MDA CLINICAL & SCIENTIFIC CONFERENCE 2024

Conference Agenda

ALL CONFERENCE SESSION TIMES LISTED ARE IN EASTERN TIME

Saturday, March 2, 2024		
12:00 – 6:00 PM	Registration Open for Care Center Directors	Lobby Level "Meeting Planner Registration" (near Lake Rooms)
12:00 – 6:00 PM	Registration Open for Large Exhibit Booth Sponsors	Orlando I-III, VI
5:30 – 7:00 PM	MDA Care Center Network Happy Hour	Bar Tela

Sunday, March 3, 2024		
7:00 AM – 8:00 PM	Registration Open	Lobby Level "Meeting Planner Registration" (near Lake Rooms)
8:00 AM – 3:30 PM	MDA Care Center Directors Meeting (Invite Only)	Lake Eola
8:00 AM – 3:30 PM	MDA Care Center Allied Health Symposium (Invite Only)	Lake Mizell
1:00 – 5:00 PM	MDA Trainee Networking Session (Invite Only)	Champions Gate
2:00 – 5:00 PM	Neuromuscular Advocacy Collaborative Meeting (Invite Only)	Lake Nona
6:00 – 8:00 PM	Welcome Reception & Posters in Exhibit Hall	Orlando Ballroom

MDA CLINICAL & SCIENTIFIC CONFERENCE 2024

Conference Agenda

ALL CONFERENCE SESSION TIMES LISTED ARE IN EASTERN TIME

Monday, March 4, 2024		
6:00 AM – 6:00 PM	Registration Open	(see Sunday)
7:00 – 8:00 AM	**Industry Forum Breakfast presented by argenx** ***MDA CIDP Program Title:*** *Our Evolving Understanding of CIDP: Pathophysiology, Current Unmet Treatment Needs, and Exploring the Role of IgG Autoantibodies*	Florida 5-7
	Industry Forum Breakfast presented by The France Foundation *Evolutions in the Management of Duchenne Muscular Dystrophy: Treatment Implications for the Present and Future*	Florida 4
8:30 – 9:30 AM	**Opening Remarks** Donald S. Wood, PhD President and CEO of the Muscular Dystrophy Association **MDA Legacy Award** Jeffrey Chamberlain, PhD	Orange Ballroom
9:30 – 9:55 AM	Networking Break in Exhibit Hall	Orlando Ballroom
9:30 – 10:30 AM	Exhibit Hall & Poster Sessions Open	Orlando Ballroom
10:00 AM – 12:00 PM	**Keynote Address** Brooke Eby ALS Patient and Advocate **Panel Discussion on NMD in the Age of Therapy** Sharon Hesterlee, PhD (moderator) PJ Brooks, PhD Barry Byrne, MD, PhD Justin Moy Elizabeth McNally, MD, PhD This panel discussion brings together experts in the fields of precision medicine, patient advocacy, healthcare systems, and future perspectives to delve into crucial aspects of neuromuscular disease treatment. The discussion aims to foster a comprehensive understanding of current challenges, breakthroughs, and collaborative efforts shaping the landscape of neuromuscular healthcare. ***P values vs. People: How Can Patient Experience be Integrated Into Regulatory Decision-Making?*** Holly Peay, PhD	Orange Ballroom
12:00 – 1:30 PM	**Industry Forum Lunch presented by Biogen** *Latest Real-World Evidence in Adults with SMA: Learn More About the Pivotal Data and One Patient's Journey*	Orlando IV-V
	Industry Forum Lunch presented by Edgewise Therapeutics *Protecting and Preserving Dystrophic Muscle: The Balance Between Exercise and Contraction-Induced Muscle Injury*	Key West
	Industry Forum Lunch presented by Genentech *A Closer Look at Male Fertility and Bulbar Function in SMA*	Florida 5-7
	Industry Forum Lunch presented by Pfizer *Caring for the DMD Patient Using a Multidisciplinary Approach*	Key Largo
	Industry Forum Lunch presented by Sarepta *Putting Into Focus: Evaluating Functional Assessments in Duchenne Across Clinical Trials and Practice*	Florida 4

