MDA CLINICAL & SCIENTIFIC CONFERENCE 2023

Conference Agenda

ALL CONFERENCE SESSION TIMES LISTED ARE IN CENTRAL TIME
SPEAKERS ARE IN ORDER OF SPEAKING POSITION

Saturday, March 18, 2023		
12:00 – 6:00 PM	Registration Open for Care Center Directors & Large Exhibit Booth Sponsors	Geppetto Foyer, Tower Side

Sunday, March 19, 2023		
7:00 AM – 8:00 PM	Registration Open	Geppetto Foyer, Tower Side
7:30 AM – 3:30 PM	Care Center Directors Meeting (Invite Only)	Stemmons Ballroom
1:00 – 5:00 PM	MDA Trainee Networking Session (Invite Only)	Carpenters Ballroom
2:00 – 5:00 PM	Neuromuscular Advocacy Collaborative Meeting (Invite Only)	Metropolitan
6:00 – 8:00 PM	Welcome Reception in Exhibit Hall	Exhibit Hall

MDA CLINICAL & SCIENTIFIC CONFERENCE 2023

Conference Agenda

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Monday, March 20, 2023		
6:00 AM – 6:00 PM	Registration Open	Geppetto Foyer, Tower Side
7:00 – 8:00 AM	Industry Forum Breakfast presented by Biogen *Treating Patients with SPINRAZA® (nusinersen) Exploring the Role of Regular Monitoring to Identify Motor Function Changes Over Time*	Chantilly West
	Industry Forum Breakfast presented by PTC *Integrating Corticosteroids into all-around Duchenne Muscular Dystrophy Care*	Chantilly East
	Industry Forum Breakfast presented by Reata *A Newly Approved Treatment for Friedreich's Ataxia*	Grand DE
8:30 – 9:30 AM	Opening and Keynote Address Donald S. Wood, PhD President & CEO, MDA Peter Marks, MD, PhD Director of the Center for Biologics Evaluation and Research (CBER) US Food and Drug Administration (FDA)	Trinity Ballroom
9:30 – 9:55 AM	Networking Break	Exhibit Hall
10:00 – 10:30 AM	2023 MDA Legacy Award for Achievement in Clinical Research Awarded to Merit Cudkowicz, MD, MSc *Presented By:* Stanley Appel, MD	Trinity Ballroom
10:30 – 10:50 AM	MDA Funded Research – Fueling the Therapies of Tomorrow Sharon Hesterlee, PhD	
10:50-11:05 AM	Gene Therapy Summit Update Barry Byrne, MD, PhD	
11:05-11:20 AM	Public Policy Priorities of the Neuromuscular Disease Community Paul Melmeyer	
11:20-11:25 AM	Introducing MDA's 2023 Development Grantees Angela Lek, PhD	
11:25-11:45 AM	Care & the Community at MDA Nora Capocci Alicia Dobosz	
9:30 AM – 1:30 PM	Exhibit Hall and Poster Sessions Open	Exhibit Hall
12:00 – 1:30 PM	Industry Forum Lunch Presented by Genentech *Expert Discussion: The Evolving Management of the SMA Patient and the Role of Evrysdi*	Chantilly West
	Industry Forum Lunch Presented by Pfizer *Management of Duchenne Muscular Dystrophy (DMD) – How Far Have We Come?*	Grand DE
	Industry Forum Lunch Presented by AKH *Moving Forward in Pompe Disease: Optimizing Outcomes in the Era of Next-Generation Therapies and Advanced Newborn Screening* Industry Forum Lunch is jointly provided by AKH and Catalyst Medical Education, LLC. This activity is supported by an educational grant from Sanofi.	Grand ABC
	Industry Forum Lunch Presented by Sarepta *Assessing the Assessments for Duchene- An Expert Panel Discussion*	Chantilly East

MDA CLINICAL & SCIENTIFIC CONFERENCE 2023

Conference Agenda

ALL CONFERENCE SESSION TIMES LISTED ARE IN CENTRAL TIME
SPEAKERS ARE IN ORDER OF SPEAKING POSITION

Monday, March 20, 2023 cont.

