MUSCLE STUDY GROUP

Annual Scientific Meeting

SEPTEMBER 25-27, 2020

Muscle Study Group

WELCOME!

On behalf of your Muscle Study Group we would like to welcome each of you to the 2020 Muscle Study Group Annual Meeting. It is an exciting time in neuromuscular research as we continue to grow and adapt.

This year is different with our virtual meeting, it is an effective way for us all to communicate at a distance. We have set up this meeting for three morning sessions, as all day sessions could contribute to ZFS (ZOOM FATIGUE SYNDROME), also our European colleges need to sleep at some point!

Our plan COVID permitting, is to meet again in person in September of 2021 in Georgetown, DC. If this online meeting is successful, we might consider adding a virtual meeting to the yearly lineup.

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.

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.

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, UCL Institute of Neurology
Co-chair, Muscle Study Group

2020 Planning Committee

Carolina Barnett-Tapia, M.D., Ph.D. // Chair
University of Toronto

Chafic Karam, M.D.
Oregon Health and Science Hospital

Gita Ramdharry, Ph.D.
Queen Square MRC Centre for Neuromuscular Disease

James B. Lilleker, MBChB, Ph.D.
The University of Manchester

Kimberly A. Hart, M.A
University of Rochester Medical Center

Lindsay Alfano, DPT
Nationwide Children's Hospital

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

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

AGENDA //
Friday, September 25

Carolina Barnett-Tapia, M.D., Ph.D. & Chafic Karam, M.D. // *Moderators*

8-8:15 A.M. — **WELCOME** Richard J. Barohn, M.D., Prof Michael G. Hanna, M.D.

8:20-8:30 A.M. — **UPDATE ON THE PHASE 2/3 STUDY OF ARIMOCLOMOL IN IBM** *
Mazen Dimachkie, M.D. // *University of Kansas Medical Center*

8:32-8:42 A.M. — **MSG INTERNATIONAL INCLUSION BODY MYOSITIS (IBM) GENETICS CONSORTIUM UPDATE** * Alaa Khan, Ph.D. // *UCL Queen's Square*

8:45-8:55 A.M. — **INFLUENCE OF NT5c1A ANTIBODIES ON DISEASE PROGRESSION, CLINICAL PHENOTYPE AND BLOOD AND MUSCLE BIOMARKERS IN SPORADIC INCLUSION BODY MYOSITIS: A PROSPECTIVE EVALUATION** * Tahseen Mozaffar, M.D. // *University of California, Irvine*

9:00-9:20 A.M. — **DEVELOPMENT WORK FOR AN APP-BASED INTERVENTION TO PROMOTE PHYSICAL ACTIVITY IN PEOPLE LIVING WITH AND BEYOND CANCER**
Dr. Abi Fisher // *Associate Professor, UCL School of Behaviour Change*

9:25-9:55 A.M. — **CHALLENGES OF HEALTH ECONOMIC ASSESSMENT FOR SPINRAZA AND ZOLGENSMA**
Rick Chapman, Ph.D. // *Institute for Clinical & Economic Review*

10-10:20 A.M. — **INCORPORATING IMPLEMENTATION SCIENCE QUESTIONS INTO CLINICAL EFFECTIVENESS TRIALS**
Geoff Curran, Ph.D. // *Professor, University of Arkansas for Medical Sciences*

10:25-10:50 A.M. — **THE HEALEY ALS PLATFORM TRIAL: INNOVATIVE TRIAL DESIGN AND COLLABORATION TO ACCELERATE DRUG DEVELOPMENT**
Sabrina Paganoni, M.D., Ph.D. // *Healey Center for ALS, Massachusetts General Hospital*

10:55-11:10 A.M. — **BREAK**

11:10-11:25 A.M. — **PROXIMAL NERVE IMAGING IN CMT1A**
Reza Sadjadi, M.D. // *MSG Fellow, Massachusetts General Hospital*