MDA CLINICAL & SCIENTIFIC CONFERENCE 2024

Conference Agenda

ALL CONFERENCE SESSION TIMES LISTED ARE IN EASTERN TIME

Monday, March 4, 2024 cont.		
12:00 – 1:30 PM	Exhibit Hall & Poster Sessions Open	Orlando Ballroom
	Clinical Considerations	
2:00 – 3:30 PM	**Rapid-Fire Clinical Case Reviews Part 1: Motor Neuron Disease & NMJ Disorders** ***(CE Accredited)*** John Day, MD, PhD (Co-Chair) Bakri Elsheikh, MBBS, FRCP, FAAN (Co-Chair) *SOD1-related Lower Motor Neuron Disease* Jennifer Roggenbuck, MS, CGC *Riboflavin Deficiency Presenting as Potential Myasthenic Syndrome* Chamindra Laverty, MD *Pediatric Amyotrophic Lateral Sclerosis (FUS-ALS)* Jacinda Sampson, MD *Intermittent Episodes of Fatigue and Weakness* Megan Waldrop, MD *Symptomatic 5q SMA in the Era of Newborn Screening* Megan Waldrop, MD *Normal Toddler to Progressively Weakening Teen* Matthew Wicklund, MD, FAAN	Orange E-G
	Lab to Life	
2:00 – 3:30 PM	**Translational research in ALS: Novel Biomarkers and Model Systems** Michael Ward, MD, PhD *The New Kids Around The Block: Contribution Of Glial Cells In ALS/FTD-Mediated Neurodegeneration* Rita Sattler, MSc, PhD (Chair) *Towards Early Prediction Of Amyotrophic Lateral Sclerosis Empowered by Machine Learning and Clinical Big Data* Askar Safipour Afshar, MS *Characterization Of Alternative Polyadenylation Events in ALS Highlights New Disease Mechanisms and Novel Gene Therapy Targets* Frederick (Eric) Arnold, PhD *Human 3D Cortico-Motor Assembloids To Model ALS* Jimena Andersen, PhD	Florida 1-3

Monday, March 4, 2024 cont.		
Clinical Approaches in Streamlining Care		
2:00 – 3:30 PM	Newborn Screening ***(CE Accredited)*** *The State of Newborn Screening in Neuromuscular Disease* Marcia Felker, MD (Chair) *Overview of Ethical Newborn Screening (USA focus)* Thomas Crawford, MD *A Consented Pilot Study in NYS To Screen Newborns For Duchenne Muscular Dystrophy* Norma Tavakoli, PhD *Newborn Screening in Pompe Disease* Nishitha Pillai, MD *Newborn Screening for Duchenne Muscular Dystrophy: Results from Early Check* Katerina Kucera, PhD	Orange D
Practical Consideration in Gene Therapy		
2:00 - 3:30 PM	Where Are We in Gene Therapy? ***(CE Accredited)*** *Potential Impact of Gene Therapy in Neuromuscular Disease* Diana Castro, MD *Known or Experienced Serious Adverse Events in Gene Therapy* Russell Butterfield, MD, PhD *Potential Short- & Long- Term Harms of Gene Therapy* Richard Finkel, MD *Weighing Benefits & Harms with People with NMD and Their Families* Perry Shieh, MD, PhD *Ethical and Family Implications of Gene Therapy* Holly Peay, PhD (Chair)	Orange A-C
3:30 – 4:30 PM	Exhibit Hall & Poster Sessions Open	Orlando Ballroom
3:30 – 3:55 PM	Networking Break	Orlando Ballroom

Monday, March 4, 2024 cont.

Clinical Considerations		
4:00 – 6:00 PM	**Rapid-Fire Clinical Case Reviews Part 2: Muscle Diseases** *(CE Accredited)* John Day, MD, PhD (Co-Chairs) Bakri Elsheikh, MBBS, FRCP, FAAN (Co-Chairs) *Pregnancy in Adult Myotonic Dystrophy Type 1* Dominic Fullenkamp, MD, PhD *Diagnosis Unknown: Congenital Myopathy? Congenital Fiber Type Disproportion?* Natalie Katz, MD, PhD *Diagnosis Unknown: Suspected Pediatric Dystrophinopathy* Natalie Katz, MD, PhD *Titinopathy Diagnosis After Decades-Long Odyssey* Jennifer Roggenbuck, MS, CGC *Adult- Onset Multiple Acyl-CoA Dehydrogenase Deficiency (MADD)* Jennifer Roggenbuck, MS, CGC *LGMD 2T/R19 Due to Heterozygous Pathogenic Variants in GMPPB (Myopathy Combined With NMJ Deficit)* Matthew Wicklund, MD, FAAN *Malignant Hyperthermia, Elevated CK, and RYR1 in a Carrier of Dystrophinopathy Gene* Matthew Wicklund, MD, FAAN	Orange E-G
Lab to Life		
4:00 – 5:00 PM	**ALS Genetic Discovery, Mechanisms, and Therapeutic Development** Matthew Harms, MD (Chair) *Rare Variant Analyses Validate Known ALS Genes in A Multi-Ethnic Population and Identifies ANTXR2 As A Candidate in PLS* Tess Pottinger, PhD *ALS: Translating Genetic Discoveries Into Therapies* Bryan Traynor, MD, PhD *Development of a UNC13A Cryptic Exon Skipping Antisense Oligonucleotide as a Treatment for ALS* Sam Alworth, MS, MBA *Investigation of ALS Caused by Mutant KIF5A* Devesh Pant, PhD	Florida 1-3

