

MUSCLE STUDY GROUP

Annual Scientific Meeting

OCTOBER 1-3, 2021



Muscle Study Group

WELCOME!

Welcome! On behalf of your Muscle Study Group, we would like to welcome each of you to the 2021 Muscle Study Group Annual Meeting. It is an exciting time in neuromuscular research as we continue to grow and adapt.

We once again are conducting our meeting virtually given the recent surge of COVID over the summer. We were also shown by our meeting last year a ZOOM conference is an effective way to communicate at a distance. Again, we have set up the meeting for three morning sessions, as all day sessions contribute to ZFS (ZOOM FATIGUE SYNDROME), and our European colleagues need to sleep at some point!

We are excited this year to have more industry involvement than we have ever had. The Shark Tank session was such a success last year we are bringing it back with the opportunity of a \$10K grant to the winner!

We also have been encouraging residents to register and have been successful. We want to encourage and stimulate residents to pursue NM Fellowships as this group is our successors to the next generation.

The MSG continues to fund a Neuromuscular Research 2-year fellowship program so at any one time we have a Fellow in the first year and one in the second year. Both our current Fellows will be presenting during the meeting.

As the Co-Chairs of the Muscle Study Group, we would like to thank this year's planning committee for putting together an excellent agenda that covers such a broad range of topics and interests within the neuromuscular field. They have spent much time planning an in-person meeting and then changing course to secure a virtual meeting. Keep a lookout on the MSG website for the 2022 meeting information as next year we are hoping to meet in person in northern Italy.

Best wishes,



Richard J. Barohn, M.D.
*Executive Vice Chancellor for Health Affairs,
University of Missouri
Co-chair, Muscle Study Group*



Prof Michael G. Hanna, M.D.
*Director, University College London Institute of Neurology
Co-chair, Muscle Study Group*

2021 Planning Committee

Chafic Karam, M.D. // Chair
University of Pennsylvania Health System

James Lilleker, MBChB, Ph.D. // Co-Chair
The University of Manchester

Senda Ajroud-Driss, M.D.
Northwestern University

Richard Barohn, M.D. // MSG Chair
University of Missouri

Laurie Gutmann, M.D.
Indiana University

Michael Hanna, M.D. // MSG Co-Chair
University College London

Melissa McIntyre, DPT
University of Utah

Colin Quinn, M.D.
University of Pennsylvania Health System

Tracey Willis, MBChB, MMedSci, M.D.
Chester University, UK

Amelia Wilson, DPT
University of Utah Health

AGENDA //

Friday, October 1

James Lilleker, MBChB, Ph.D.; Senda Ajroud-Driss, M.D. // Moderators

8:00 - 8:05 A.M. OPENING Richard J. Barohn, M.D., Prof Michael G. Hanna, M.D.

8:05 - 9:05 A.M. **ROBERT C. GRIGGS, M.D. ANNUAL MSG LECTURE:**
IDIOPATHIC DOES NOT MEAN CRYPTOGENIC: THERAPEUTIC DEVELOPMENT
FOR METABOLIC NEUROPATHY
Gordon Smith, M.D. // Virginia Commonwealth University

9:10 - 9:25 A.M. **IBM INTERNATIONAL GENETICS CONSORTIUM STUDY UPDATE**
Prof Michael G. Hanna, M.D.

SESSION 1: INFLAMMATORY MYOPATHIES

9:25 - 9:45 A.M. **AUTOANTIBODIES IN MYOSITIS** Prof Neil McHugh, MBChB, M.D. // University of Bath

9:50 - 10:10 A.M. **INTERVENTIONAL STUDIES IN MYOSITIS (IVIG, TOC, RITUX)**
Rohit Aggarwal, M.D. // University of Pittsburgh

10:15 - 10:35 A.M. **RAPAMYCIN STUDY** Prof Olivier Benveniste, M.D., Ph.D. // AP-HP

10:37 - 10:50 A.M. **BREAK** Sponsored by CSL Behring

SESSION 2: FLASH PRESENTATIONS

10:50 - 11:00 A.M. **MOLECULAR BIOMARKERS IN MYOTONIC MUSCULAR DYSTROPHY TYPE 2**
Paloma Gonzalez-Perez, M.D., Ph.D. // MSG Fellow, Massachusetts General Hospital