Ultra-Rare Track	Coronado A
2:00 – 2:30 PM	**Myopathies/CMD** Alan Beggs, PhD (Chair) The Expanding Clinical Phenotype of PYROXD1 Myopathy Frances Evesson, PhD Recessive and Dominant Mutations in DNAJB4 Cause Novel Chaperonopathies Michio Inoue, MD, PhD
2:30 – 3:00 PM	**LGMD** LGMD: Emerging Solutions Peter Kang, MD (Chair) Preliminary Results from MLB-01-003: An Open Label Phase 2 Study of BBP-418 in Patients with Limb-girdle Muscular Dystrophy Type 2I Douglas Sproule, MD, MSc iPSC-Based Cell Therapy for Limb-Girdle Muscular Dystrophy Type 2A/R1 Peter Andersen, PhD
3:00 – 3:30 PM	**CMT** Then and Now: General Landscape of Genetic Discoveries and Clinical Trials in CMT Michael Shy, MD (Chair) Advancing Clinical Trial Readiness for TRPV4 Neuromuscular Disease Brett McCray, MD, PhD
Technology Track	**Coronado BCD**
2:00 – 3:30 PM	**Translation of Gene Editing Technologies** Melissa Spencer, PhD (Chair) Immunological Barriers to CRISPR Editing Therapy for Duchenne Muscular Dystrophy Dongsheng Duan, PhD Upregulation of LAMA1 as a Mutation-Independent therapeutic Approach for LAMA2-CMD Dwi Kemaladewi, PhD Preclinical development of a gene editing therapy targeting a large hotspot of DMD patients Courtney Young, PhD Afrooz Rashnonejad, MSc, PhD Functional Rescue of Periodic Paralysis by in Vivo Gene Editing Stephen Cannon, MD, PhD CRISPR Correction of Cardiomyopathy Eric Olson, PhD

MDA CLINICAL & SCIENTIFIC CONFERENCE 2023

Conference Agenda

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Monday, March 20, 2023 cont.		
DMD Beyond Muscle: A Holistic Approach Track (CE Accredited Session)		Cortez AB
2:00 – 2:45 PM	Making Visible the Invisible: Cognitive and Behavioral Health in Muscular Dystrophy Conceptualizing Neurocognitive & Behavioral Functioning in DBMD Natalie Truba, PhD V Preethish Kumar, MBBS, MRCP, MRCPE, PhD Mathula Thangarajh, MD (Chair) Pragmatic Solutions: Cognition-Focused Interventions Alisha Pollastri, PhD	
2:45 – 3:30 PM	Bone Health - Where Have We Been, Where Are We Now, and Where Are We Going? Where Have We Been? Meilan Rutter, MD (Chair) Where We are Now: Clinical Approach David Weber, MD, MSCE Where are We Going? Nat Nasomyont, MD, MS	
NMD Care Delivery: Care Throughout the Patient Journey Track (CE & Genetic Counselor CEUs Accredited Session)		Cortez CD
2:00 - 3:30 PM	Diagnostic Testing Options Katherine Mathews, MD (Co-Chair) The Role of Biopsy Pathology in the Era of Neuromuscular Disease Genetic Testing Steven Moore, MD, PhD Diagnostic Imaging in the Era of Genetic Testing Diana Bharucha-Goebel, MD Detection and Implication of Intronic Mutations and Other Novel Genetic Variants in Dystrophinopathies Kevin Flanigan, MD (Co-Chair)	
3:30 – 4:30 PM	Exhibit Hall and Poster Sessions Open	Exhibit Hall
3:30 – 3:55 PM	Networking Snack Break sponsored by Entrada Therapeutics	Exhibit Hall

Monday, March 20, 2023 cont.

Ultra-Rare Track	Coronado A
4:00 – 5:00 PM	**Therapies for Primary Mitochondrial Myopathies** Introduction to Clinical Trials for Primary Mitochondrial Myopathies Michio Hirano (Chair) Mavodelpar (REN001) for Primary Mitochondrial Myopathies Will Chou, MD Development of Elamipretide for the Treatment of Primary Mitochondrial Myopathy Anthony Abbruscato, PharmD, BCPS TK2 Deficiency Susan VanMeter, MD
5:00 – 6:00 PM	**MDA Kickstart Program for Ultra-Rare Gene Therapy Development** Ultra-Rare Medical Products: Integrated Development, Access and Use Perspectives and Case Studies Review Maryna Kolochavina, PharmD, PhD, PMP A New Program from MDA to Tackling Ultra-Rare Neuromuscular Disease Sharon Hesterlee, PhD (Chair) AAV9-Mediated Gene Therapy of Choline Acetyltransferase Deficient Mice Ricardo Maseli, MD Panelists: Alan Beggs, PhD; Michio Hirano, MD, PhD; Peter Kang, MD & Michael Shy, MD, PhD