Monday, March 4, 2024 cont.		
5:00 – 6:00 PM	**Panel Session: Natural History Studies: Most Recent Collaborative Consortia Efforts** Amelie Gubitz, PhD Robert Bowser, PhD James Berry, MD, MPH Stephanie Fradette, PharmD	
Clinical Approaches in Streamlining Care		
4:00 – 6:00 PM	**Pediatric Neuromuscular Disease** ***(CE Accredited)*** Elena Caron, MD (Chair) *Primary Care Perspective Of How to Best Collaborate On Pediatric NM Patient Care From Rural to Urban Settings* Faria Abdullah, MD *Collaboration With Adult Specialists to Transition Care Of Pediatric Neuromuscular Patients to Adult Care* Amanda Witt, MD *Help Me, Help You: What An ER Physician Needs to Know About The NM Patient and How They Need to Know It* Brian Lindsey, DO *School Based Physical Therapy* Jessica Herron, PT, DPT	Orange D
Practical Consideration in Gene Therapy		
4:00 – 6:00 PM	**Is My Institution Equipped for Gene Replacement Therapies?** ***(CE Accredited)*** *Is My Institution Prepared for Gene Transfer Therapies?* Crystal Proud, MD (Chair) *Gene Transfer Therapy: Dosing Day Logistics* Omer Abdul-Hamid, MD *A Leadership Perspective* Aravindhan Veerapandiyan, MD *Clinical Care Consideration for Patient & Families: Prior to and Following Gene Transfer Therapy* Nancy Kuntz, MD	Orange A-C
6:00 – 8:00 PM	Poster & Networking Reception	

MDA CLINICAL & SCIENTIFIC CONFERENCE 2024

Conference Agenda

ALL CONFERENCE SESSION TIMES LISTED ARE IN EASTERN TIME

Tuesday, March 5, 2024		
6:00 AM – 6:00 PM	Registration Open	(See Sunday)
7:00 – 8:00 AM	**Industry Forum Breakfast presented by Biogen** *Genetic Counseling and Testing: Advancements, Barriers, and Opportunities in Rare Disease*	Florida 4
	Industry Forum Breakfast presented by Catalyst *A New Treatment Option for DMD: Integration Into Clinical Practice*	Florida 5-7
	Industry Forum Breakfast presented by ML Bio Solutions *Biomarkers in the Limb Girdle Muscular Dystrophies*	Key West
	Industry Forum Breakfast presented by UCB *Mitochondrial Myopathies: Mimicking Neuromuscular Diseases*	Key Largo
Clinical Considerations		
8:30 – 10:30 AM	**Challenges Facing the Clinician in the Era of Treatment** ***(CE Accredited)*** Richard Finkel, MD (Chair) *Types of Therapies: Relative Risks & Burden of Treatment* Anne Connolly, MD *FDA-Approved Therapies: Implications of Approval Type on Clinical Benefit* Perry Shieh, MD, PhD *Real-World Challenges* Katherine Mathews, MD *When is Enough, Enough?* Julie Parsons, MD	Orange E-G
Lab to Life		
8:30 – 10:30 AM	**Development of Gene Therapy for Rare and Ultra-Rare Neuromuscular Disease** Alan Beggs, PhD (Chair) Anna Buj-Bello, PhD *Canine Model Of X-Linked Myotubular Myopathy: Discovery to IND-Enabling Studies* Casey Childers, DO, PhD *AAV Gene Therapy, Liver Toxicity, and X-Linked Myotubular Myopathy* James Dowling, MD, PhD *Rapid Transition of AT132 to Clinical Testing by an Industry Sponsor* John Gray, PhD	Lake Eola A-B