10:27-11:37 A.M. — **LONG-TERM SAFETY AND EFFICACY OF GOLODIRSEN IN MALE PATIENTS WITH DUCHENNE MUSCULAR DYSTROPHY AMENABLE TO EXON 53 SKIPPING**
Francesco Mutoni, M.D. // *Chair of Paediatric Neurology, UCL GOS Institute of Child Health*

11:39-11:49 A.M. — **SATISFACTION WITH ANKLE FOOT ORTHOSES IN INDIVIDUALS WITH CHARCOT-MARIE-TOOTH**
Riccardo Zuccarino, M.D., PMR // *Fondazione Serena Onlus, Centro Clinico Nemo*

11:50 A.M.-12 P.M. — **EXPLORING MUSCLE STRUCTURE, FUNCTION AND GAIT PATTERNS IN PEOPLE WITH DISTAL HEREDITARY MOTOR NEUROPATHY: NATURAL HISTORY AND THE EFFECT OF REHABILITATION INTERVENTIONS, STUDY PROTOCOL**
Aljwhara Alangary, PT // *Ph.D. Student, UCL institute of Neurology, UK*

12-12:20 P.M. — **EXPANDING POSSIBILITIES IN THE TREATMENT OF SPINAL MUSCULAR ATROPHY**
Perry Shieh, M.D., Ph.D. // *Representative, Genentech*

12:25-12:45 P.M. — **SAREPTA PIPELINE OVERVIEW AND THE EVOLUTION OF REMOTE ASSESSMENT AND STRATEGIES FOR IMPLEMENTATION IN THE COVID-19 ERA AND BEYOND**
Linda P. Lowes, PT, Ph.D. // *Associate Professor, The Ohio State University of College of Medicine*
Louise Rodino-Klapac, Ph.D. // *Senior Vice President, Gene Therapy, Sarepta Therapeutics, Inc.*

12:45 P.M. — **CLOSING** Richard J. Barohn, M.D., Prof Michael G. Hanna, M.D.

12:45-1:30 P.M. — **FLASH PRESENTER AND SPONSOR NETWORKING**
ZOOM BREAKOUT ROOMS (OPTIONAL)

**Not for CME Credit*

All times US Central Time

MUSCLE STUDY GROUP

AGENDA //
Saturday, September 26

Lindsay Alfano, DPT & James B. Lilleker, MBChB, Ph.D. // *Moderators*

8-8:10 A.M. OPENING Richard J. Barohn, M.D., Prof Michael G. Hanna, M.D.

8:10-9:05 A.M. ROBERT C. GRIGGS, M.D. ANNUAL MSG LECTURE: HOW THE FSHD PUZZLE WAS SOLVED
Rabi Tawil, M.D. // *Fields Endowed Professor of Neurology, University of Rochester Medical Center*

9:07-9:27 A.M. RARE DISEASES: WHY CONSIDER ECONOMICS? Karen Lee // *CADTH*

9:30-9:50 A.M. A NOVEL APPROACH TO OPTIMIZING MOVEMENT IN TREATED CHILDREN WITH SPINAL MUSCULAR ATROPHY Megan Iammarino, PT, DPT // *Nationwide Children's Hospital*

9:55-10:10 A.M. BREAK

10:10-10:25 A.M. MOLECULAR BIOMARKERS IN MYOTONIC DYSTROPHY TYPE 2
Paloma Gonzalez Perez, M.D., Ph.D. // *MSG Fellow, Massachusetts General Hospital*

10:27-10:37 A.M. RESULTS FROM A NATIONAL CROSS-SECTIONAL STUDY OF DISEASE-BURDEN IN AMYOTROPHIC LATERAL SCLEROSIS (ALS): RESULTS FROM A NATIONAL CROSS-SECTIONAL STUDY Jennifer Weinstein, MS // *University of Rochester*

10:39-10:49 A.M. EXOME SEQUENCING IDENTIFIES NOVEL CANDIDATE GENES AND PHENOTYPIC EXPANSION IN A NEUROMUSCULAR COHORT
Daniel Calame, M.D., Ph.D. // *Baylor Medical College*