11:02 - 11:12 A.M. **CD8 POSITRON EMISSION TOMOGRAPHY (PET/CT) IMAGING WITH 89ZR-DF-IAB22M2C**
IN PATIENTS WITH INCLUSION BODY MYOSITIS
Colin Quinn, M.D. // University of Pennsylvania

11:14 - 11:24 A.M. **10 PS MNEMONIC FOR DX OF IIM** Amir Sabouri, M.D. // Kaiser Permanente

11:26 - 11:36 A.M. **INFLAMMATORY MYOPATHIES PRESENTING WITH AXIAL WEAKNESS**
Elie Naddaf, M.D. // Mayo Clinic

11:38 - 11:58 A.M. **OPTIMIZING CIDP CARE WITH SCIG: PATH OLE AND BEYOND**
Mazen Dimachkie, M.D. // CSL Behring Update

12:03 - 12:23 P.M. **THE ROLES OF AUTOANTIBODIES IN IMMUNE CHECKPOINT INHIBITOR THERAPY:**
FROM BIOMARKERS TO MEDIATORS OF NEUROLOGICAL SYNDROMES
Karin Hoang Woodman, M.D. // Argenx Update

12:28 - 12:48 P.M. **FORCE™ PLATFORM DELIVERS EXON SKIPPING PMO, LEADS TO DURABLE INCREASES IN**
DYSTROPHIN PROTEIN IN MDX MICE AND IS WELL TOLERATED IN NHPS
Oxana Beskrovnaya, Ph.D. // Dyne Therapeutics Update

12:50 P.M. **CLOSING** Richard J. Barohn, M.D., Prof Michael G. Hanna, M.D.

12:50-1:30 P.M. **MINGLING TIME (OPTIONAL)**

Zoom Rooms: Cigar Mingling Room | CSL Behring Room
Dyne Therapeutics Room | Argenx Room

MUSCLE

STUDY GROUP

All times US Central Time

AGENDA //

Saturday, October 2

Chafic Karam, M.D.; Amelia Wilson, DPT // Moderators

8:00 - 8:08 A.M. OPENING Richard J. Barohn, M.D., Prof Michael G. Hanna, M.D.

8:09 - 8:19 A.M. ARIMOCLOMOL IN IBM TRIAL UPDATE
Mazen Dimachkie, M.D. // University of Kansas Medical Center

SESSION 3: INFLAMMATORY NEUROPATHIES AND OUTCOME MEASURES

8:20 - 8:40 A.M. EXPANDING THE SPECTRUM OF CHRONIC IMMUNE SENSORY POLYRADICULOPATHY:
THE CISP-PLUS SYNDROME James Dyck, M.D. // Mayo Clinic

8:45 - 9:05 A.M. BIOMARKERS IN CIDP/GBS Karissa Gable, M.D. // Duke University Medical Center

9:05 - 9:25 A.M. 2021 EAN/PNS CIDP GUIDELINES: A FOCUSED REVIEW OF THE NEW DIAGNOSTIC
AND TREATMENT GUIDELINES Jeff Allen, M.D. // University of Minnesota

9:30 - 9:50 A.M. OUTCOME MEASURES Gita Ramdharry, DPT // University College London

9:55 - 10:10 A.M. BREAK Sponsored by PTC Pharma

SESSION 4: FLASH PRESENTATIONS

10:10 - 10:25 A.M. POST-TRANSLATIONAL MODIFICATIONS OF DUX4
Renatta Knox, M.D. // MSG Fellow, Nationwide Children's Hospital

10:27 - 10:37 A.M. LOSS OF TDP-43 FUNCTION AND RIMMED VACUOLES PERSIST AFTER T CELL DEPLETION
IN A XENOGRAFT MODEL OF INCLUSION BODY MYOSITIS
Chiseko Ikenaga, M.D., Ph.D. // Johns Hopkins University