Technology Track	Coronado BCD
4:00 – 5:00 PM	**Non-Viral Delivery Approaches for Genetic Therapies** Stanley Froehner (Chair) Effect of Myonuclear Domain Structure on the Efficacy of DMD Therapeutics Thomas Roberts, PhD Targeted Non-Viral Delivery of the Full-Length Dystrophin Gene to Dystrophic Muscle Nick Whitehead, PhD Endosomal Escape Vehicles (EEV™) to Enhance the Functional Delivery of Oligonucleotides in Muscular Dystrophy Mahasweta Girgenrath, PhD

MDA CLINICAL & SCIENTIFIC CONFERENCE 2023

Conference Agenda

ALL CONFERENCE SESSION TIMES LISTED ARE IN CENTRAL TIME
SPEAKERS ARE IN ORDER OF SPEAKING POSITION

Monday, March 20, 2023 cont.		
Technology Track		Coronado BCD
5:00 – 6:00 PM	Using iPS Cells to Model Neuromuscular Disease Using patient-specific iPS cells to model and target FKRP mutations Rita Perlingeiro (Chair) Advanced In Vitro Modelling of Neuromuscular Diseases and Therapeutics Francesco Saverio Tedesco, PhD Uncovering Strategies to Bolster Sarcomere and Sarcolemma Stability with DMD Microphysiological Systems Penney Gilbert, PhD Using 3D Engineered Cardiac and Skeletal Muscle Tissues for Disease Modeling and Drug Discovery David Mack, PhD	
DMD Beyond Muscle: A Holistic Approach Track (CE Accredited Session)		Cortez AB
4:00 – 5:30 PM	Digestive Health Impacts from Top to Bottom David Brumbaugh, MD, MSCS, FAAP (chair) Deglutition - DMD Beyond Muscle: Digestive Health Impacts from Top to Bottom Katlyn McGrattan, PhD Ajay Kaul, MD The Distinct Nutritional Phases and Challenges of Nutritional Assessment in Patients with Duchenne Muscular Dystrophy Laura Watne, MS, RD, CSP	
NMD Care Delivery: Care Throughout the Patient Journey Track (CE & Genetic Counselor CEUs Accredited Session)		Cortez CD
4:00 – 5:30 PM	Supporting the Patient Throughout Their Journey Ericka P. Greene, MD, MACM, FAAN (Chair) Rebecca Axline, LCSW-S, CSM, APHSW-C Keelie Denson, MD Timothy Lotze, MD	
6:00 – 8:00 PM	Poster & Networking Reception	Exhibit Hall

MDA CLINICAL & SCIENTIFIC CONFERENCE 2023

Conference Agenda

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SPEAKERS ARE IN ORDER OF SPEAKING POSITION

Tuesday, March 21, 2023		
6:00 AM – 6:00 PM	Registration Open	Geppetto Foyer, Tower Side
7:00 – 8:00 AM	Industry Forum Breakfast presented by Amylyx *Incorporating newly Approved RELYVRIO™ Into Treatment Approach for Adults in Living with ALS: A Case Study*	Chantilly West
	Industry Forum Breakfast presented by Sanofi *The Next Step Forward: Real World Experience in Patients with Late-Onset Pompe Disease*	Grand DE
	Industry Forum Breakfast presented by Sarepta *Exploring the Treatment Galaxy: AAV-based Gene Transfer Research for Patients Living with Neuromuscular Diseases*	Chantilly East
9:00 – 11:45 AM	Insights in Research Investor Summit (agenda link)	Wedgwood Ballroom
	ALS Track	Coronado A
8:30 – 10:30 AM	ALS Collaborations: A Continuum from Research to Care Melanie Leitner, PhD (Chair) Target ALS Approach to Accelerating ALS Research and Drug Development Manish Raisinghani, MBBS, PhD Multimodal Collaboration in ALS: Sharing Skills and Data Fernando Viera, MD Clare Durrett Coordinated Philanthropy for Transforming ALS Kuldip Dave, PhD Sharon Hesterlee, PhD Darleen Sawicki, MSN, NP-BC	
	Digital Outcomes & Big Data Track	Coronado BCD
8:30 – 10:30 AM	Remote Outcomes and Decentralized Studies Jeffrey Statland, MD (Chair) Digital and Home-based Outcome Measures in ALS Jeremy Shefner, MD, PhD Stakeholder Informed; Remote Video Assessments, Quantifying What is Meaningful to Patients and Caregivers Christine McSherry, RN BSN The First-Ever Regulatory Qualification of a Real-World Digital Outcome Measure and Its Transformative Impact on Drug Development in Duchenne Muscular Dystrophy Paul Strijbos, PhD Multi-Modal Wearable Sensors and Digital Health Measures for Neurological Diseases Ashkan Vaziri, PhD Digital Home Monitoring for Patients with Neuromuscular Diseases Amir Lahav, ScD	