10:51-11:01 A.M. A SAFETY STUDY OF WEEKLY STEROIDS IN MUSCULAR DYSTROPHY (WSIMD)
Senda Ajroud-Driss, M.D. // *Associate Professor of Neurology, Northwestern University Feinberg School of Medicine*

11:03-11:13 A.M. AAV GENE THERAPY FOR TNNT1-ASSOCIATED NEMALINE MYOPATHY
Eleonora D'Ambrosio, M.D. // *University of Massachusetts*

11:15-11:25 A.M. FOLLOW-UP CARE IN MYASTHENIA GRAVIS DURING COVID-19: COMPARISON OF TELEMEDICINE AND IN-PERSON ENCOUNTERS
Constantine Farmakidis, M.D. // *Assistant Professor, University of Kansas Medical Center*

11:28-11:38 A.M. POST-COVID GUILLAIN-BARRE SYNDROME MIMICKING MYOSITIS
Sai Si Thu, M.D. // *SUNY Downstate Medical Center, USA*

11:38-11:45 A.M. BREAK

11:45 A.M.-12:05 P.M. TOTALITY OF EVIDENCE: CONTROLLING DYSTROPHIN AS AN ANTIGEN IN DUCHENNE MUSCULAR DYSTROPHY (DMD)
Brian E. Pfister, Ph.D., MBA // *Executive Director, US Medical Head-Neurology, PTC Therapeutics*

12:10-12:30 P.M. THE FUTURE OF MYASTHENIA GRAVIS TREATMENT, SPECIFICALLY THINKING ABOUT WHEN AND WHERE COMPLEMENT AND FCRN INHIBITORS MIGHT BE USED MOST EFFECTIVELY BASED ON AVAILABLE DATA James Howard, M.D. // *Representative, UCB*

12:30 P.M. CLOSING Richard J. Barohn, M.D., Prof Michael G. Hanna, M.D.

12:30-1:30 P.M. FLASH PRESENTER AND SPONSOR NETWORKING
ZOOM BREAKOUT ROOMS (OPTIONAL)

MUSCLE STUDY GROUP

All times US Central Time

AGENDA //
Sunday, September 27

Kimberly A. Hart, M.A & Gita Ramdharry, Ph.D. // *Moderators*

8-8:15 A.M. OPENING Richard J. Barohn, M.D. Prof Michael G. Hanna, M.D.

8:20-9:45 A.M. SHARK TANK SESSION ($10K GRANT AWARDED TO BEST PRESENTATION)
Vera Bril, BSc, M.D., FRCPC, Laurie Gutmann, M.D., James Lilleker, MBChB, Ph.D., William David, M.D., Ph.D. // *Sharks*
Will Meurer, M.D. // *Moderator*

- HEAD TO HEAD Dr. Vino Vivekanandam // *UCL Institute of Neurology*
- CLASH OF THE TITINS Jennifer Roggenbuck, MS, LGC // *The Ohio State University College of Medicine*
- PROJECT NMD MUSE: INSPIRING A DEEPER UNDERSTANDING OF MOTOR UNIT BEHAVIOR IN NEUROMUSCULAR DISEASE Kristina M. Kelly, PT, DPT, EdM, NCS, CPT, PES // *The Ohio State University*
- EXPLORING CSF BIOMARKERS IN PREPARATION FOR CLINICAL TRIALS TARGETING CNS IN DM1 Carola Rita Ferrari-Aggradi // *Medical Student, University of Milan*

9:45-10 A.M. BREAK

10-10:10 A.M. RESPIRATORY FUNCTION AND THE ROLE OF NON-INVASIVE VENTILATION IN MYOTONIC DYSTROPHY TYPE 1: A RETROSPECTIVE STUDY
Carola Rita Ferrari-Aggradi // *Medical Student, University of Milan*

10:13-10:23 A.M. MAGNETIC RESONANCE IMAGING (MRI) IN PERIODIC PARALYSIS
Dr. Vinojini Vivekanandam // *UCL Institute of Neurology*