MDA CLINICAL & SCIENTIFIC CONFERENCE 2024

Conference Agenda

ALL CONFERENCE SESSION TIMES LISTED ARE IN EASTERN TIME

Tuesday, March 5, 2024

Clinical Approaches to Streamlining Care		
8:30 – 10:30 AM	**Adult Neuromuscular Disease: Engaging Primary Care and Other Non-NMD Experts** ***(CE Accredited)*** *Adult NMD: Engaging Primary Care & Other Non-NMD Experts* Lora Clawson, MSN, CRNP (Chair) *Adult NMD: Engaging Primary Care and other "Non-NMD Experts* Becky Hopfinger, MSN, MBA, FNP-BC *Building a Neuromuscular Disease Care Community* Bruce Rabin, MD, PhD	Orange D
Practical Considerations in Gene Therapy		
8:30 – 10:30 AM	**My Institution is Not Equipped to Administer Gene Replacement Therapy: Now What?** Barry Byrne, MD, PhD (chair) Taylor Kaschak, BSN, RN	Orange A-C
Disease Mechanism & Therapeutic Strategies		
8:30 – 10:30 AM	**Role of Immune System and the Microenvironment in Neuromuscular Disorders** Aaron Beedle, PhD (Co-Chair) *Treg and Macrophage Interactions Regulate Muscle Fibrosis* Armando Villalta, PhD *Targeting the Innate Immune System to Improve Gene Therapy in Mdx Mice.* Melissa Morales, PhD *Targeting Inflammation to Improve Muscle Regeneration and Enhance Dystrophin Restorative Therapy* Jyoti Jaiswal, MSc, PhD (Co-Chair) *Spatiotemporal Macrophage Subtype Specification Guides The Formation Of Dynamic Multilayered Regenerative Inflammation Zones During Tissue Repair* Andreas Patsalos, MS, PhD *Where Immunology Meets Genetics: A Snapshot Of Immunophenotypes in LAMA2-CMD Pathophysiology and The Impact Of Genetic Therapy* Yonne Tenorio De Menezes, MS, PhD *The Super-Healing MRL Strain Promotes Muscle Growth in Muscular Dystrophy Through a Regenerative Extracellular Matrix* Alexis Demonbreun, PhD *Intercellular Communication During Muscle Regeneration and Disease* Daniel Kopinke, PhD	Florida 1-3

MDA CLINICAL & SCIENTIFIC CONFERENCE 2024

Conference Agenda

ALL CONFERENCE SESSION TIMES LISTED ARE IN EASTERN TIME

Tuesday, March 5, 2024 cont.		
10:30 AM – 1:30 PM	Exhibit Hall & Poster Sessions Open	Orlando Ballroom
10:30 – 10:55 AM	Networking Break sponsored by UCB	Orlando Ballroom
Clinical Considerations		
11:00 AM – 12:00 PM	**Setting Patients Up for Success: Life Transitions** ***(CE Accredited)*** Jamie Eskuri, MD (Chair) Emelia Rogers, LICSW Candice Johnson, OTD, ORT/L Kaitlin Lewis, CTRS	Orange E-G
Lab to Life		
11:00 AM – 12:00 PM	**Neuromuscular Junction Disease** *The Present Landscape of Myasthenia Gravis* Linda Kusner, PhD (Chair) *Recent Advances in the Diagnosis and Treatment of Congenital Disorders of the Neuromuscular Junction* Ricardo Maselli, MD *Pathophysiology of Lambert-Eaton Myasthenic Syndrome* Stephen Meriney, PhD *Digital Endpoints and Artificial Intelligence Application to Neuromuscular Disease* Henry Kaminski, MD	Lake Eola A-B
Clinical Approaches to Streamline Care		
11:00 AM – 12:00 PM	**Novel Strategies for Clinical Trial Recruitment & Participation** ***(CE Accredited)*** *A Path Forward* Gil Wolfe, MD, FAAN (Chair) *What Constitutes a Recruitment Plan?* Amy Bartlett, BA, CCRC *Inclusive & Intersectional Research Engagement Across the Lifespan* Helen Hemley, MPA	Orange D
Practical Considerations in Gene Therapy		
11:00 AM – 12:00 PM	**Gene Therapy in DMD: Is It Now or Never?** ***(CE Accredited)*** *Gene Therapy in DMD: Is It Now or Never?* Natalie Goedeker, DNP, CPNP (Chair) *The Future of Gene Therapy in Duchenne: Building on Current Milestones* Emma Ciafaloni, MD, FAAN	Orange A-C

