



**SMA**  
FOUNDATION

The SMA Foundation has invested over \$100M to support basic, translational and clinical research that will accelerate progress towards a treatment for Spinal Muscular Atrophy (SMA). For a list of active and completed research projects and programs, please see below.

## RESEARCH PORTFOLIO

### Active

#### **DNA Learning Center** (Cold Spring Harbor Laboratory)

SMA educational website

#### **Enzo Life Sciences** (formerly Assay Designs)

Optimization and technical support of quantitative enzyme-linked immunosorbent assay (ELISA) for SMN protein

#### **Isis Pharmaceuticals**

Antisense oligonucleotide therapeutics for SMA

#### **Samie Jaffrey, M.D., Ph.D.** (Cornell University)

Use of axon-specific ablation of SMN to explore RNA (*as part of the Axon Working Group grant*)

#### **Jasper Clinic**

Pilot study of SMA biomarkers in blood

Optimization of SMN protein measurements in blood

#### **Chien-Ping Ko, Ph.D.** (University of Southern California)

Synapse loss in mouse models of SMA

#### **Adrian Krainer, Ph.D.** (Cold Spring Harbor Laboratory)

Effects of systemic and CNS administration of ISIS-SMN<sub>RX</sub>

#### **Cathleen Lutz, Ph.D.** (The Jackson Laboratory)

Genetic models for SMA research and therapeutics development

#### **George Mentis, Ph.D.** (Columbia University)

Characterization of the physiological mechanisms in motor neuron dysfunction in SMA (*as part of the Motor Neuron Center grant*)

#### **Motor Neuron Center** (Columbia University)

#### **Novartis Institute for Biomedical Research**

Development of small molecule therapeutics for SMA

#### **Pediatric Neuromuscular Clinical Research Network (PNCN Network)**

Wendy Chung, M.D. (Columbia University)

Basil Darras, M.D. (Children's Hospital of Boston)

Darryl De Vivo, M.D. (Columbia University)

Richard Finkel, M.D. (Children's Hospital of Pennsylvania)

Rabi Tawil, M.D. (University of Rochester)

#### **PharmOptima**

Pharmacokinetic and pharmacodynamic analyses in mouse models of SMA

SMN biomarker development in blood

## RESEARCH PORTFOLIO



### Active *(continued)*

#### **PsychoGenics, Inc.**

Phenotypic and treatment studies in SMA mice

#### **Roche and PTC Therapeutics, Inc.**

Discovery and development of small molecule activators of SMN gene expression to treat SMA

#### **Lee Rubin, Ph.D.** (Harvard University)

Motor neuron screens to identify an effective therapeutic for SMA

#### **Rules-Based Medicine**

Validation of SMA Biomarkers

Development of a multiplexed immunoassay plasma protein panel for SMA

#### **Seward Rutkove, M.D.** (Beth Israel Deaconess Medical Center)

Measuring disease progression in SMA through the use of electrical impedance myography

#### **Charlotte Sumner, M.D.** (Johns Hopkins University)

Collection and analysis of SMA autopsy tissues

#### **Lee Sweeney, Ph.D.**

Evaluation of muscle function in mild SMA mouse models; impact of potential SMA therapeutics on muscle function

### Completed

#### **Spyros Artavanis-Tsakonas, Ph.D.** (Harvard University)

Invertebrate genetic approaches to identify drug targets for SMA

#### **Assay Designs, Inc.**

Analysis of SMN protein levels by ELISA of PBMC samples *(as part of the Biomarkers for SMA study)*

#### **Gary Bassell, Ph.D.** (Albert Einstein College of Medicine)

The role of SMN in axonal localization of mRNP complexes *(as part of the Axon Working Group grant)*

#### **Gary Bassell, Ph.D.** (Emory University)

Imaging of SMN function in RNA dynamics in motor neurons using lentiviral vectors *(as part of the Axon Working Group grant)*

