



Annual SMA Research & Clinical Care Meeting
June 25th- June 27th, 2025

7:00 AM **Research & Clinical Care Symposium: Living with SMA: Considering Patients' Perspectives and the Evolving Landscape** , Sponsored by Novartis

Track A: Clinical Care Session		
Time	Abstract Number	Presentation Title
8:30 AM		Welcome and Cure SMA Update Mary Schroth, MD, Chief Medical Officer, Cure SMA
8:50 AM	A	Having a Plan of Attack – Outpatient and Inpatient Escalation of Care During an Acute Illness at Home and Away Hank Meyer, MD, Robert Graham, MD, Allyson Henkel, SMA Parent, Cure SMA Board Member
9:50 AM	B	Orthopedic Management Discussion Moderator: Luca Labianca, MD Effect of Scoliosis on Pulmonary Function in Spinal Muscle Atrophy (SMA) Brian Snyder, MD, PhD Trajectories of lower limb joint mobility in SMA; hip extension matters. Elizabeth Harding, PT
10:30 AM		BREAK
10:50 AM	C	A Discussion on Genitourinary Issues Angelena Edwards, MD, Douglas Storm, MD

Track B: Research Meeting		
Time	Abstract Number	Presentation Title
8:30 AM		Welcome Jackie Glascock, PhD, Chief Scientific Officer, Cure SMA
		<i>Genetic Modifiers of Disease</i>
8:45 AM	1	A SYNAPTIC CHAPERONE MODIFIES THE SMA PHENOTYPE IN THE SMN2B/- MOUSE MODEL OF THE DISEASE Narendranath Jha, PhD
9:05 AM	2	DETERMINING THE ROLE OF SARM1-MEDIATED AXON DEGENERATION IN SMA Madeline Denton, BS
9:25 AM	3	DLC1-I1 RESCUES MOTOR NEURONS IN SPINAL MUSCULAR ATROPHY BY ENHANCING SMN2 SPLICING AND AXON GROWTH, OFFERING A NOVEL THERAPEUTIC STRATEGY Tianyuan Shi, PhD
9:45 AM	4	IDENTIFICATION OF KIF5A AS A KEY FACTOR LINKING AXONAL PATHOLOGY IN SMA AND ALS Tetsuya Akiyama, MD, PhD
10:05 AM		BREAK
		<i>Gene Expression Changes & Transcript Diversity in SMA</i>
10:35 AM	5	DIRECT RNA NANOPORE SEQUENCING REVEALS EXTENSIVE TRANSCRIPT ISOFORM DIVERSITY IN CONTROL AND SMA HUMAN IPSC DERIVED MOTOR NEURONS Stephen Brown, BS
10:55 AM	6	LONG-READ SEQUENCING REVEALS NOVEL INSIGHTS INTO SMN LOCUS COMPLEXITY IN SMA Ewout Groen, PhD
11:15 AM	7	COMPREHENSIVE SPATIAL TRANSCRIPTOME ANALYSIS OF SPINAL CORD AND SURROUNDING TISSUES IN NORMAL AND SMA MODEL MICE Elliot Androphy, MD
11:35 AM	8	GENE EXPRESSION CHANGES IN MOTOR NEURONS IN A LARGE ANIMAL MODEL OF MOTOR NEURON DISEASE Megan Baird, PhD

12:00 PM align="center">**LUNCH**



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Combined Research & Clinical Care Meeting	
Clinical CME Content Provided	
Abstract Number	Presentation Title
	<i>Neurocognition & SMA</i> <i>Moderator: Julie Parsons, MD</i>
1:00 PM	9 PANEL DISCUSSION: Uncovering Cognitive Implications in Spinal Muscular Atrophy: Research Gaps, Clinical Insights, and Future Directions Sarah Wright, DO, Eduardo Tizzano, MD, PhD, Gretchen Weatherly, PhD, LP
2:00 PM	10 Investigating the developmental expression of SMN in the brain in a mouse model of Spinal Muscular Atrophy Charalambos Demetriou, PhD
2:20 PM	BREAK
	<i>Understanding Bulbar Function: Impacts of Disease and Treatment in Mice & Man</i>
2:40 PM	11 Creating human hindbrain motor neurons: a platform to model bulbar symptoms in SMA Kyle Loh, PhD
3:00 PM	12 Defects of sensory-motor circuits - controlling facial muscles - may provide mechanistic insights in suckling and masticatory behavioral deficits in SMA mice Danny Florez-Paz, PhD
3:20 PM	13 Differences in Swallowing Physiology Between Pre- and Post-Symptomatically Treated Patients with SMA Katlyn McGrattan, PhD
3:40 PM	14 Enhancing Bulbar Assessment in Spinal Muscular Atrophy: A Rasch Analysis of the International Bulbar Assessment Tool (iBAT) Pilot Study Sally Dunaway Young, PT, DPT
4:30 PM	Poster Session A: Odd Numbered Posters Presented



