:00 a.m. 100 Adults with Spinal Muscular Atrophy at the Dawn of Treatment: A Bone Health Focus
Dr. Rebecca Johnson
National Hospital for Neurology and Neurosurgery

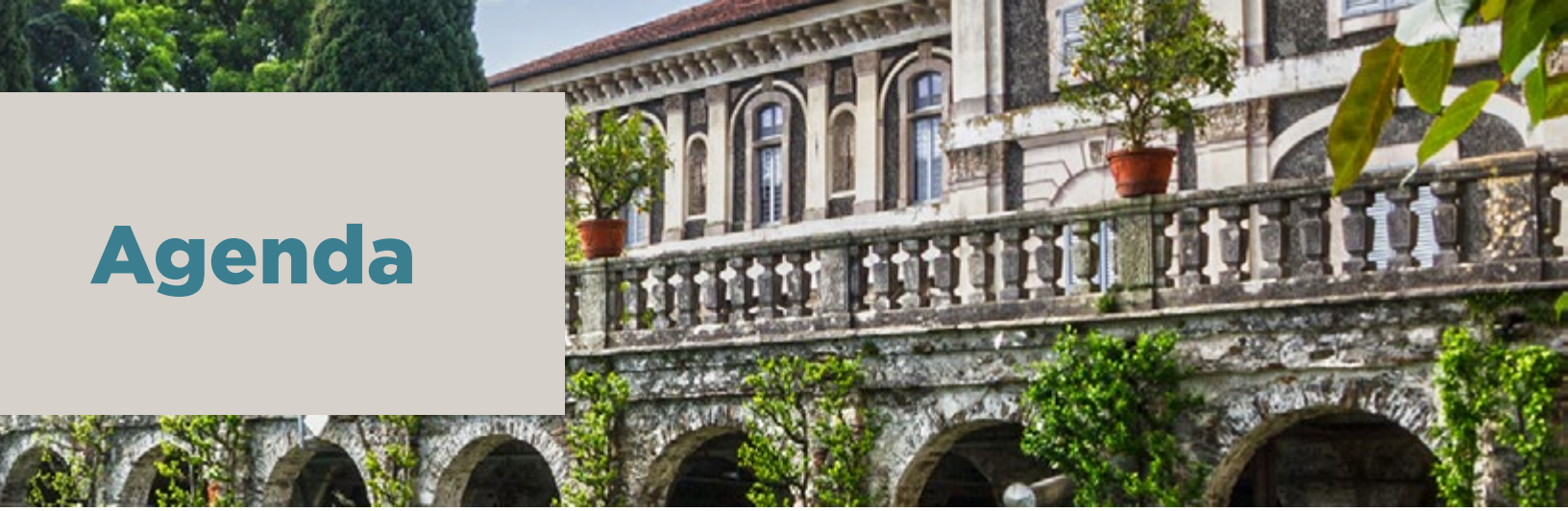
11:20 a.m. Nutritional and Swallowing Assessment in Patients with Myotonic Dystrophy Type 1: A Cross-Sectional Study
Andrea Barp, M.D.
Centro Clinico NeMO Trento

11:40 a.m. Uncovering Brain Structure and Cognitive Dysfunction in Myotonic Dystrophy Type 2
Araya Puwanant, M.D.
Wake Forest University School of Medicine

12:00 p.m. Lunch | *Liberty Lago*

NMSG Executive Committee Meeting Breakout Lunch | *Borromean Room*





Agenda

SESSION III: FLASH PRESENTATIONS

Moderators: Salman Bhai, M.D., & Jose Manual Sanz Mengibar, Ph.D.
Lalique

1:15 p.m. Altered BCR signaling mediates defects in early B cell tolerance checkpoints: insights from single cell analysis of NF155-mediated autoimmune nodopathies
Bhasker Roy, M.D.
Yale University School of Medicine

1:22 p.m. Development and validation of a deep learning-based facial weakness score for objective assessment in facioscapulohumeral muscular dystrophy
Karlein Mul, M.D., Ph.D.
Radboud University Medical Center

1:29 p.m. An omics-based investigation of changes at the molecular level in differing severities of Spinal Muscular Atrophy
Sharon Owen, Ph.D.
Keele University