Tuesday, March 5, 2024 cont.		
Disease Mechanism & Therapeutic Strategies		
11:00 AM – 12:00 PM	**Single-Cell and Spatial Transcriptomics to Understand Disease** Doug Millay, PhD (Chair) *Muscle Remodeling in DMD and in Response to Gene Therapy at Single Cell/Nuclei Resolution* Carrie Miceli, PhD *Transcriptome-based Insights Into Brain Manifestations in Dystrophinopathy* Mathula Thangarajh, MD, PhD *snRNAseq of Muscle Biopsies As a New Tool to Identify New Biomarkers and Response to Treatment in Patients With Muscle Disorders* Jordi Diaz Manera, PhD *Single-cell Dissection Of The Human Motor and Prefrontal Cortices in ALS and FTLD* Veronique Belzil, MS, PhD	Florida 1-3
12:00 – 1:30 PM	**Industry Forum Lunch presented by argenx** *Consideration of the Treatment Journey in gMG: Perspectives on the Use of VYVGART and VYVGART HYTRULO in Adults With gMG Who Are AChR-Ab Positive*	Orlando IV-V
	Industry Forum Lunch presented by Biogen *The Future of Friedreich Ataxia: Exploring Advances in Treatment*	Florida 4
	Industry Forum Lunch presented by The CME Institute *Advancements in Understanding Facioscapulohumeral Muscular Dystrophy (FSHD) and What Clinically Meaningful Outcomes Look Like*	Key Largo
	Industry Forum Lunch presented by Sanofi *It's Time to Go: Transition of Care in Neuromuscular Conditions*	Key West
	Industry Forum Lunch presented by Scholar Rock *Muscle Matters in SMA: Experiences of a Person Living with SMA, a Neurologist, and a Physical Therapist*	Florida 5-7

Tuesday, March 5, 2024 cont.

Lab to Life		
2:00 PM – 4:00 PM	**Dystrophies** Katherine Mathews, MD (Chair) *The Impact Of Long Read Sequencing on Diagnostics of Neuromuscular Diseases* Stephan Züchner, MD, PhD *Long Read Sequencing For Comprehensive Genetic Analysis In FSHD* Russell Butterfield, MD, PhD *Post-Translational Modifications of the DUX4 Protein Impact Toxic Function in FSHD Cell Models* Renatta Knox, MD, PhD *Synaptic Defects in Mouse Models of Dystroglycanopathy* Kevin Wright, PhD *Steroids in Limb Girdle Muscular Dystrophy* Elizabeth McNally, MD, PhD *The Use of Ribitol For Treating FKRP-Related Dystroglycanopathy* Anthony Blaeser, PhD *What Lies Ahead For Myotonic Dystrophy* Charles Thornton, MD	Lake Eola A-B
Clinical Approaches to Streamline Care		
2:00 PM – 4:00 PM	**Underserved Communities & Working with What We've Got: Overcoming Challenges in the Healthcare System** ***(CE Accredited)*** *What are Medically Underserved Communities?* Meeta Cardon, MD (Co-Chair) *Reaching Out to Underserved Communities in NM* Leslie Morrison, MD *Inclusive & Intersectional Research Engagement Across the Lifespan* Anna Kostera-Pruszczyk, MD, PhD *Healthcare considerations in treating indigenous communities through the Native American experience in the Southwest* Monika Krzesniak-Swinarska, MD (Co-Chair)	Orange D

Tuesday, March 5, 2024 cont.

Practical Considerations in Gene Therapy		
2:00 PM – 4:00 PM	**Day-of: Orchestrating the Team & Planning for the Unexpected & We Did It - Now What? Navigating post GTT Monitoring in "Real Life"** *(CE Accredited)* *DMD Gene Therapy Day Of: Institutional and Patient/Family Readiness- When We Were Not a Trial Site* Leigh Ramos-Platt, MD *Orchestrating the Team & Planning for the Unexpected* Diana Bharucha-Goebel, MD *Post Gene Therapy Delivery Issues* Erika Finanger, MD *MDA Gene Therapy Support Network* Nora Capocci, MPH *Q&A & Wrap Up* Julie Parsons, MD (Chair)	Orange A-C
Disease Mechanism & Therapeutic Strategies		
2:00 – 4:00 PM	**Multi-Disease and Next Generation Genetic Therapies** Dwi Kemaladewi, PhD (Co-Chair) *Novel Protein Therapeutics Targeting Sarcolemma Membrane Repair* Noah Weisleder, PhD (Co-Chair) *Next-Generation Genome Editing* Thomas Gaj, PhD *Leveraging Skeletal Muscle Fusion Properties For Skeletal Muscle Gene Therapy* Sajedah Hindi, PhD *Therapeutic Targeting of a Pathological MicroRNA in Becker Muscular Dystrophy and Duchenne Muscular Dystrophy* Anders Naar, PhD *AUF1 Gene Therapy For Traumatic Skeletal Muscle Injury: Significantly Accelerated Muscle Regeneration in Preclinical Models* Dounia Abbadi, PhD *Directed Evolution of Novel MyoAAV Capsid Variants Enabling Effective Systemic Muscle Transduction While De-Targeting The Liver in Non-Human Primates* Sharif Tabebordbar, PhD *Expression of Large and Highly Functional Dystrophins Using Split Inteins and Myotropic AAV Vectors* Hichem Tasfaout, PharmD, PhD *Myospreader propagates gene therapies throughout syncytial myofibers* Eric Wang, PhD	Florida 1-3