10:39 - 10:49 A.M. PROTOCOL FOR A HYBRID II STUDY EXPLORING THE FEASIBILITY OF DELIVERING,
EVALUATING, AND IMPLEMENTING A SELF-MANAGEMENT PROGRAMME FOR PEOPLE WITH
NEUROMUSCULAR DISEASES AT A SPECIALIST NEUROMUSCULAR CENTRE (ADAPT-NMD)
Louie Lee, BSc (HONS) // University College London

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10:51 - 11:01 A.M. ESTABLISHING CLINICAL TRIAL READINESS FOR VALOSIN CONTAINING PROTEIN-
ASSOCIATED MULTISYSTEM PROTEINOPATHY: BASELINE RESULTS FROM A 1-YEAR
PROSPECTIVE STUDY Megan A. Iammarino, PT, DPT // Nationwide Children's Hospital

11:03 - 11:13 A.M. COVID-19 RELATED OUTCOMES IN PRIMARY MITOCHONDRIAL DISEASES:
AN INTERNATIONAL STUDY Chiara Pizzamiglio, M.D. // University College London

11:15 - 11:25 A.M. SELF-MANAGEMENT IN NEUROMUSCULAR DISEASES: PRELIMINARY FINDINGS
FROM A QUALITATIVE EXPLORATION OF THE PATIENT PERSPECTIVE
Louie Lee, BSc (HONS) // University College London

11:27 - 11:37 A.M. PROSPECTIVE CLINICAL TRIAL READINESS IN LGMDR9 FKRP-RELATED MUSCULAR DYSTROPHY:
A GRASP CONSORTIUM STUDY Megan A. Iammarino, PT, DPT // Nationwide Children's Hospital

11:37 - 11:45 A.M. BREAK Sponsored by Argenx

11:45 A.M. - 12:05 P.M. DEFLAZACORT EVIDENCE FOR ITS ROLE IN SLOWING DMD DISEASE
Richard A. Able, Jr., Ph.D. // PTC Therapeutics Inc. Update

12:10 - 12:30 P.M. EXPLORE THE SCIENCE FOR AN APPROVED SPINAL MUSCULAR ATROPHY TREATMENT
Elizabeth Kichula, M.D., Ph.D. // Genentech Update

12:35 - 12:37 P.M. SAREPTA THERAPEUTICS SPONSOR INTRODUCTION Dr. Gilmore O'Neill

12:40 P.M. CLOSING Richard J. Barohn, M.D., Prof Michael G. Hanna, M.D.

12:40 - 1:30 P.M. MINGLING TIME (OPTIONAL)

Zoom Rooms: Cigar Mingling Room | Genentech Room
PTC Therapeutics Room

All times US Central Time

AGENDA //

Sunday, October 3

James Lilleker, MBChB, Ph.D.; Laurie Gutmann, M.D.; Melissa McIntyre, DPT // *Moderators*

MUSCLE STUDY GROUP

8:00 - 8:05 A.M. **OPENING** Richard J. Barohn, M.D., Prof Michael G. Hanna, M.D.

8:05 - 8:15 A.M. **NIH SIBM NATURAL HISTORY STUDY TRIAL UPDATE**
Tahseen Mozaffar, M.D. // *University of California, Irvine*

8:17 - 8:27 A.M. **DUCHENNE MUSCULAR DYSTROPHY RESPIRATORY PROFILES FROM REAL WORLD REGISTRY DATA**
Mona Hnaini, M.D. // *Pediatric Neuromuscular Fellow, London Health Science Centre, Western University*

SESSION 5: SHARK TANK

8:30 - 9:45 A.M. **SHARK TANK SESSIONS** James Lilleker, MBChB, Ph.D. // *Moderator*
W. David Arnold, M.D., Prof Tracey Willis, MBChB, MMedSci, M.D., Gita Ramdharry, DPT // *Shark Panel*

THE INNATE IMMUNE SYSTEM IN
MYASTHENIA GRAVIS
Katherine Dodd, MBChB MRCP,
Ph.D. Student // *University of Manchester, UK*

THE THERAPEUTIC PLAY GYM
PILOT STUDY
Jenna Linn Lammers, MSR/PT,CNT, PCS // *University of Florida*