10:25-10:35 A.M. INCIDENCE AND RISK FACTORS FOR PATELLOFEMORAL DISLOCATION IN ADULTS WITH CHARCOT-MARIE-TOOTH DISEASE: AN OBSERVATIONAL STUDY
Enza Leone, PT, MSc // *UCL Great Ormond Street Institute of Child Health*

10:35-10:45 A.M. ANNOUNCEMENT OF SHARK TANK AWARD

10:45-10:55 A.M. REFRACTORY CIDP: CHARACTERISTICS, ANTIBODIES AND RESPONSE TO ALTERNATIVE TREATMENT
Jamila Godil // *Medical Student, Oregon Health and Science University*

10:57-11:07 A.M. TIMED MOTOR FUNCTION TESTS IN BOYS WITH NONSENSE DMD MUTATIONS
Darina Dinov, DO // *PGY-2 Child Neurology Resident, Virginia Commonwealth University*

11:09-11:19 A.M. PATIENT ACCEPTABLE SYMPTOM STATES (PASS) IN CHRONIC INFLAMMATORY DEMYELINATING POLYNEUROPATHY (CIDP) Meg Mendoza, Ph.D. // *Research Analyst I, UHN*

11:21-11:31 A.M. SCREENING FOR GENETIC MUTATIONS IN PATIENTS WITH NEUROPATHY WITHOUT DEFINITE ETIOLOGY IS USEFUL Braden Vogt // *Medical Student, Brown University*

11:33-11:43 A.M. OPTIMIZING HAND-FUNCTION PATIENT OUTCOME MEASURES FOR INCLUSION BODY MYOSITIS
Ava Lin, M.D., Ph.D. // *Clinical Assistant Professor, University of Michigan*

11:45-11:55 A.M. FURTHER INSIGHT INTO DYSPHAGIA USING MBS-IMP IN ADULT PATIENTS WITH NEPHROPATHIC CYSTINOSIS AND MYOPATHY
Stacey Sullivan MS, CCC-SLP // *Massachusetts General Hospital*

11:57 A.M.-12:07 P.M. LIVE CELL-BASED ASSAY FOR ANTIBODIES TO CLUSTERED ACETYLCHOLINE RECEPTOR IN MYASTHENIA GRAVIS, CROSS VALIDATION, INTER-ASSAY STABILITY AND UTILITY IN A PAEDIATRIC COHORT SUSPECTED FOR MG Hans Frykman, M.D., Ph.D., FRCPC // *Clinical Assistant Professor, The University of British Columbia*

12:09-12:19 P.M. DIAGNOSTIC OUTCOME OF GENETIC TESTING ON NEUROMUSCULAR DISORDERS IN A TERTIARY CENTER Husam Al Sultani, M.D. // *Nerve and Muscle Center of Texas*

12:20-12:30 P.M. PATIENT PREFERENCE IN VIRTUAL VERSUS IN-PERSON VISITS IN NEUROMUSCULAR CLINICAL PRACTICE Komal Naeem // *Neurology Resident (3rd Year), Baylor College of Medicine*

12:30 P.M. CLOSING Richard J. Barohn, M.D., Prof Michael G. Hanna, M.D.

12:35-1:35 P.M. FLASH PRESENTER AND SPONSOR NETWORKING ZOOM BREAKOUT ROOMS (OPTIONAL)

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MEETING SUPPORT //
Thank You!

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NOW APPROVED

to treat spinal muscular atrophy in infants, children, and adults

In patients 2 months and older

Learn more at www.Evrysdi-HCP.com/NowApproved

Indication

Evrysdi is indicated for the treatment of spinal muscular atrophy (SMA) in patients 2 months of age and older.