Tuesday, March 5, 2024 cont.		
3:30 – 4:30 PM	Exhibit Hall & Poster Sessions Open	Orlando Ballroom
4:00 – 4:25 PM	Networking Break sponsored by UCB	Orlando Ballroom
	Clinical Considerations	
4:30 – 6:00 PM	**Overview of Therapeutics for FSHD** In partnership with TREAT-NMD ***(CE Accredited)*** Jeffrey Statland, MD (Chair) *FSHD: Presentation, Progression and Disease Mechanism* Rabi Tawil, MD *Exercise and FSHD* Natalie Katz, MD, PhD *Emerging Therapies for FSHD* Enrico Bugiardini, PhD	Orange E-G
	Lab to Life	
4:30 – 6:00 PM	**CMT (Peripheral Nerve Disease)** Michael Shy, MD (Chair) *Towards Clinical Trial Readiness for CMT1A* David Herrmann, MBBCh Stephan Züchner, MD, PhD *Aggregation of Lamin Protein in Aging and Disease* Alysia Vrailas-Mortimer, PhD *Pathogenic Variation in R-Loop Helicase Gene DHX9 Causes Axonal Neuropathy and Neurodevelopmental Disorders* Daniel Calame, Md, PhD	Lake Eola A-B
	Clinical Approaches to Streamline Care	
4:30 – 6:00 PM	**Multidisciplinary Care: Meeting the Needs of the Patient TODAY** ***(CE Accredited)*** *A Multidisciplinary Approach for Adult Neuromuscular Patients Possibility vs. Reality* Amanda Peltier, MD (Chair) *Multidisciplinary Care Model: Meeting the Needs of Pediatric Patients* Urvi Desai, MBBS, MD, FAAN	Orange D

MDA CLINICAL & SCIENTIFIC CONFERENCE 2024

Conference Agenda

ALL CONFERENCE SESSION TIMES LISTED ARE IN EASTERN TIME

Tuesday, March 5, 2024 cont.		
Practical Considerations in Gene Therapy		
4:30 – 6:00 PM	Navigating Difficult Conversations *(CE Accredited)* *Navigating Difficult Conversations: Ineligible patients* Kaitlin Batley, MD *Navigating Difficult Conversations: Financial Coverage and Resources Restraints* Aravindhan Veerapandiyan, MD *Navigating Difficult Conversations: An Adult Perspective* Deidre Devier, PhD (Chair)	Orange A-C
Disease Mechanism & Therapeutic Strategies		
4:30 – 6:00 PM	Dysfunction in Cellular Organelles and Trafficking in Neuromuscular Disorders Carlos Moraes, PhD (Co-Chair) *Principles of Gene Regulation in a Large, Dense Syncytium* Eric Wang, PhD *The Mitochondrial Protein CHCHD10 Has Different Roles in Health and Neuromuscular Diseases* Giovanni Manfredi, MD, PhD *Mechanism of Protein Trafficking and Quality Control In Skeletal Muscle* Vandana Gupta, PhD, MSc (Co-Chair) *Altered Mitochondria Calcium Homeostasis In MICU1KO Is Associated With Synaptic Dysfunction in Mice Cortical Neurons* Raghavendra Singh, PhD *Multivesicular Bodies and Their Exosomes Link Poor Myofiber Repair With Muscle Degeneration in Muscular Dystrophy* Daniel Christopher Bittel, PhD	Florida 1-3
6:00 – 8:00 PM	Poster & Networking Reception	

