



Annual SMA Research & Clinical Care Meeting

June 28-30, 2023

	<i>Abstract Number</i>	<u>Wednesday June 28th, 2023</u>
7:00 AM		Research Breakfast with Symposium Sponsored by Biogen: SMN Restoration in Adults Living with SMA
8:30 AM		Welcome Jackie Glascock, PhD, Vice President, Research, Cure SMA
8:35 AM		Basic Research: Session 1 Moderator: Elliot Androphy, MD
8:35 AM	1	TOWARDS A PRENATAL THERAPY FOR PATIENTS WITH SEVERE SPINAL MUSCULAR ATROPHY - PRECLINICAL EXPERIMENTS IN MOUSE AND SHEEP MODELS Beltran Borges, MD
8:55 AM	2	TARGETING HISTONE ACETYLATION SPECIFICALLY TO SMN2 INTRON 6 USING A CRISPR-DEAD CAS9 STRATEGY COOPERATES WITH NUSINERSEN TO PROMOTE EXON 7 INCLUSION Alberto Kornblihtt, PhD
9:15 AM	3	A PERMANENT GENETIC TREATMENT FOR SPINAL MUSCULAR ATROPHY USING BASE EDITORS Christiano Alves, PhD
9:35 AM	4	MOTOR UNIT RECOVERY FOLLOWING AAV9-SMN TREATMENT IN A MOUSE MODEL OF SPINAL MUSCULAR ATROPHY Laura Comley, PhD
9:55 AM	5	SURVIVAL MOTOR NEURON (SMN) GENE THERAPY: ADDRESSING CONCERNS OF SENSORIMOTOR TOXICITY Maria Balch, PhD
10:15 AM		Coffee Break
10:45 AM		Basic Research Session 2 Moderator: Rashmi Kothary, PhD
10:45 AM	6	CEREBELLAR STRUCTURAL, GLIAL, AND NEURONAL DEFECTS IN A MOUSE MODEL OF SPINAL MUSCULAR ATROPHY Nicholas Cottam, BS
11:05 AM	7	CEREBELLAR PATHOLOGY IN TWO MOUSE MODELS FOR SPINAL MUSCULAR ATROPHY Florian Gerstner, MS
11:25 AM	8	HUMAN SPINAL CORD ORGANOID MODELS OF SPINAL MUSCULAR ATROPHY EXHIBIT EARLY NEURODEVELOPMENTAL DEFECTS Natalia Rodriguez-Muela, PhD
11:45 AM		Lunch
1:00 PM		Basic Research Session 3 Moderator: Adrian Krainer, PhD



1:00 PM	9	ONE RING TO RULE THEM ALL: ARE SM-RING ASSOCIATED FUNCTIONS OF SMN CENTRAL TO THE DEVELOPMENT OF SMA? Anton Blatnik, PhD
1:20 PM	10	RAPID CYTOPLASMIC PHASE SEPARATION OF SMN IN RESPONSE TO CELLULAR STRESS Oliver Gruss, PhD
1:40 PM	11	MIR34 CONTRIBUTES TO SPINAL MUSCULAR ATROPHY (SMA) AND AAV9-MEDIATED DELIVERY OF MIR34A AMELIORATES THE MOTOR DEFICITS IN SMA MICE Tai-Heng Chen, MD, PhD
2:00 PM	12	NUCLEOLIN, AN SMN INTERACTING PROTEIN, BINDS WD-40 DOMAIN OF ACOP THROUGH ITS DILYSINE MOTIF Timra Gilson, PhD
2:20 PM		Break
2:40 PM	13	SMA PROTECTIVE MODIFIER PLASTIN 3 IS EPIGENETICALLY REGULATED BY THE MACROSATELLITE DXZ4 AND THE TRANSCRIPTION REGULATOR CHD4 Brunhilde Wirth, PhD
3:00 PM	14	MITIGATING EFFECTS OF A SYNAPTIC CHAPERONE IN INTERMEDIATE SMN2B/- MODEL MICE Yoon Ra Her, PhD
3:20 PM	15	COMBINATORIAL THERAPY USING A ZOLGENSMA-LIKE DRUG WITH MIFEPRISTONE TO TARGET WHOLE-BODY PATHOLOGIES IN THE SMN2B/- MOUSE MODEL OF SMA Emma Sutton, PhD
4:30 PM		Poster Session A: Odd Numbered Posters Presented
		<u>Thursday June 29th, 2023</u>
7:00 AM		Researcher Breakfast with Symposium <i>Sponsored by Genentech: Addressing the Evolving Needs of the SMA Community: The Next Phase of Evidence Generation</i>
8:30 AM		Special Session: The State of SMA – Towards Improved Therapeutics and Care
8:30 AM		Part 1: The Evolving SMA Patient Population: Unmet Needs and impact of Early Treatment Moderator: Charlotte Sumner, MD
8:30 AM		INTRODUCTION TO THE SESSION Charlotte Sumner, MD and Stephen Kolb, PhD
8:40 AM		SMA IN THE UNITED STATES: PERSPECTIVES FROM MULTIPLE REAL WORLD DATA SOURCES Lisa Belter, MPH and Sarah Whitmire
9:00 AM		WHERE ARE THEY NOW? THE SMA NEWBORN SCREENING IDENTIFIED INFANTS' CLINICAL JOURNEY