Important Safety Information

Interactions with Substrates of MATE Transporters

- Based on in vitro data, Evrysdi may increase plasma concentrations of drugs eliminated via MATE1 or MATE2-K, such as metformin
- Avoid coadministration of Evrysdi with MATE (multidrug and toxin extrusion) substrates. If coadministration cannot be avoided, monitor for drug-related toxicities and consider dosage reduction of the coadministered drug if needed

Important Safety Information (continued)

Pregnancy

- In animal studies, administration of Evrysdi during pregnancy or throughout pregnancy and lactation resulted in adverse effects on development
- Based on animal data, advise pregnant women of the potential risk to the fetus. Pregnancy testing is recommended for females of reproductive potential prior to initiating Evrysdi. Advise female patients of reproductive potential to use effective contraception during treatment with Evrysdi and for at least 1 month after the last dose

Breastfeeding

- There is no data on the presence of Evrysdi in human milk, the effects on the breastfed infant, or the effects on milk production. The developmental and health benefits of breastfeeding should be considered along with the mother's clinical need for Evrysdi and any potential adverse effects on the breastfed infant

Potential Effects on Male Fertility

- Male fertility may be compromised by treatment with Evrysdi. Counsel male patients on the potential effects on fertility. Male patients may consider sperm preservation prior to treatment

Hepatic Impairment

- The safety and efficacy of Evrysdi in patients with hepatic impairment have not been studied
- Because Evrysdi is predominantly metabolized in the liver, hepatic impairment may potentially increase the exposures to Evrysdi. Avoid use of Evrysdi in patients with impaired hepatic function

Most Common Adverse Reactions

- The most common adverse reactions in later-onset SMA (incidence in at least 10% of patients treated with Evrysdi and more frequent than control) were fever, diarrhea, and rash
- The most common adverse reactions in infantile-onset SMA were similar to those observed in later-onset SMA patients. Additionally, adverse reactions with an incidence of at least 10% were upper respiratory tract infection, pneumonia, constipation, and vomiting

You may report side effects to the FDA at **1-800-FDA-1088** or **www.fda.gov/medwatch**. You may also report side effects to Genentech at **1-888-835-2555**.

Please see full Prescribing Information for additional Important Safety Information.

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PTC THERAPEUTICS

At PTC Therapeutics, it is our mission to provide access to best-in-class treatments for patients who have an unmet need.

PTC is a science-led, 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 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 September 26, 2020 at 11:45 am–12:05 pm CT to hear more about our latest updates at the 2020 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.

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

MED-US-DMD-2000147 | September 2020

Recognize the signs of Duchenne muscular dystrophy (DMD) for earlier diagnosis and intervention

Sarepta Therapeutics supports healthcare professionals to reduce missed or delayed diagnosis.

Perform the steps to identify early developmental delays that may lead to a diagnosis of DMD.[1]