1:36 p.m. A Phase 2, Open Label Trial of Repeated Intrathecal Autologous Adipose-Derived MSCs in ALS
Nathan Staff, M.D., Ph.D.
Mayo Clinic

1:43 p.m. Exploring accuracy and utility of artificial intelligence in the real-world management of myasthenia gravis
Sara Francesca Santagostino, Ph.D.
University of Vermont

SESSION IV: CLINICAL TRIAL UNITS

Moderators: Emma Ciafaloni, M.D., & Jasper Morrow, MBChB, Ph.D.
Lalique

1:50 p.m. Establishing a clinical trial site from A to Z
Yessar Hussain, M.D.
Austin Neuromuscular Center
Aziz Shaibani, M.D.
Nerve and Muscle Center of Texas

2:15 p.m. Placebo response and placebo effect: is there a difference?
Fabrizio Benedetti, M.D.
University of Turin Medical School

2:40 p.m. Setting up a Clinical Trial Unit: the experience at the NeMO site
Valeria Sansone, M.D., Ph.D.
NeMO Milan

3:05 p.m. Allied Health professionals in Clinical trial delivery — a QS experience
Gita Ramdharry, Ph.D.
University College London

3:25 p.m. Refreshment/Exhibitor Break | *Gallè*

SESSION V: YOUNG INVESTIGATOR, EVALUATOR AND COORDINATOR SESSION

Lalique

3:35 p.m. Panel
Mamatha Pasnoor, M.D., Emma Ciafaloni, M.D., Valeria Sansone, M.D., Ph.D., Chad Heatwole, M.D., Nicholas Johnson, M.D., Karen Suetterlin, MBBS, MRCP, Ph.D.

