Friday June 28, 2019

7:00 AM  Researcher Breakfast with Symposium: Sponsored by AveXis
          TBD

8:30 AM  Welcoming Remarks
          Jill Jarecki PhD, Cure SMA Chief Scientific Officer

Special Session: Spinal and Neuromuscular Circuitry: Exploring defects in SMA and Potential Therapeutic Targets, Moderator, Samuel Pfaff PhD, Professor, Gene Expression Laboratory, Howard Hughes Medical Institute Investigator, Benjamin H. Lewis Chair

Understanding how the intricate circuitry connecting nerves and muscles functions and what defects in that circuitry are present in SMA is critical for better understanding the pathology of SMA. Furthermore, identifying defects in the circuitry may identify potential targets for future therapeutics. The goal of this session is to better understand this circuitry in the context of SMA and discuss how this knowledge may shape and impact further therapeutic development.

8:35 AM  Circuit Based Research in the Spinal Cord
          Samuel Pfaff PhD, Chief, Professor, Gene Expression Laboratory, Howard Hughes Medical Institute Investigator, Benjamin H. Lewis Chair, Salk Institute for Biological Studies

9:00 AM  Sensory-Motor Dysfunction in SMA
          George Mentis, PhD, Associate Professor in Pathology and Cell Biology, Member Faculty of Motor Neuron Center, Columbia University

9:25 AM  Dysfunction of the Spinal-Neuromuscular Circuitry in SMA
          Chien-Ping Ko, PhD, Professor of Biological Sciences, University of Southern California

9:50 AM  Break

10:20 AM  Christin Simon: Chronic Increase of Neuronal Activity by 4-Aminopyridine Improves Sensory-Motor Dysfunction in a Mouse Model of SMA

10:40 AM  Jianli Sun: The Function of Cerebellum and Motor Cortex Output Neurons in Spinal Muscular Atrophy Mice

11:00 AM  Closing Remarks from Moderator and Panel Discussion

11:30 AM  Lunch

11:30 PM  Researcher Lunch with Symposium: Sponsored by Biogen
The Evolving Clinical Management of Spinal Muscular Atrophy: From Diagnosis to Transition of Care

Basic Research 1: Identification of Candidate Therapeutic Targets and Disease Modifiers
Session Moderator: Umrao Monani, PhD

1:00 PM  Plenary Talk: Bridging Biophysics and Neurology: Aberrant Phase Transitions in Neurological Diseases
J. Paul Taylor, MD, PhD, Chair, Cell & Molecular Biology Department, Investigator Howard Hughes Medical Institute, Associate Director Basic Research Comprehensive Cancer Center, Edward F. Barry Endowed Chair in Cell and Molecular Biology St Jude Hospital

1:25 PM  Kevin Kaifer: SMN-Independent Therapeutic Targets: miR-23a, a Novel Protective Modifier in SMA Model Mice

2:45 PM  Brunhilde Wirth: ASO-mediated NCALD Reduction Ameliorates Spinal Muscular Atrophy Pathology, while Homozygous Ncald Knockout Impairs Adult Neurogenesis

2:05 PM  Yongchao Ma: Targeting Mitochondrial Dynamics to Alleviate Disease Symptoms in SMA Mice

2:25 PM  Darija Šoltić: Lamin A is Dysregulated in Spinal Muscular Atrophy

2:45 PM  Laxman Gangwani: Role of Senataxin in Degeneration and Prevention of Motor Neurons in Spinal Muscular Atrophy

3:05 PM  Joe Hoolachan: Identification of New Muscle-Specific Targets in SMA

3:25 PM  Min Jeong Kye: Muscle-Secre ted CTRP3 Enhances SMN Protein Synthesis in SMA Motor Neurons

3:45 PM  Monica Nizzardo: Cell Penetrating Peptide-Conjugated Morpholino: An Improved and Promising Treatment for SMA Symptomatic Cases

4:05 PM  Christiano Alves: Analysis of Transcriptome Profiles from Whole-Blood Reveals Down-Regulation of Actin Cytoskeleton Pathway and Immunosuppression in Spinal Muscular Atrophy Type 3 Subjects

4:30 PM  Poster Session A and Cocktail Reception
Odd Numbered Posters Presented

6:30 PM  Meet and Greet and Annual Relay Race with SMA Families
Annual Relay Race for Researchers and Kids

Saturday June 29
### Basic Research 2: SMN Function, Expression, and Splicing
Session Moderator: Adrian Krainer, PhD

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<tr>
<td>8:30 AM</td>
<td><strong>Plenary Talk: Developmental and Temporal Needs of SMN Protein</strong></td>
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<td>Charlotte Sumner, MD, Professor of Neurology, Johns Hopkins</td>
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<td>8:50 AM</td>
<td>Daniel Ramos: SMN Expression in Normal Development, Disease, and Post-therapy: Implications for Treating SMA patients</td>
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<td>9:10 AM</td>
<td>Utz Fischer: Regulation of the SMN complex by the mTOR pathway and its Implications for SMA</td>
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<td>9:30 AM</td>
<td>Alberto Kornblitt: Why Histone Deacetylase Inhibitors Help Upregulation of SMN2 Exon 7 Inclusion by Antisense Oligonucleotides</td>
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<td>9:50 AM</td>
<td>Sarah Tisdale: SMN Regulates Neuromuscular Junction Integrity Through U7 snRNP</td>
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<td>10:10 AM</td>
<td><strong>Break</strong></td>
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### Basic Research 3: SMA Pathology and Tissue Requirements
Session Moderator: Christine DiDonato, PhD