MDA CLINICAL & SCIENTIFIC CONFERENCE 2023

Conference Agenda

ALL CONFERENCE SESSION TIMES LISTED ARE IN CENTRAL TIME
SPEAKERS ARE IN ORDER OF SPEAKING POSITION

Tuesday, March 21, 2023 cont.		
Practical Considerations in Gene Therapy Track (Genetic Counselor CEUs Accredited Session)		Cortez AB
8:30 – 10:30 AM	**Updates on Gene Therapy in NMD: Current & Emerging Therapies** Natalie Goedeker, MSN, CPNP (Chair) **Review: Current and Emerging Gene Therapies in DMDs** Emma Ciafaloni, MD, FAAN **Review: Current and Emerging Gene Therapies in SMA** Richard Finkel, MD **Future of Gene Therapy in NMD: Pressing Questions** Alex Murphy, BBM, CRT **Safety Issues: Gene Therapy Administration and Management** Kevin Flanigan, MD	
NMD Care Delivery: Care Throughout the Patient Journey Track (CE Accredited Session)		Cortez CD
8:30 – 9:30 AM	**Registry Engagement Successes** **MOVR Registry Engagement Success** Daragh Heitzman, MD, FAAN (Chair) **United States National ALS Registry: A Multi-Faceted Research Platform** Paul Mehta, MD **Hurdles of Registry from Research Coordinator Perspective** Rupa Nallamothu **Data Registries** R. Bradley Troxler, MD	
9:30 – 10:30 AM	**Leveraging Providers in the Community** Craig McDonald, MD (Chair) **Leveraging Providers in the Community: Neurodevelopmental and Psychosocial Care** Molly Colvin, PhD **Leveraging Providers in the Community: Rehabilitation Therapy Services** Tina Duong, PT, PhD **Leveraging Providers in the Community: Respiratory Therapy and Sleep Medicine** Tony Mozzone, CRT **Leveraging Providers in the Community: Technology-enabled Care** Erik Henricson, PhD, MPH	
10:30 AM – 1:30 PM	Exhibit Hall and Poster Sessions Open	Exhibit Hall
10:30 – 10:55 AM	**Networking Snack Break sponsored by UCB** **Networking Refreshment Break sponsored by Reata**	Exhibit Hall

MDA CLINICAL & SCIENTIFIC CONFERENCE 2023

Conference Agenda

ALL CONFERENCE SESSION TIMES LISTED ARE IN CENTRAL TIME
SPEAKERS ARE IN ORDER OF SPEAKING POSITION

Tuesday, March 21, 2023 cont.

ALS Track		Coronado A
11:00 AM – 12:00 PM	**Advances in the Understanding in ALS** Stathmin-2: an Emerging Therapeutic Target in TDP-43 Proteinopathies Clotilde Lagier-Tourenne, MD, PhD A Combined Stem Cell and Gene Therapy Approach to Treat ALS Clive Svendsen, PhD Novel Treg-Enhancing Therapy Suppresses Neuroinflammation in ALS Stanley Appel, MD (Chair)	
Digital Outcomes & Big Data Track		Coronado BCD
11:00 AM – 12:00 PM	**Large-Scale Data Approaches to NMD Research** New Trends in Genetic Studies of Rare Diseases Stephan Zuchner, PhD Utilizing Data From the *All of Us* Research Program Anastasia Wise, PhD Using Large Genomic Data Sets to Identify Regions of Constraint in the Mitochondrial Genome Monkol Lek, PhD The Clinical Genome Resource: Advancing Genomic Knowledge through Global Curation Jonathan Berg, MD, PhD	
Practical Considerations in Gene Therapy Track (CE & Genetic Counselor CEUs Accredited Session)		Cortez AB
11:00 AM – 12:00 PM	**Future of Gene Therapy in NMD: Considerations for Newborn Screening & Clinical Trial Design** Natalie Goedeker, MSN, CPNP (Chair) NBS in NMD: What Have We Learned and Where Are We Going? Don Bailey, PhD Future of Clinical Trial Design in DMD and SMA Katherine Mathews, MD	
NMD Care Delivery: Care Throughout the Patient Journey Track (CE Accredited Session)		Cortez CD
11:00 AM – 12:00 PM	**Holistic Approach to Transitioning into Adulthood** Jamie Twanow, MD Anne Connolly, MD (Chair) Richard Shell, MD	