MDA CLINICAL & SCIENTIFIC CONFERENCE 2024

Conference Agenda

ALL CONFERENCE SESSION TIMES LISTED ARE IN EASTERN TIME

Wednesday, March 6, 2024		
7:00 AM – 1:00 PM	Registration Open	(see Sunday)
8:30 AM – 1:00 PM	**New, Novel, and Noteworthy: NMD Highlights** We are thrilled to have had an overwhelming number of incredible abstract submissions as we reconvene in 2024. To that end, we have redesigned our Clinical Trial Presentations to include a wider range of topics which also allows for even more live presentations selected directly from the abstract submissions. This is a final day not to be missed!	
8:30 AM – 1:00 PM	**Clinical Trial Updates**	**Florida 1-3**
8:30 – 8:45 AM	*The Go-Digital Study: Activity Monitoring and Fine Motor Skills Assessment in People With ALS* Sharon Tamin	
8:45 – 9:00 AM	*Interim Analysis of the Radicava/Edaravone Findings in Biomarkers from ALS (REFINE-ALS) Study* James Berry, MD	
9:00 – 9:15 AM	*Interim Results From the RESPOND Study of Nusinersen in Children With Spinal Muscular Atrophy (SMA) Previously Treated With Onasemnogene Abeparvovec* Crystal Proud, MD	
9:15 – 9:30 AM	*Long-Term Safety and Efficacy of Subcutaneous Efgartigimod PH20 in Participants With Generalized Myasthenia Gravis: Interim Results of ADAPT-SC+ Study* Tuan Vu, MD	
9:30 – 9:45 AM	*Two-year Safety and Exploratory Efficacy of AT845 Gene Replacement Therapy for Late Onset Pompe Disease: FORTIS, a Phase 1/2 Open-label Clinical Study* Tahseen Mozaffar, MD	
9:45 – 10:00 AM	*Switching Treatment to Cipaglucosidase alfa+miglustat Positively Affects Motor Function and Quality of Life in Patients With Late-Onset Pompe Disease* Barry Byrne, MD, PhD	
10:00 – 10:15 AM	*Safety and Efficacy of Vatiquinone Treatment in Friedreich Ataxia Patients from MOVE-FA: a Phase 3, Double-blind, Placebo-controlled Trial* David Lynch, MD, PhD	
10:15 – 10:30 AM	*Initial Data from the ACHIEVE Trial of DYNE-101 in Adults with Myotonic Dystrophy Type 1 (DM1)* Valeria Sansone, PhD	
10:30 – 10:45 AM	Break	
10:45 – 11:00 AM	*Safety and Efficacy of Delandistrogene Moxeparvovec Versus Placebo in Duchenne Muscular Dystrophy (EMBARK): Pivotal Phase 3 Primary results* Jerry Mendell, MD	
11:00 – 11:15 AM	*Longitudinal Multicentric Study to Validate SV95C in Ambulant DMD aged 2-15 Years Old* Laurent Servais, MD	
11:15 – 11:30 AM	*Functional and Muscle Damage Biomarker Changes Following Treatment with EDG-5506, a Fast Myosin Modulator, in Adults with Becker Muscular Dystrophy* Han Phan, Md	
11:30 – 11:45 AM	*Initial Data from the DELIVER Trial of DYNE-251 in Males with DMD Mutations Amenable to Exon 51 Skipping* Kevin Flanigan, MD	