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<tr>
<td>10:40 AM</td>
<td>Jeong-Ki Kim: Low SMN Protein in Muscle Acts in a Cell-Autonomous Manner to Trigger a Late-Onset Neuromuscular Disease Phenotype</td>
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<td>11:00 AM</td>
<td>Wilfried Rossoll: SMA-Specific Differences in the Translatome of Motor Neurons In Vivo</td>
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<td>11:20 AM</td>
<td>Lingling Kong: Serum Neurofilament Light Chain is a Biomarker of Early Postnatal Degeneration of Immature SMA Motor Axons</td>
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<td>11:45 PM</td>
<td><strong>Boxed Lunch and Poster Session B</strong></td>
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<td>Even Numbered Posters Presented</td>
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### Clinical Research Studies for SMA
Session Moderator: Tom Crawford, MD

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<tr>
<td>1:50 PM</td>
<td><strong>Plenary Talk: Fluid-based biomarkers for ALS</strong></td>
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<td>Robert Bowser, PhD, Chairman, Professor of Neurology and Neurobiology, Barrow Neurological Institute &amp; St. Joseph's Hospital and Medical Center</td>
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<td>2:15 PM</td>
<td>Thomas Crawford: Comparison of Phosphorylated Neurofilament Heavy Chain (pNF-H) and Neurofilament Light Chain (NF-L) Concentrations in Cerebrospinal Fluid (CSF) Among Multiple Spinal Muscular Atrophy (SMA) Populations</td>
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<tr>
<td>2:30 PM</td>
<td>Charlotte Sumner: Phosphorylated Neurofilament Heavy Chain (pNF-H) and Motor Function Achievement in Nusinersen-Treated Individuals With Spinal Muscular Atrophy (SMA)</td>
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2:45 PM  Jacqueline Montes: Diminished Muscle Oxygenation During Exercise in Ambulatory Spinal Muscular Atrophy Patients

3:05 PM  Break

3:30 PM  Bart Bartels: The Endurance Shuttle Tests as Outcome Measures for Fatigability in Spinal Muscular Atrophy: Validity and Reliability

3:50 PM  Lindsay Alfano: Development of the Neuromuscular Gross Motor Outcome as an Outcome Measure in Spinal Muscular Atrophy

4:10 PM  Linda Lowes: Pre-symptomatic Spinal Muscular Atrophy: Reality or Myth?

4:30 PM  Katlyn McGrattan: Natural History of Physiologic Swallowing Deficits in Spinal Muscular Atrophy Type 1

6:30 PM  Evening Activity: Family Friendly Poster Sessions with SMA Families

Sunday June 30

8:30 AM  Podium Poster Highlights
           Session Moderator: Jacqueline Glascock, PhD
           Poster presentations are selected by the Cure SMA SAB for five-minute presentations

Clinical Drug Development
           Session Moderator: Stephen Kolb, MD, PhD

9:05 AM  John Day: Onasemnogene Abeparvovec Gene-Replacement Therapy for Spinal Muscular Atrophy Type 1: Pivotal Phase 3 Study (STR1VE) Update

9:20 AM  TBD: Onasemnogene Abeparvovec Gene-Replacement Therapy in Presymptomatic Spinal Muscular Atrophy (SPRINT)

9:35 AM  Richard Finkel: Intrathecal Administration of Onasemnogene Abeparvovec Gene-Replacement Therapy for Spinal Muscular Atrophy Type 2 (STRONG)

9:50 AM  Julie Parsons: Nusinersen in Infants who Initiate Treatment in a Presymptomatic Stage of Spinal Muscular Atrophy (SMA): Interim Efficacy and Safety Results from the Phase 2 NURTURE study

10:10 AM  Basil Darras: Interim Report on the Safety and Efficacy of Longer-Term Treatment with Nusinersen in Later-Onset Spinal Muscular Atrophy (SMA): Results from the SHINE Study
10:30 AM    Giovanni Baranello: FIREFISH Part 1: 1-year Results on Motor Function in Infants with Type 1 Spinal Muscular Atrophy (SMA) Receiving Risdiplam (RG7916)

10:45 AM    Basil Darras: FIREFISH Part 1: Survival, Ventilation and Swallowing Ability in Infants with Type 1 Spinal Muscular Atrophy (SMA) Receiving Risdiplam (RG7916)

11:00 AM    Laurent Servais: SUNFISH Part 1: Safety, Tolerability, Pharmacokinetics (PK), Pharmacodynamics (PD) and Exploratory Efficacy Data in Patients with Type 2 or 3 Spinal Muscular Atrophy (SMA) Receiving Risdiplam (RG7916)

11:10 AM    Yung Chyung: Interim Results from a Phase 1 Study of SRK-015, a Fully Human Monoclonal Antibody that Inhibits Myostatin Activation

11:30 AM    Marloes Stam: SPACE trial. A Phase II, Monocenter, Double-Blind, Placebo-Controlled, Cross-Over Trial to Assess Efficacy of Pyridostigmine in Patients with Spinal Muscular Atrophy Types 2, 3 and 4

11:50 AM    Closing Remarks
            Jacqueline Glascock, PhD, Senior Scientific Program Manager, Cure SMA

12:00 PM    Meeting Adjourns