MDA CLINICAL & SCIENTIFIC CONFERENCE 2023

Conference Agenda

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SPEAKERS ARE IN ORDER OF SPEAKING POSITION

Tuesday, March 21, 2023 cont.		
12:00 – 1:30 PM	Industry Forum Lunch Presented by Biogen *Genetic Counseling and Testing in Neuromuscular Disease: Spotlight on Spinal Muscular Atrophy and Amyotrophic Lateral Sclerosis*	Chantilly West
	Industry Forum Lunch Presented by Edgewise *Targeting Fast Muscle Myosin: A Novel Approach to Protecting Muscle in the Dystrophinopathies*	Grand ABC
	Industry Forum Lunch Presented by Fulcrum *Addressing Unmet Needs in FSHD: Data from Losmapimod Long-Term Clinical Trials*	Chantilly East
	Industry Forum Lunch Presented by NS Pharma *A Treatment Option for Patients with Duchenne Muscular Dystrophy Amenable to Exon 53 Skipping*	Grand DE
1:30 – 4:10 PM	Insights in Research Investor Summit (agenda link)	Wedgwood Ballroom
	ALS Track	Coronado A
2:00 – 4:00 PM	Gene Therapy in ALS Gene Directed Therapy for Sporadic ALS: The story of Stathmin Angela Genge, MD, FRCP An Update on our ALS Gene Therapy Programs: C9orf72 ALS Program (ALT-161) &SOD1 ALS Program (AMT-162) Meg Bradbury, MS, CGC, MSHS Neil Shneider, MD, PhD Evidence-based Consensus Guidelines for Genetic Counseling and Testing in ALS Jennifer Roggenbuck, MS, LGC 30 years of ALS Genetic Research: Separating Wheat from Chaff Matthew Harms, MD (Chair) A Tofersen Model: Clinical Optimization for SOD1 Patients Jennifer Morganroth, MD, MDBA	
	Digital Outcomes & Big Data Track	Coronado BCD
2:00 – 4:00 PM	Lab to Life: CMT Michael Shy, MD (Chair) Repairing Genetic Forms of CMT with CRISPR Bruce Conklin, MD Clinical Trials and Outcome Measures Across the Lifespan Joshua Burns, PhD Katy Eichinger, PhD, DPT Development and Assessment of Novel Biomarkers for Clinical trials in CMT John Svaren, PhD	

Tuesday, March 21, 2023 cont.		
	Practical Considerations in Gene Therapy Track (CE & Genetic Counselor CEUs Accredited Session)	Cortez AB
2:00 – 4:00 PM	Practical Management of Gene Therapy: Procurement & Administration Lessons Learned: 3 years of Onasemnogene Abeparvovec Administration & DMD GT Trials Natalie Goedeker, MSN, CPNP (Chair) Building an Infrastructure for Safe and Efficient Gene Therapy Administration Crystal Proud, MD FDA Perspective: Gene Therapy Approval Process + Fast Track Michael Singer, MD, PhD, FDA Parent Perspective: The NBS SMA Diagnosis and Treatment Journey Kody & Sydney Graves Parent Perspective: Experience of Participating in a GT Clinical Trial Brent Furbee	
	NMD Care Delivery: Care Throughout the Patient Journey Track (CE Accredited Session)	Cortez CD
2:00 – 4:00 PM	Therapy Interventions – Modify Therapy Approaches with Evolving Market Landscape Tina Duong, PT, PhD (Chair) Therapeutic Play Gym: Feasibility of a Caregiver-Mediated Exercise System for Infants and Young Children with Severe Neuromuscular Weakness Jenna Lammers, MSR/PT, CNT, PCS A Novel Approach to Optimizing Movement in Treated Children with Spinal Muscular Atrophy Megan Iammarino, PT, DPT Considerations and Guidance for Using Online or Application-Based Resources for Exercise Intervention in Patients with Neuromuscular Diseases Leslie Nelson, PT, PhD Development of a Strength Training Program in Duchenne Muscular Dystrophy Donovan Lott, PT, PhD, CSCS AI-Enabled Shoe Insoles to Assess Ambulatory Function in SMA and DMD Jacqueline Montes, PT, EdD, NCS Does a Week Really Matter? Melissa McIntyre, DPT	
3:30 – 4:30 PM	Exhibit Hall and Poster Sessions Open	Exhibit Hall
4:00 – 4:25 PM	Networking Snack Break sponsored by PepGen	Exhibit Hall

MDA 2023 CLINICAL & SCIENTIFIC CONFERENCE

Conference Agenda

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Tuesday, March 21, 2023 cont.