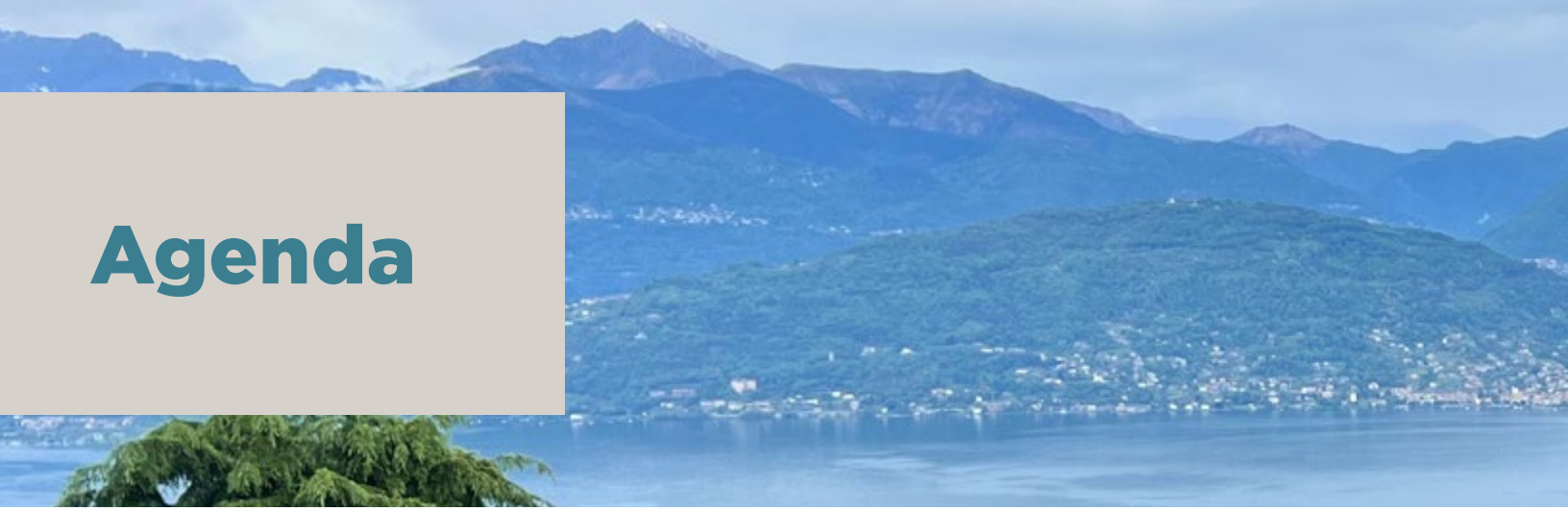
EVENING ACTIVITIES

6 - 8 p.m. Abstract poster session | *Tiffany*

8:05 p.m. Group picture | *Rotunda*

8:15 p.m. Dinner | *New Liberty*

9:30 p.m. Reception | *Hotel Bar*



Agenda

DAY 2: SATURDAY, SEPTEMBER 27

7:00 a.m. Buffet Breakfast at your hotel

WELCOME
Lalique

8:00 a.m. Welcome
Richard J. Barohn, M.D. & Prof Michael Hanna

Opening
Richard J. Barohn, M.D. & Prof Michael Hanna

SESSION VI: AI/E-HEALTH
Moderators: Prof Eugenio Mercuri & Dr. Nicholas Streicher
Lalique

8:15 a.m. MRI as a biomarker in neuromuscular disorders
John Thornton, Ph.D.
University College London

8:40 a.m. AI in healthcare. How can it help and where do we stand?
Saverio d'Amico, MSc
IRCCS Humanitas Research Hospital

9:05 a.m. Clinical Trials Powered by AI
Gaelen Ritter
Global Head of Digital Clinical Development, Sanofi

SESSION VII: DEBATE
Moderator: Prof Michael Hanna
Lalique

9:30 a.m. To Treat or not to Treat
Angela Genge, M.D.
McGill University
Prof John Vissing, M.D.
Copenhagen University Hospital

10:15 a.m. Refreshment/exhibitor break | *Gallè*

SESSION VIII: OUTCOME MEASURES AND HOW TO APPROACH REGULATORY REQUIREMENTS: LESSONS LEARNED
Moderators: Michaela Walker, MPH, CCRP, & Jackie Montes
Lalique

10:30 a.m. From clinical trials to drug approval: lessons learnt for regulatory success
Chad Heatwole, M.D.
University of Rochester

10:55 a.m. One size does not fil all: Lessons learnt from DMD
Prof Eugenio Mercuri, M.D., Ph.D.
Catholic University, Rome

11:25 a.m. Lunch | *Liberty Lago*

SESSION IX: SMORGASBORD NEW CLINICAL PHENOTYPES
Moderators: Paloma Gonzalez Perez, M.D., Ph.D., & Renatta Knox, M.D., Ph.D.
Lalique

12:15 p.m. New phenotypes and challenges in Myasthenia Gravis
Michael Hehir, M.D.
University of Vermont

12:40 p.m. New phenotypes and outcome measures for myotonic dystrophy
Johanna Hamel, M.D.
University of Rochester Medical Center

1:05 p.m. Clinical features of the peripheral neuropathies that occur in large arteriole vasculitis as compared to nerve microvasculitis
P. James Dyck, M.D.
Mayo Clinic

1:30 p.m. Refreshment/exhibitor Break | *Gallè*

Agenda

SESSION X: INDUSTRY UPDATES

Moderators: Amanda Guidon, M.D., MPH, & Dr. Enrico Bugiardini
Lalique

1:45 p.m. Targeted by Design: Avidity's Antibody-Oligonucleotide Conjugates (AOC™) for the potential treatment for Neuromuscular Disease
Joshua Lilienstein, M.D.
DMD Global Medical Lead Medical Affairs, Avidity Biosciences

2:10 p.m. A Case for Complement Inhibition: Clinical Activity of Riliprubart, an Activated C1s-Complement Inhibitor, in Chronic Inflammatory Demyelinating Polyneuropathy - A Phase 2, Multicentre, Open-Label Trial
Nazem Atassi, M.D.
Head of Early Development Neurology, Sanofi

2:35 p.m. The Clinical Perspective on Sustained Disease Control in Myasthenia Gravis
Johnson & Johnson

3:00 p.m. Understanding myasthenia gravis immune pathology: The role of B and T cells
Prof Raffaele Iorio, M.D., Ph.D.
Merck

3:25 p.m. A Focus on Muscle in SMA: Myostatin as a Therapeutic Target
Valeria Sansone, M.D., Ph.D.
Scholar Rock