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Abstract Number	<u>Thursday June 26th, 2025</u>
7:00 AM	<p><i>Research & Clinical Care Symposium: An Evolving Treatment for SMA: Discover a New Formulation and the Latest Clinical Results Across SMA Types and Age Groups</i> <i>Sponsored by Genentech</i></p> <p><i>Neurodevelopmental Deficits, Neurodegeneration, and Capacity for Regeneration after Treatment in SMA</i></p>
8:30 AM	<p>15 Aberrant Angiogenesis, Blood–Spinal Cord Barrier Compromise, and Neuroinflammation in Severe Spinal Muscular Atrophy Patient Spinal Cord Hazel Allardyce, PhD</p>
8:50 AM	<p>16 Premature differentiation of motor neuron progenitors renders motor neurons vulnerable in spinal muscular atrophy Zeynep Dokuzluoğlu, MscR.</p>
9:10 AM	<p>Profile time course of motor unit regeneration and stability following administration of Smn-upregulating therapeutics Inga Partlova, MscR</p> <p align="center"><i>Phenotypic and Genotypic Considerations in SMA Treatment</i></p>
9:30 AM	<p>17 Spinal muscular atrophy among U.S. Hutterites: Phenotype Variability in the setting of conserved ancestral haplotype and 4 SMN2 copies Kathryn Swoboda, MD</p>
9:50 AM	<p>18 Novel SMN1 mutations identified by newborn screening: Therapeutic hurdles and functional solutions Brunhilde Wirth, PhD</p>
10:10 AM	BREAK
10:40 AM	<i>Adults Living with SMA: Panel Q&A</i>
11:45 AM	LUNCH
12:00 PM	<p><i>Research & Clinical Care Symposium: Muscle Matters, Understanding the Role of Muscle in SMA</i> <i>Sponsored by Scholar Rock</i></p> <p align="center"><i>Assessing the Adult Experience: Outcomes and Biomarkers</i></p>
1:00 PM	<p>19 Exploring Quantitative Muscle Testing Among Treated Adults with Spinal Muscular Atrophy: A Real-World Perspective Constance de Monts, PT, DPT</p>
1:20 PM	<p>20 Long-Term Motor Responses to Disease-Modifying Therapies in Spinal Muscular Atrophy (SMA) Adults: A Prospective Study Tina Duong, MPT, PhD</p>
1:40 PM	<p>21 Longitudinal evaluation of adult patients with type 3 SMA with spinal cord imaging and motor scales: in search of new biomarkers Felipe Franco da Graça, MD</p>
2:00 PM	BREAK
	<i>Outcome Measures for Research and Clinical Care</i>
2:20 PM	<p>22 Quantitative description of movement and posture in infants with SMA using a wearable sensor suit and a Machine Learning algorithm</p>



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		Jinseok Oh, PhD
2:40 PM	23	Gait Speed in SMA: a Key Vital Sign Linked to Strength and Fatigability Carly Mueller, <i>PT, DPT, PCS</i>
3:00 PM	24	Rasch Analysis of the Spinal Muscular Atrophy Person-Reported Outcome Measure (SMA-PRO) Mobility Domain in Children and Adults with SMA. Amy Pasternak, <i>PT, DPT, PCS</i>
3:20 PM	25	Retrospective Assessment of feeding and Nutrition after 2 years of Risdiplam Treatment in children with Type 1 SMA using a novel scale Giovanni Baranello MD, PhD
4:00 PM		Poster Session B: Even Numbered Posters Presented
6:30 PM		Meet and Greet with Families: Featuring Annual Relay Race



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Abstract Number	Friday June 27th, 2025
7:00 AM	Research & Clinical Care Symposium: Elevating Expectations in SMA Care: Optimizing Patient Outcomes <i>Sponsored by Biogen</i>
8:30 AM	Podium Poster Highlights
	Clinical Therapeutic Development
9:10 AM	25 First-in-human study of epidural spinal cord stimulation in individuals with spinal muscular atrophy Genis Prat-Ortega, PhD
9:30 AM	26 PIERRE Clinical Trial: Safety and Performance of ThecaFlex DRx™: an Implantable Intrathecal Catheter and Subcutaneous Port System for Repeated Intrathecal Delivery of Nusinersen Cassandra Dennys, PhD
9:50 AM	27 NURTURE Study: Long-term Benefits of Nusinersen in Presymptomatic Spinal Muscular Atrophy (SMA) over 8-years of Follow-up Nancy L Kuntz, MD
10:10 AM	BREAK
10:40 AM	28 DEVOTE Part C Results: Exploring Higher Doses of Nusinersen in Nusinersen - Experienced Participants with Spinal Muscular Atrophy Richard Finkel, MD
11:00 AM	29 Efficacy and safety of apitegromab in individuals with type 2 and type 3 spinal muscular atrophy evaluated in the phase 3 SAPPHIRE trial Basil T. Darras, MD
11:20 AM	30 Intrathecal Onasemnogene Abeparvovec for Patients with Spinal Muscular Atrophy: Phase 3, Randomized, Sham-Controlled, Double-Blind STEER Study Crystal M. Proud, MD
11:40 AM	31 Intrathecal Onasemnogene Abeparvovec for Treatment-Experienced Patients with Spinal Muscular Atrophy: Phase 3b, Open-Label STRENGTH Jennifer M. Kwon MD, MPH, FAAN
12:00 PM	Closing Remarks Jackie Glascock, PhD, Vice President Research, Cure SMA
12:10 PM	Research and Clinical Care Meeting Adjourns
6:30 PM	Family Friendly Researcher Poster Session