ALS Track		**Coronado A**
4:30 – 6:00 PM	**Industry Updates** **Current Landscape and Overview of Current ALS Clinical Trials** Jinsy Andrews, MD, MSc **Accelerating ALS Drug Development Through an Innovative Clinical Trial Design - Through the Lens of a Statistical Scientist** Melanie Quintana, PhD **Relyvrio Drug Development in ALS - Lessons to Share with the Scientific Community** Joshua Cohen & Justin Klee **The Scope of Expanded Access Programs and Open-Label Extension in ALS Clinical Trials** Suma Babu, MD, MPH, MBBS (Chair)	
Digital Outcomes & Big Data		**Coronado BCD**
4:30 – 6:00 PM	**Lab to Life: Myositis** **Evolving Classification Schema in Myositis - Where does Polymyositis Fit in this scheme** Tahseen Mozaffar, MD (Chair) **Immune Checkpoint Inhibitor-Associated Myopathy and Atypical Immune-Mediated Necrotizing Myopathy** Teerin Liewluck, MD **Inclusion Body Myositis: Update on Pathogenesis and Treatment** Thomas Llyod, MD **Idiopathic Inflammatory Myopathies and Cancer; Different Sides of the Same Coin** Eleni Tiniakou, MD **Dermatomyositis: A Diagnostic and Therapeutic Approach Based on Antibodies** Suur Bilicier, MD	
Practical Considerations in Gene Therapy (CE & Genetic Counselor CEUs Accredited Session)		**Cortez AB**
4:30 – 5:30 PM	**Clinical Challenges in the Gene Therapy Era: Roundtable Discussion of Difficult Cases** Natalie Goedeker, MSN, CPNP (Chair) Richard Finkel, MD Jerry Mendell, MD Crystal Proud, MD	
NMD Care Delivery: Care Throughout the Patient Journey (CE Accredited Session)		**Cortez CD**
4:30 – 5:30 PM	**End of Life: Care and Considerations** Lauren Treat, MD Becky Tesch Mathew Jacobson, MDiv, BCC Ambereen Mehta, MD, MPH (Chair)	
6:00 – 8:00 PM	**Poster & Networking Reception**	Exhibit Hall

MDA CLINICAL & SCIENTIFIC CONFERENCE 2023

Conference Agenda

ALL CONFERENCE SESSION TIMES LISTED ARE IN CENTRAL TIME
SPEAKERS ARE IN ORDER OF SPEAKING POSITION

Wednesday, March 22, 2023		
7:00 AM – 1:00 PM	Registration Open	Geppetto Foyer, Tower Side
New, Novel, and Noteworthy: NMD Highlights We are thrilled to have had an overwhelming number of incredible abstract submissions as we reconvene in 2023. To that end, we have redesigned our Clinical Trail 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 – 11:15 AM	Insights into Neuromuscular Disease Mechanisms	Chantilly East
8:30 – 8:45 AM	The role of pathological miRNAs in Duchenne and Becker Muscular Dystrophy Alyson Fiorillo, PhD	
8:45 – 9:00 AM	Altered muscle niche contributes to myogenic deficit in the D2-mdx model of severe DMD James Novak, PhD	
9:00 – 9:15 AM	Zebrafish and cellular model assays for the study of SELENON-Related Myopathy Pamela Barraza, PhD	
9:15 -9:30 AM	Variegated silencing in Friedreich Ataxia Sanjay Bidichandani	
9:30 – 9:45 AM	FSHD Xenografts for Biomarker Discovery: SLC34A2 Robert Bloch, PhD	
9:45 – 10:00 AM	Mitochondrial ROS signaling enables repair of injured cells and its disruption contributes to disease onset in Limb Girdle Muscular Dystrophy Marshall Hogarth, PhD	
10:00 – 10:15 AM	New concepts in the pathogenesis of TPM3-related myopathy Mathias Lambert, PhD	
10:15 – 10:30 AM	SMN acts as a genetic modifier of GEMIN5-mediated neurodegeneration Udai Pandey, PhD	
10:30-10:45 AM	Functional consequences of a human-specific tandem repeat in WDR7 associated with ALS Sam Smukowski	
10:45 – 11:00 AM	BREAK	
11:00 – 11:15 AM	Leveraging microglia states to understand ALS Martine Therrien, PhD	
11:15 AM – 12:00 PM	Improving Diagnostics and Variant Interpretation	Chantilly East
11:15 – 11:30 AM	Dystroglycanopathies: A Workflow to Improve Variant Interpretation, Disease Mechanism Understanding and Pathogenicity Predictions Kaiyue Ma	
11:30 – 11:45 AM	Evaluation of transcriptome forward computational strategies to improve molecular diagnosis in rare pediatric neuromuscular disease Sarah Silverstein	
11:45 AM – 12:00 PM	Clinical spectrum and genotype-phenotype correlations of FLVCR1-related hereditary neuropathies and neurodegeneration Daniel Calame, MD, PhD	