LISTEN
to parents regarding concerns about their child's development

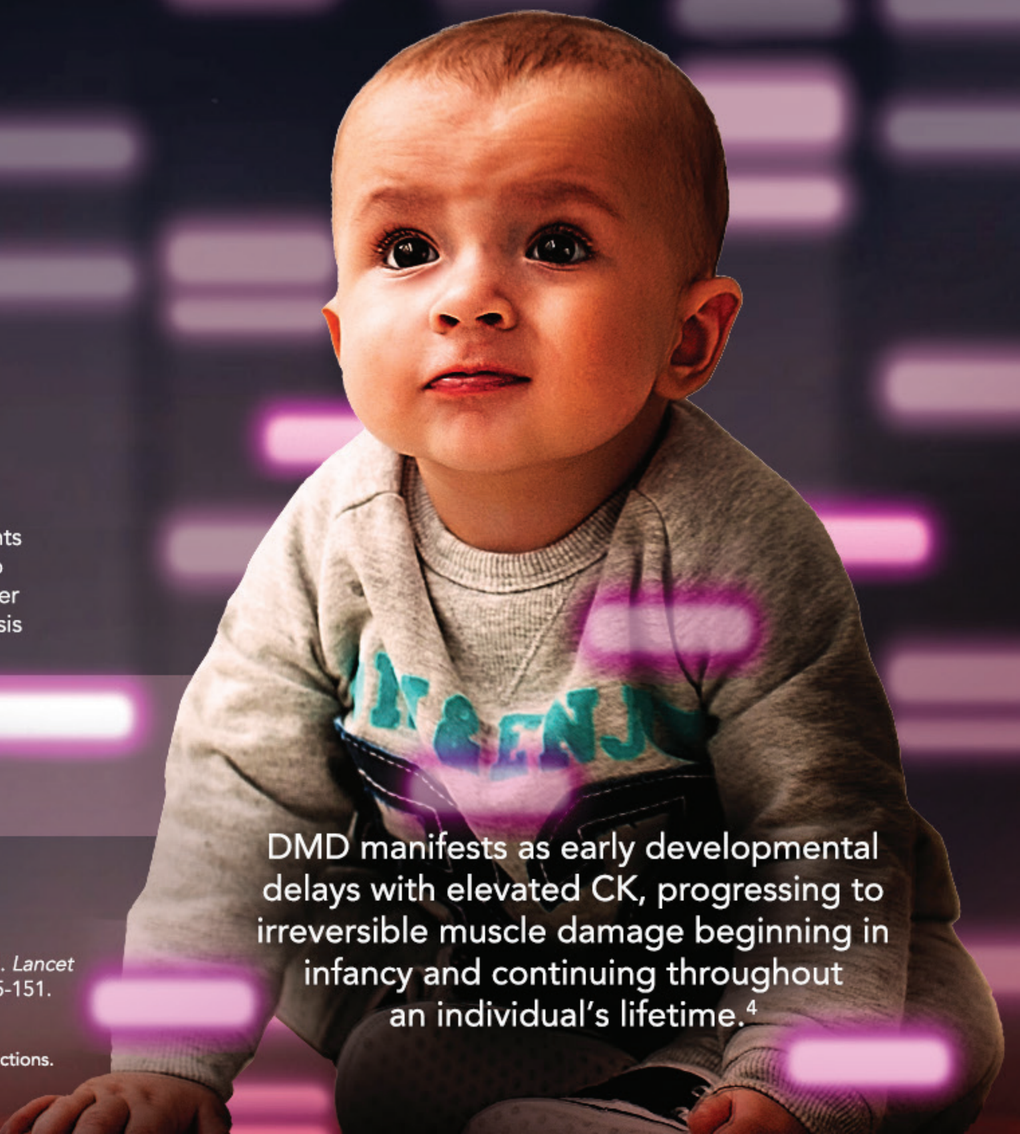
OBSERVE
signs of muscle weakness

EVALUATE
motor development to determine the cause of muscle weakness

TEST
for creatine kinase (CK) if a developmental delay is confirmed

REFER
elevated CK warrants prompt referral to neurology for further testing and diagnosis

Prompt and accurate diagnosis of DMD is a crucial aspect of care, allowing for improved patient management and timely genetic counseling.[2,3]



DMD manifests as early developmental delays with elevated CK, progressing to irreversible muscle damage beginning in infancy and continuing throughout an individual's lifetime.[4]

REFERENCES:

1. National Task Force for Early Identification of Childhood Neuromuscular Disorders. Surveillance and referral aid for primary care providers. https://childmuscleweakness.org/wp-content/uploads/2019/05/PrimaryCareProviderPacket.pdf. Accessed February 25, 2020. 2. Birnkrant DJ, et al. *Lancet Neurol.* 2018;17(3):251-267. 3. van Ruiten HJA, et al. *Arch Dis Child.* 2014;99(12):1074-1077. 4. Aartsma-Rus A, et al. *J Med Genet.* 2016;53(3):145-151.

2021 MSG MEETING // September 24-26

Georgetown University Hotel and Conference Center
Washington, DC

CONTINUING EDUCATION (CE)

Muscle Study Group
Annual Scientific Meeting 2020
Online // September 25 – 27, 2020

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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Prof Michael Hanna, M.D. // Co-Chair
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Mazen Dimachkie, M.D. //
Investigator Member, Treasurer
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University of Rochester Medical Center

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Melissa Mcintyre, DPT, DPT //
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