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		Crystal Proud, MD
9:20 AM		WHAT'S NEXT? DRUGS IN THE PIPELINE Thomas Crawford, MD
9:40 AM		Part 1: Panel Discussion
10:20 AM		Coffee Break
10:40 AM		Part 2: The Importance of Understanding SMA pathobiology and Identification of New Druggable Targets Moderator: Stephen Kolb, MD, PhD
10:40 AM		WHAT GENETICS AND BIOCHEMISTRY HAVE TOLD US ABOUT SMA AND SMN: WHERE WE GO FROM HERE Arthur Burghes, PhD
11:00 AM		THE ROLE OF SMN AT THE NEUOMUSCLAR JUNCTION: KNOWN FUNCTIONS AND REMAINING QUESTIONS Umrao Monani, PhD
11:20 AM		UNDERSTANDING NEURONAL VULNERABILITY: LESSONS FROM MOUSE MODELS Lyndsay Murray, PhD
11:40 AM		Part 2: Panel Discussion
12:20 PM		Lunch
1:20 PM		Clinical Research Session Moderator: Kathy Klinger, PhD
1:20 PM	16	ASSESSING PERCEIVED FATIGABILITY IN ADOLESCENTS AND ADULTS WITH SMA; RESULTS FROM A PILOT STUDY Rafael Rodriguez-Torres, DPT
1:40 PM	17	MUSCLE ULTRASOUND IN CHILDREN WITH SMA TREATED WITH EARLY MONO- OR DUAL THERAPY Susan E. Matesanz, MD
2:00 PM	18	SERUM NEUROFILAMENT LEVELS AND MAXIMUM ULNAR CMAP IN THE REAL-WORLD SETTING TO MONITOR IMPACT OF MOLECULAR THERAPIES ON CLINICAL OUTCOMES IN INFANTS WITH SMA INDICATE A POTENTIAL PROTECTIVE EFFECT OF NUSINERSEN DOSING PRIOR TO IV ONASEMNOGENE DOSING IN VULNERABLE SMA INFANTS Kathryn Swoboda, MD
2:20 PM	19	PRELIMINARY RESULTS FROM AN ONGOING PILOT CLINICAL TRIAL TESTING THE EFFECTS OF SPINAL CORD STIMULATION ON MOTOR FUNCTION IN HUMANS WITH TYPE 3 SMA Marco Capogrosso, PhD
2:40 PM	20	RISDIPLAM EXPERIENCE FOLLOWING ONASEMNOGENE ABEPARVOVEC IN INDIVIDUALS



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		WITH SPINAL MUSCULAR ATROPHY: A MULTICENTER CASE SERIES Melissa D Svoboda, MD
3:00 PM	21	EVALUATION OF MOLECULAR RESPONSE TO RISDIPLAM IN SMA PATIENTS THROUGH THE ANALYSIS OF SMN2 TRANSCRIPT LEVELS ON PERIPHERAL BLOOD Emanuela Abiusi, MD
4:00 PM		Poster Session B: Even Numbered Posters Presented
		<u>Friday June 30th, 2023</u>
7:00 AM		<i>Research Breakfast with Symposium</i> <i>Sponsored by Novartis Gene Therapies: How Far We've Come with SMA: Two Patients' Journeys</i>
8:30 AM		Podium Poster Highlights Moderator: Jackie Glascock, PhD
9:40 AM		Clinical Drug Development Session Moderator: Samuel Pfaff, PhD
9:40 AM	22	INTERIM RESULTS FROM THE ONGOING RESPOND STUDY EVALUATING NUSINERSEN IN CHILDREN WITH SPINAL MUSCULAR ATROPHY PREVIOUSLY TREATED WITH ONASEMNOGENE ABEPARVOVEC Crystal Proud, MD
10:00 AM	23	FIREFISH PARTS 1 AND 2: 4-YEAR EFFICACY AND SAFETY OF RISDIPLAM IN TYPE 1 SPINAL MUSCULAR ATROPHY Giovanni Baranello, MD
10:20 AM	24	SUNFISH PARTS 1 AND 2: 4-YEAR EFFICACY AND SAFETY DATA OF RISDIPLAM IN TYPES 2 AND 3 SPINAL MUSCULAR ATROPHY (SMA) John W. Day MD, PhD
10:40 AM	25	EFFECT OF APITEGROMAB ON PEDI-CAT AND PROMIS-FATIGUE QUESTIONNAIRE AT 36-MONTHS IN PATIENTS WITH TYPE 2 AND NONAMBULATORY TYPE 3 SPINAL MUSCULAR ATROPHY Thomas O. Crawford, MD
11:00 AM	26	EFFECT OF APITEGROMAB ON MOTOR FUNCTION AT 36-MONTHS IN PATIENTS WITH TYPE 2 AND NONAMBULATORY TYPE 3 SPINAL MUSCULAR ATROPHY Thomas O. Crawford, MD
11:20 AM		Closing Remarks Jackie Glascock, PhD, Vice President Research, Cure SMA
11:30 AM		Research and Clinical Care Meeting Adjourns