IDENTIFICATION OF NOVEL
BIOMARKERS FOR INCLUSION BODY
MYOSITIS USING SINGLE-NUCLEI RNA
SEQUENCING OF MUSCLE BIOPSIES
Chiseko Ikenaga, M.D., Ph.D. // *Johns Hopkins University School of Medicine*

9:45 - 10:00 A.M. **BREAK** Sponsored by Genentech

SESSION 6: GENETICS

10:00 - 10:20 A.M. **VUS** Tanya Bardkjian, MS, LCGC // *University of Pennsylvania*

10:25 - 10:45 A.M. **RESOLUTION OF GENETIC VARIANTS OF UNCERTAIN SIGNIFICANCE USING MUSCLE BIOPSIES**
Karra Jones, M.D., Ph.D. // *University of Iowa*

10:50 - 11:10 A.M. **GENETIC TESTING IN NEUROMUSCULAR DISORDERS** Shawna Feely, MS, LGC // *University of Iowa*

11:10 - 11:15 A.M. **ANNOUNCEMENT OF SHARK TANK AWARD**

SESSION 7: FLASH PRESENTATIONS

11:15 - 11:25 A.M. **2020 SHARK TANK AWARD WINNER PRESENTATION:**
MEND (MEXILETINE VERSUS LAMOTRIGINE IN NON-DYSTROPHIC MYOTONIA)
Dr. Vinojini Vivekanandam, MBBS // *University College London Institute of Neurology*

11:27 - 11:37 A.M. **ADAPTING MRI AS A CLINICAL OUTCOME MEASURE FOR A FACIOSCAPULOHUMERAL
MUSCULAR DYSTROPHY TRIAL OF PREDNISONE AND TACROLIMUS**

Leo Wang, M.D., Ph.D. // *University of Washington*

11:38 - 11:48 A.M. **RACIAL DISPARITIES IN SKIN TONE REPRESENTATION OF DERMATOMYOSITIS RASHES**
Salman Bhai, M.D. // *University of Texas Southwestern*

11:49 - 11:59 A.M. **LONGITUDINAL DYSPHAGIA ASSESSMENT IN PATIENTS WITH CYSTINOSIS USING MBSIMP**
Stacey Sullivan, MS, CCC, SLP // *Mass General Hospital*

12:00 - 12:10 P.M. **CMT-COVID SURVEY** Riccardo Zuccarino, M.D. // *NEMO*

12:11 - 12:21 P.M. **CASE SERIES OF MYASTHENIA GRAVIS (MG) PATIENTS PRESCRIBED SUBCUTANEOUS
IMMUNOGLOBULIN (SCIG) THERAPY AND MONITORED BY PATIENT REPORTED OUTCOME
MEASURES (PROMS) BY A SPECIALTY INFUSION PHARMACY USING SOLEMETRICS**
Timothy Walton, MHS, CCRP // *Soleo Health*

12:22 - 12:32 P.M. **FLOW CYTOMETRY AND SORTING OF SINGLE ANTIBODY SECRETING CELLS FROM
FROZEN MUSCLE TISSUE** Vladimir Liarski, M.D. // *University of Chicago Medicine*

12:33 - 12:43 P.M. **EFFECT OF DISTAL HEREDITARY MOTOR NEUROPATHY ON MUSCLE STRUCTURE,
FUNCTION, AND GAIT PATTERNS: TWO CASE REPORTS**
Aljwhara Alangary, PT, Ph.D. Student // *University College London*

12:44 - 12:54 P.M. **MONOTHERAPY WITH ECULIZUMAB IN REFRACTORY ACETYLCHOLINE RECEPTOR
POSITIVE GENERALIZED MYASTHENIA GRAVIS**
Xiaoyang Li, MBBS, M.D. // *University of North Carolina at Chapel Hill*

12:55 P.M. **CLOSING** Richard J. Barohn, M.D., Prof Michael G. Hanna, M.D.

1:00 - 1:30 P.M. **AFTER HOURS OPEN MINGLING** (all welcome to attend)

All times US Central Time

MEETING SUPPORT //

Thank You!

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We are proud to sponsor the 2021 Muscle Study Group Annual Scientific Meeting.

Please join us on October 1, 2021 for an update on our efforts in Duchenne muscular dystrophy.

