



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 Mayer, 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, EdD
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
		Genetic Modifiers of Disease Moderator: Rashmi Kothary, PhD
8:45 AM	1	A SYNAPTIC CHAPERONE MODIFIES THE SMA PHENOTYPE IN THE SMN2B/- MOUSE MODEL OF THE DISEASE Narendra Nath 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
		Gene Expression Changes & Transcript Diversity in SMA Moderator: Arthur Burghes, PhD
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	IDENTIFICATION OF EARLY SYMPTOMATIC GENE EXPRESSION CHANGES IN MOTOR NEURONS IN A LARGE ANIMAL MODEL OF MOTOR NEURON DISEASE Megan Baird, PhD

12:00 PM

LUNCH



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

Combined Research & Clinical Care Meeting		
Clinical CME Content Provided		
	Abstract Number	
		NEUROCOGNITION & SMA MODERATOR: JULIE PARSONS, MD
1:00 PM	9	INVESTIGATING THE DEVELOPMENTAL EXPRESSION OF SMN IN THE BRAIN IN A MOUSE MODEL OF SPINAL MUSCULAR ATROPHY Charalambos Demetriou, PhD
1:20 PM	10	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
2:20 PM		BREAK
		UNDERSTANDING BULBAR FUNCTION: IMPACTS OF DISEASE AND TREATMENT IN MICE & MAN Moderator: Katherine Klinger, PhD
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



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

	Abstract Number	Thursday June 26 th , 2025
7:00 AM		RESEARCH & CLINICAL CARE SYMPOSIUM: AN EVOLVING TREATMENT FOR SMA: DISCOVER A NEW FORMULATION AND THE LATEST CLINICAL RESULTS ACROSS SMA TYPES AND AGE GROUPS Sponsored by Genentech
		NEURODEVELOPMENTAL DEFICITS, NEURODEGENERATION, AND CAPACITY FOR REGENERATION AFTER TREATMENT IN SMA Moderator: Charlotte Sumner, MD
8:30 AM	15	ABERRANT ANGIOGENESIS, BLOOD-SPINAL CORD BARRIER COMPROMISE, AND NEUROINFLAMMATION IN SEVERE SPINAL MUSCULAR ATROPHY PATIENT SPINAL CORD Hazel Allardyce, PhD
8:50 AM	16	PREMATURE DIFFERENTIATION OF MOTOR NEURON PROGENITORS RENDERS MOTOR NEURONS VULNERABLE IN SPINAL MUSCULAR ATROPHY Zeynep Dokuzluoğlu, MscR.
9:10 AM	17	PROFILE TIME COURSE OF MOTOR UNIT REGENERATION AND STABILITY FOLLOWING ADMINISTRATION OF SMN-UPREGULATING THERAPEUTICS Inga Partlova, MscR
		Phenotypic and Genotypic Considerations in SMA Treatment Moderator: Kathryn Swoboda, MD
9:30 AM	18	SPINAL MUSCULAR ATROPHY AMONG U.S. HUTTERITES: PHENOTYPE VARIABILITY IN THE SETTING OF CONSERVED ANCESTRAL HAPLOTYPE AND 4 SMN2 COPIES Kathryn Swoboda, MD
9:50 AM	19	NOVEL SMN1 MUTATIONS IDENTIFIED BY NEWBORN SCREENING: THERAPEUTIC HURDLES AND FUNCTIONAL SOLUTIONS Brunhilde Wirth, PhD
10:10 AM		BREAK
10:30 AM		ADULTS LIVING WITH SMA: PANEL Q&A Panelists: Brad Nunemaker, Lyza Weisman, Carolyn Barrett Moderators: Lyndsay Murray, PhD Tom Crawford, MD
11:30 AM		LUNCH
11:45 PM		RESEARCH & CLINICAL CARE SYMPOSIUM: MUSCLE MATTERS, UNDERSTANDING THE ROLE OF MUSCLE IN SMA Sponsored by Scholar Rock
		Assessing the Adult Experience: Outcomes and Biomarkers Moderator: Stephen Kolb, MD, PhD
1:00 PM	20	EXPLORING QUANTITATIVE MUSCLE TESTING AMONG TREATED ADULTS WITH SPINAL MUSCULAR ATROPHY: A REAL-WORLD PERSPECTIVE Constance de Monts, PT, DPT
1:20 PM	21	LONG-TERM MOTOR RESPONSES TO DISEASE-MODIFYING THERAPIES IN SPINAL MUSCULAR ATROPHY (SMA) ADULTS: A PROSPECTIVE STUDY Tina Duong, MPT, PhD