MDA CLINICAL & SCIENTIFIC CONFERENCE 2023

Conference Agenda

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SPEAKERS ARE IN ORDER OF SPEAKING POSITION

Wednesday, March 22, 2023 cont.		
8:30 – 10:45 AM	Novel Therapeutic Strategies	Grand DE
8:30 – 8:45 AM	Therapeutic effect of linker protein-mediated gene therapy in a mouse model for LAMA2-related muscular dystrophy Judith Reinhard, PhD	
8:45 – 9:00 AM	An RNA-mediated gene therapy approach to target polyglutamine-expanded AR co-regulators and attenuate SBMA Maria Pennuto, PhD	
9:00 – 9:15 AM	A novel calcium channel gating modifier that enhances synaptic function and maintains innervation in a mouse model of Amyotrophic Lateral Sclerosis Yomna Badawi, PhD	
9:15 -9:30 AM	PGN-EDODM1 Nonclinical Data Demonstrate Potential for Meaningful Impact in Myotonic Dystrophy Type 1 (DM1): Support for Phase 1 Clinical Trial Design Jane Larkindale, PhD	
9:30 – 9:45 AM	Efficient mini dCas13X mediated base editing for personalized treatment of Duchenne muscular dystrophy Guoling Li, PhD	
9:45 – 10:00 AM	Oral delivery of a novel noncoding RNA drug in mdx mice Alice Rannou, PhD	
10:00 – 10:15 AM	Multicenter AAV Studies for SMARD1/CMT2S Establish Dose-Dependent Efficacy in Multiple Models and Pave the Way for Initiation of a Phase I/II Trial Julieth Sierra Delgado, MD, M.Sc.	
10:15 – 10:30 AM	EPI-321, a Potential Cure for FSHD Alexandra Collin de l'Hortet	
10:30 – 10:45 AM	AOC 1020: An Antibody Oligonucleotide Conjugate (AOC) in Development for the Treatment of FSHD Barbora Malecova, PhD	
10:45 – 11:00 AM	BREAK	
11:00 AM – 12:00 PM	Patient Registries and Databases	Grand DE
11:00 – 11:15 AM	Population-based genetic epidemiology of selected muscular dystrophies: the Muscular Dystrophy Surveillance, Tracking and Research Network (MDSTARnet) Peter Kang, MD	
11:15 – 11:30 AM	Newborn Screening for Spinal Muscular Atrophy in the United States: Perspectives from Multiple Real World Data Sources Sarah Whitmire, MS	
11:30 – 11:45 AM	Comprehensive Database for Ryanodine Receptor-1 Related Disorders Joshua Todd, PhD	
11:45 AM – 12:00 PM	The United States National Amyotrophic Lateral Sclerosis (ALS) Registry Advances Research Domestically and Internationally Paul Mehta, MD, PhD	