3:50 p.m. Complementing the Future: Advances in Rare Disease Neurology
Emma Weiskopf, M.D.
Therapeutic Area Advisor, Senior Medical Director, Alexion Pharmaceuticals

4:15 p.m. argenx's quest to improve lives of patients with rare severe neuromuscular diseases through innovation and collaboration
Jeff Guptill, M.D.
argenx

4:40 p.m. Genetic medicine pipeline programs for rare genetic diseases of the muscle and central nervous system
Sarepta

6:00 p.m. 2025 NMSG Fellow | *Lalique*

Interrogating a role for endothelial cells in modulating pathophysiology and gene therapy response in human DMD using single nuclei transcriptomics
Sophie Rengarajan, M.D., Ph.D.
UCLA Medical Center

Robert C. Griggs Keynote Lecture: It takes a team: lessons in leadership and collaboration in Myotonic Dystrophy and beyond
Prof Valeria Sansone, M.D., Ph.D.
NeMO Clinic

EVENING ACTIVITIES

6:45 p.m. Leave for dinner | *Meet buses at the back entrance of Regina Palace Hotel*

8:00 p.m. Dinner and reception
La Tenuta del Golfo

Agenda

DAY 3: SUNDAY, SEPTEMBER 28

WELCOME
Lalique

8:00 a.m. Welcome
Richard J. Barohn, M.D. & Prof Michael Hanna

8:30 a.m. INSPIRE-IBM: An NIH-funded, two-year, multicenter, observational study in inclusion body myositis (IBM)- an update
Isela Hernandez, MS
University of California, Irvine

SESSION XI: NMSG YOUNG INVESTIGATOR PROJECTS
Moderators: Dr. Karen Suetterlin, MBBS, MRCP, Ph.D., & Michael Hehir, M.D.
Lalique

8:45 a.m. MAPP: MRI as a biomarker in Periodic Paralysis. A prospective longitudinal pilot study in periodic paralysis
Dr. Murva Asad
University College London

9:05 a.m. IgA autoantibodies demonstrate a novel mechanism of MuSK myasthenia gravis pathology
Gianvito Masi, M.D.
Yale University

SESSION XII: SHARK TANK SESSION
Lalique
Moderator: James Lilleker, MBChB, MRCP, Ph.D.
Sharks: Gordon Smith, M.D.
Nikoletta Nikolenko, M.D., Ph.D.
Karlein Mul, M.D., Ph.D.
Jasper Morrow, MBChB, Ph.D.

9:25 a.m. **Shark Tank Sessions**

Investigating Cancer-Triggered Autoimmunity in MuSK Myasthenia Gravis
Silvia Falso, M.D.
Catholic University of the Sacred Heart

Exploring Autoimmune Myasthenia Gravis in Facioscapulohumeral Muscular Dystrophy: A Pilot Study on a Novel Disease Association
Grace McMacken, MBBCh, Ph.D.
Queen's University Belfast

Gastrointestinal involvement in Myotonic Dystrophy type 1: characterization of a variable phenotype by PROMIS GI
Laura Tufano, M.D.
Sapienza University of Rome

Beyond Balance: Redefining Cerebellar Function in Myotonic Dystrophy,
Carola Rita Ferrari Aggradi, M.D.
NeMO Milan

11:00 a.m. Refreshments/Exhibitor Break | *Gallė*

Agenda

SESSION XIII: CAUTION IN THE INTERPRETATION AND CONCLUSIONS ON SOME TRIALS

Moderators: Karen Suetterlin, MBBS, MRCP, Ph.D., & Michael Hehir, M.D.
Lalique

11:15 a.m. Unresolved issues with efgartigimod in CIDP, and broader concerns for future trial designs
Jonathan S. Katz, M.D.
Sutter Health

11:35 a.m. Considerations for Interpretation of ALS ASO Trial Results- lessons learned from SOD1 and C9orf72 trials
Suma Babu, MBBS, MPH
Mass General Hospital, Harvard Medical School

11:55 a.m. Translarna and Beyond
Prof Laurent Servais, M.D., Ph.D.
University of Oxford

MEETING CLOSING

Lalique

12:15 p.m. Shark Tank Winner Announcement

12:20 p.m. Lunch | *Liberty Lago*

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SEPTEMBER 25-27, 2026

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