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

1:40 PM	22	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
2:00 PM		BREAK
		<i>Outcome Measures for Research and Clinical Care</i> Moderator: Allison Ebert, PhD
2:20 PM	23	QUANTITATIVE DESCRIPTION OF MOVEMENT AND POSTURE IN INFANTS WITH SMA USING A WEARABLE SENSOR SUIT AND A MACHINE LEARNING ALGORITHM Jinseok Oh, PhD
2:40 PM	24	GAIT SPEED IN SMA: A KEY VITAL SIGN LINKED TO STRENGTH AND FATIGABILITY Carly Mueller, PT, DPT, PCS
3:00 PM	25	RASCH ANALYSIS OF THE SPINAL MUSCULAR ATROPHY PERSON-REPORTED OUTCOME MEASURE (SMA-PRO) MOBILITY DOMAIN IN CHILDREN AND ADULTS WITH SMA. Amy Pasternak, PT, DPT, PCS
3:20 PM	26	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



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

	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 <i>Moderator: Jackie Glascock, PhD</i>
		CLINICAL THERAPEUTIC DEVELOPMENT <i>Moderator: Adrian Krainer, PhD</i>
9:10 AM	27	FIRST-IN-HUMAN STUDY OF EPIDURAL SPINAL CORD STIMULATION IN INDIVIDUALS WITH SPINAL MUSCULAR ATROPHY Genis Prat-Ortega, PhD
9:30 AM	28	PIERRE CLINICAL TRIAL: SAFETY AND PERFORMANCE OF THECAFLEX DRX™: AN IMPLANTABLE INTRATHECAL CATHETER AND SUBCUTANEOUS PORT SYSTEM FOR REPEATED INTRATHECAL DELIVERY OF NUSINERSEN Kathrin Meyer, PhD
9:50 AM	29	TALDEFGROBEP ALFA IN SPINAL MUSCULAR ATROPHY: RESULTS OF THE PHASE 3 RESILIENT STUDY Lindsey Lair, MD
10:10 AM	30	NURTURE STUDY: LONG-TERM BENEFITS OF NUSINERSEN IN PRESYMPTOMATIC SPINAL MUSCULAR ATROPHY (SMA) OVER 8-YEARS OF FOLLOW-UP Nancy L Kuntz, MD
10:30 AM		BREAK
11:00 AM	31	DEVOTE PART C RESULTS: EXPLORING HIGHER DOSES OF NUSINERSEN IN NUSINERSEN-EXPERIENCED PARTICIPANTS WITH SPINAL MUSCULAR ATROPHY Richard Finkel, MD
11:20 AM	32	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
11:40 AM	33	INTRATHECAL ONASEMNOGENE ABEPARVOVEC FOR PATIENTS WITH SPINAL MUSCULAR ATROPHY: PHASE 3, RANDOMIZED, SHAM-CONTROLLED, DOUBLE-BLIND STEER STUDY Crystal M. Proud, MD
12:00 PM	34	INTRATHECAL ONASEMNOGENE ABEPARVOVEC FOR TREATMENT-EXPERIENCED PATIENTS WITH SPINAL MUSCULAR ATROPHY: PHASE 3B, OPEN-LABEL STRENGTH Jennifer M. Kwon MD, MPH, FAAN
12:20 PM		CLOSING REMARKS Jackie Glascock, PhD, Chief Scientific Officer, Cure SMA
12:30 PM		RESEARCH AND CLINICAL CARE MEETING ADJOURNS
6:30 PM		FAMILY FRIENDLY RESEARCHER POSTER SESSION