Wednesday, March 22, 2023 cont.		
8:45 AM – 2:45 PM	Clinical Trial Updates	Chantilly West
8:45 – 9:00 AM	Results of a Phase 3 Randomized, Double-blind, Placebo-controlled Trial in Adults with Amyotrophic Lateral Sclerosis James Berry, MD	
9:00 – 9:15 AM	Results from 96 Weeks Open-Label Extension of a Phase 2 Trial of Losmapimod in Subjects with FSHD: ReDUX4 Leon Wang, MD, PhD	
9:15 -9:30 AM	Ravulizumab reduces clinical deteriorations in patients with generalized myasthenia gravis: Results from the CHAMPION MG study Tuan Vu, MD	
9:30 – 9:45 AM	Efficacy of rozanolixizumab in muscle-specific kinase antibody-positive generalized myasthenia gravis: The Phase 3 randomized MycarinG study Ali Habib, MD	
9:45 – 10:00 AM	Outcomes in Patients with Spinal Muscular Atrophy and Four or More SMN2 Copies Treated with Onasemnogene Abeparvovec: Findings from RESTORE Richard Finkel, MD	
10:00 – 10:15 AM	Baseline Characteristics and Interim Safety in RESPOND: A Phase 4 Study in Children with SMA Treated with Nusinersen After Onasemnogene Abeparvovec John Brandsema, MD	
10:15 – 10:30 AM	Apitegromab in SMA (TOPAZ trial): Covariates of Multiple Efficacy Endpoints From 24 Month Data Thomas Crawford, MD	
10:30 – 10:45 AM	Long-term efficacy and safety of cipaglucosidase alfa/miglustat in patients with Pompe disease: a Phase III open-label extension study (ATB200-07) Tahseen Mozaffar, MD	
10:45 – 11:00 AM	BREAK	
11:00 – 11:15 AM	Efficacy and safety of avalglucosidase alfa in participants with late-onset Pompe disease after 145 weeks of treatment during the COMET trial Volker Straub	
11:15 – 11:30 AM	Phase 1/2 Clinical Trial Evaluating the Safety and Pharmacokinetics of AOC 1001 in Adults with Myotonic Dystrophy Type 1: MARINA Trial in Progress Nicholas Johnson, MD, MSCI, FAAN	
11:30 – 11:45 AM	An Endovascular Motor Neuroprosthesis Enables Patients with Paralysis to Control Digital Devices by Thinking: Experience of Field Clinical Engineers Maria Nardozzi	
11:45 AM – 12:00 PM	Givinostat in DMD: results of the Epidys Study with particular attention to MR measures of muscle fat fraction Krista Vandenborne, PhD	
12:00 – 12:15 PM	Long-term safety and sustained functional benefit in patients with DMD 4 years post-treatment with delandistrogene moxeparvovec in a Phase 1/2a study Jerry Mendell, MD	
12:15 – 12:30 PM	One-year data from ENDEAVOR, a Phase 1b trial of delandistrogene moxeparvovec in patients with DMD Crystal Proud, MD	
12:30 – 1:00 PM	BREAK	

Wednesday, March 22, 2023 cont.		
1:00 – 1:15 PM	Expression of apparent full-length dystrophin in skeletal muscle after administration of the scAAV9.U7-ACCA vector: 12 to 36 month follow up Megan Waldrop, MD	
1:15 – 1:30 PM	Two Year Muscle MRI observations from a Phase 1b trial of fordadistrogene movaparvovec (PF-06939926) for Duchenne muscular dystrophy (DMD) Sarah Sherlock, PhD	
1:30 – 1:45 PM	WVE-N531 supports 53% mean exon 53 skipping in skeletal muscle of boys with Duchenne Muscular Dystrophy (DMD) after six weeks of treatment Michael Tillinger, MD	
1:45 – 2:00 PM	Assessment of Preliminary Safety and Efficacy of DT-DEC01 Therapy in Duchenne Muscular Dystrophy Patients, up-to 12-months post administration Ahlke Heydemann, PhD	
2:00 – 2:15 PM	Positive Results from a First-in-Human Study Support Continued Development of PGN EDO51 for the Treatment of Duchenne Muscular Dystrophy (DMD) Michelle Mellion, MD	
2:15 – 2:30 PM	Safety and efficacy of ataluren in nmDMD patients from Study 041, a phase 3, randomized, double-blind, placebo-controlled trial Craig McDonald, MD	
2:30 – 2:45 PM	Effects of EDG-5506, a Fast Myosin Modulator, on Biomarkers of Muscle Damage and Function in Adults with Becker Muscular Dystrophy (BMD) Han Phan, MD	
2:45 – 3:30 PM	Late-Breakers	Chantilly West
2:45 – 3:00 PM	CAP-1002, an Allogeneic Cell Therapy Demonstrates Disease Modification in Later-Stage DMD Patients: 18-Month Results from the HOPE-2-Open Label Extension Craig McDonald, MD	
3:00 – 3:15 PM	Muscle glycogen reduction in healthy adults treated with MZE001, an oral inhibitor of GYS1 and potential substrate reduction therapy for Pompe Disease Julie Ullman, PhD	
3:15 – 3:30 PM	Management of Select Adverse Events Following Delandistrogene Moxeparvovec Gene Therapy in Patients with DMD: Delphi Consensus Guidance Natalie Goedeker, CPNP	