MDA CLINICAL & SCIENTIFIC CONFERENCE 2024

Conference Agenda

ALL CONFERENCE SESSION TIMES LISTED ARE IN EASTERN TIME

Wednesday, March 6, 2024 cont.		
11:45 AM – 12:00 PM	*Long Term Safety and Efficacy of CAP-1002 in late-stage patients with DMD: A New Treatment Approach to Target Skeletal and Cardiac Muscle Pathogenesis* Craig McDonald, MD	
12:00 – 12:15 PM	*RGX-202, an Investigational Gene Therapy for the Treatment of Duchenne Muscular Dystrophy: Interim Clinical Data* Aravindhan Veerapandiyan, MD	
12:15 – 12:30 PM	*Pamrevlumab Failed to Meet Primary and Secondary Endpoints for nonambulatory Patients With Duchenne Muscular Dystrophy (DMD) in LELANTOS-1* Eugenio Mercuri, MD, pHd	
12:30 – 12:45 PM	*SAT-3247: An Oral Small Molecule Inhibitor Targeting AAK1, a Critical Effector Of Skeletal Muscle Regeneration* Phil Lambert, PhD	
12:45 – 1:00 PM	*Fordadistrogene movaparvovec for Duchenne muscular dystrophy: 3-year functional outcomes and changes in thigh and upper-limb muscle volume* Perry Shieh, MD, PhD	
8:45 AM – 12:00 PM	**Pathogenic Variants and Novel Disease Links**	**Florida 5-7**
8:45 - 9:00 AM	*A Variant in CCDC78 Collaborates with a Mutation in LMNA to Cause LGMD and Core Myopathy in a Large Family* Lori Wallrath, PhD	
9:00 – 9:15 AM	*Impaired Iron-Sulfur Cluster Assembly Due to Biallelic Variants in CIAO1 Leads to a Novel Neuromuscular Disease* Rotem Orbach, MD	
9:15 – 9:30 AM	*Dominant Cardioskeletal Titinopathies Reflect Distinct Mechanisms of Disease* Jennifer Roggenbuck, MS CGC	
9:30 – 9:45 AM	*RNAseq Analysis in a Cohort of Undiagnosed Congenital Myopathy Patients* Pamela Barraza-Flores, PhD	
9:45 – 10:00 AM	*A novel COL11A1 Variant in a Child With Neuromuscular Findings: Expanding the Genotypic and Phenotypic Spectrum of COL11A1-related Disease* Meghan McAnally, MD	
10:00 – 10:15 AM	*DNA Sequence Variation in SMAD7 Enhances LMNA-associated Skeletal Muscle Disease Severity and OImplicates SMAD Signaling as a Therapeutic Target* Nathan Mohar, PhD	
10:15 – 10:30 AM	BREAK	
10:30 – 10:45 AM	*Modulating the RNA Exosome Complex Influences C9orf72 ALS/FTD Pathology* Sebastian Michels, MD	
10:45 – 11:00 AM	*Blocking Translation to Rescue ALS/FTD Phenotypes Associated with C9ORF72 Repeat Expansion* Xin Jiang, PhD	
11:00 – 11:15 AM	*PTPσ-Mediated PI3P Regulation as a Therapeutic Strategy for C9ORF72-ALS/FTD* Zhe Zhang, PhD	
11:15 – 11:30 AM	*Regeneration of Neuromuscular Synapses by Inhibition of the Prostaglandin Degrading Enzyme 15-PGDH* Elena Monti, PhD	
11:30 – 11:45 AM	*Molecular Insights into Neuromuscular Decline in Spinal and Bulbar Muscular Atrophy* Anastasia Gromova, PhD	
11:45 AM – 12:00 PM	*Cerebrospinal Fluid Proteomic Changes after Nusinersen in Patients with Spinal Muscular Atrophy* Marie Beaudin, MD, MS	

Wednesday, March 6, 2024 cont.		
8:45 AM – 12:00 PM	Preclinical Studies in Dystrophies and Myopathies	Key West
8:45 – 9:00 AM	*AGAMREE (Vamorolone) Improves Becker Muscular Dystrophy and Increases Dystrophin Protein in Novel Bmx Model Mice* Christopher Heier, PhD	
9:00 – 9:15 AM	*A Small Noncoding tRNA Half Exerts Disease-Modifying Bioactivity by Modulating Macrophage Function in Duchenne Muscular Dystrophy* Russel Rogers, PhD	
9:15 – 9:30 AM	*Comparisons of Microdystrophin Gene Therapy for Duchenne Muscular Dystrophy* Asuka Eguchi, PhD	
9:30 – 9:45 AM	*Muscle-specific JAG1 Overexpression Increases Skeletal Muscle Mass in a Mouse Model of Duchenne Muscular Dystrophy* Felipe de Souza Leite, PhD	
9:45 – 10:00 AM	*Duchenne Muscular Dystrophy-Related Cardiomyopathy: Beyond Dystrophin Deficiency in Skeletal Muscle* Luca Caputo, PhD	
10:00 – 10:15 AM	*Developing Novel Epigenetic Treatments For DMD Using Human Stem Cell-Derived Engineered Muscle Tissues* Philip Barrett, PhD	
10:15 – 10:30 AM	*New Insight Into The Mechanism Of Eccentric Contraction-Induced Force Loss In Skeletal Muscle of the Mdx Mouse Model Of Duchenne Muscular Dystrophy* James Ervasti, PhD	
10:30 – 10:45 AM	BREAK	
10:45 – 11:00 AM	*Analysis of Extracellular Vesicles Reveals Disordered Renal Metabolism In Myotonic Dystrophy Type 1* Preeti Kumari, PhD	
11:00 – 11:15 AM	*Correction of Myotonia And Reduction Of Toxic DMPK Foci After Treatment of the Hsalr Mouse Model of Myotonic Dystrophy Type 1 Using An Adeno-Associated Virus Delivering U7snrna Interfering With CTG Repeat Expansion* Nicholas Wein, PhD	
11:15 – 11:30 AM	*Clinical and Translational Studies In In-Vitro And An In-Vivo Mouse Model Of A Unique HSPB8 Associated Vacuolar Myopathy* Lan Weiss, MD, PhD	
11:30 – 11:45 AM	*A Single Cell Atlas Mapping Disease Progression in Dysferlinopathy Offers New Therapeutic Insights* Marshall Hogarth, PhD	
11:45 AM – 12:00 PM	*Discovery Driven Development of Circulating Biomarkers for FSHD* Joel Chamberlain, PhD	