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gMG=generalized myasthenia gravis.

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PTC is a science-driven, global biopharmaceutical company focused on the discovery, development and commercialization of clinically differentiated medicines that provide benefits to patients with rare disorders. PTC's ability to globally commercialize products is the foundation that drives investment in a robust and diversified pipeline of transformative medicines and our mission to provide access to best-in-class treatments for patients who have an unmet medical need.

Please join us on Saturday October 2, 2021 at 11:45 am–12:05 pm CT to hear more about our latest updates at the 2021 Muscle Study Group Annual Scientific Meeting.

Following the presentation, we will be hosting a virtual 'Zoom room' at 12:30–1:30 pm CT. We welcome you to attend, meet the PTC team and ask us any questions you have about our work and our webinar.

To learn more about PTC, follow us on Facebook, on Twitter at @PTCBio, and on LinkedIn.

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OUR MISSION

Armed with the most advanced science in genetic medicine, we are in a daily race to rescue lives otherwise stolen by rare disease. At Sarepta, every day is another twenty-four hours to stand up for patients, advance technology, challenge convention and drag tomorrow into today.

KAI
Living with Duchenne muscular dystrophy

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| | | |
|----|--------------------------|-----------------------------------|
| S | Laundry | Arms tired again |
| M | | |
| T | Sam's 3rd Birthday Party | Cancelled: weak legs |
| W | Ballet pickup | COULDN'T DRIVE Vision problems |
| Th | Brunch with friends | Good day! |
| F | | |
| S | Big family dinner | Trouble chewing |

DISCOVER WHY UNCONTROLLED
gMG NEVER RESTS

Patient Portrayal

~50%
OF PATIENTS WITH GENERALIZED MYASTHENIA GRAVIS (gMG) REMAIN UNCONTROLLED¹

UP TO
70%
OF PATIENTS

FIND IT CHALLENGING TO DO
HOUSEHOLD CHORES^{2,3}

33%
OF PATIENTS

HAVE DIFFICULTY DRIVING
DUE TO ARM WEAKNESS AND
VISION PROBLEMS⁴

~40%
OF PATIENTS

HAVE DIFFICULTY CHEWING
AND SWALLOWING⁴

UNCONTROLLED. UNPREDICTABLE. UNEXPECTED.
Get the facts on uncontrolled disease at gMGFacts.com

REFERENCES: 1. Cutter G, et al. *Muscle Nerve*. 2019;60(6):707-715. 2. Nowak RJ. *Neurology Reviews*. March 2018;(Suppl):S1-S4.
3. The CE. *The cost to patients and the community of Myasthenia Gravis*. Centre for International Economics; November 2013.
4. Teck S, et al. *Health Qual Life Outcomes*. 2010;8:129.



2022 MSG MEETING //

September 30-October 2

Regina Palace, Stresa, Lake Maggiore, Italy

CONTINUING EDUCATION (CE)

Muscle Study Group
Annual Scientific Meeting 2021
Online // October 1-3, 2021

Physicians

In support of improving patient care, this activity has been planned and implemented by Amedco LLC and Muscle Study Group. 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.

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Our goals are to be the premier neuromuscular clinical and translational research organization and to create an environment to establish the next generation of researchers with your active involvement in the MSG we can accomplish these goals.



MSG Executive Committee

Richard J. Barohn, M.D. // Chair *University of Missouri*
Prof Michael Hanna, M.D. // Co-Chair *University College London*
Robert C. Griggs, M.D. // Chair Emeritus
University of Rochester Medical Center
William David, M.D., Ph.D. // Investigator Member
Massachusetts General Hospital
Michael Hehir, M.D. // Investigator Member
University of Vermont Medical Center
Valeria Sansone, M.D. // Investigator Member
NEMO Clinical Center
Melissa McIntyre, DPT // Evaluator Member *University of Utah*
Marie Wencel, CCRP // Study Coordinator Member
University of California, Irvine
Michael McDermott, Ph.D. // Biostatistician
University of Rochester Medical Center
Rabi Tawil, M.D. // Director, MSG Coordination Center
University of Rochester Medical Center
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