#### **Giorgio Battaglia, M.D.** (IRCCS Foundation, Neurological Institute “Carlo Besta”)

Identification and functional characterization of Axonal SMN protein (a-SMN)

#### **Christine Beattie, Ph.D.** (The Ohio State University)

Using zebrafish motor axons as an assay for SMN function *(as part of the Axon Working Group grant)*

A drug screen for SMA in zebrafish

#### **BG Medicine, Inc.**

Proteomic and metabolomic analyses of plasma and urine samples *(as part of the Biomarkers for SMA study)*

Plasma biomarkers of drug efficacy in SMA mice

#### **Steven Burden, Ph.D.** (New York University School of Medicine)

Examining the autonomous role of SMN in muscle development

## RESEARCH PORTFOLIO



### Completed *(continued)*

#### **Christine DiDonato, Ph.D.** (Northwestern University)

Pharmacological treatment strategies using neuroprotective agents in cellular and animal models of SMA (as part of the 2004 Young Investigator Awards co-sponsored with the American Academy of Neurology)

#### **Enzo Life Sciences Inc. (formerly Assay Designs, Inc.)**

Development of a quantitative enzyme-linked immunosorbent assay (ELISA) and antibodies for detection and measurement of SMN protein

#### **Expression Analysis, Inc.**

Gene expression profiling of blood samples study using the Affymetrix GeneChip® exon array platform and determination of SMN1 and SMN2 copy number from blood samples (as part of the Biomarkers for SMA study)

#### **Cold Spring Harbor Laboratory** – Banbury Meetings on SMA

#### **CombinatoRx, Inc.**

Application of combination high-throughput screening to discover drug combinations that upregulate SMN protein levels

#### **Curis, Inc.**

Development of motor neuron assays to screen for drug candidates for the treatment of SMA

#### **Families of SMA**

##### **Frimorfo**

Histopathological analysis of Delta7 mouse model

#### **Hans Goebel, M.D.** (Johannes Gutenberg University)

SMA muscle biopsy studies

#### **Tessa Gordon, Ph.D.** (University of Alberta at Edmonton)

Studies of the mechanisms of progressive muscle weakness in SMA

#### **Zhigang He, Ph.D.** (Children's Hospital of Boston)

Assessment of possible involvement of axon regeneration and sprouting defects in the pathology of SMA

#### **International SMA Patient Registry** (Indiana University)

#### **Adrian Krainer, Ph.D.** (Cold Spring Harbor Laboratory)

Directed method to correct splicing of SMN2

#### **Christian Lorson, Ph.D.** (University of Missouri)

Generation of a SMN read-through mouse

#### **Lotus Separations, LLC**

Chiral purification of R-trichostatin A

#### **Gordon Lutz, Ph.D.** (Drexel University)

Carrier-oligonucleotide therapeutics for SMA

#### **Alexander Mackenzie, M.D., Ph.D.** (Children's Hospital of Eastern Ontario)

Screening of HDAC inhibitors for SMN2 upregulation

#### **MethylGene, Inc.**

Evaluation of isoform-selective HDAC modulators as potential SMA therapeutics

## RESEARCH PORTFOLIO



### Completed *(continued)*

#### **Umrao Monani, Ph.D.** (Columbia University)

The role of SMN in motorneuron development (*as part of the 2004 Young Investigator Awards co-sponsored with the American Academy of Neurology*)

SMA - translating basic findings into a potential treatment (*as part of the Motor Neuron Center grant*)

#### **New England Research Institutes (NERI)**

Trial management and data coordination (*as part of the Biomarkers for SMA study*)

#### **Livio Pellizzoni, Ph.D.** (Columbia University)

Dissecting SMN interactions and RNP assembly activity in motor neurons (*as part of the Axon Working Group grant*)

Neuronal RNA metabolism and the molecular basis of SMA (*as part of the Motor Neuron Center grant*)

#### **Regeneron Pharmaceuticals**

Generating mouse models for SMA

#### **Mustafa Sahin, M.D., Ph.D.** (Children's Hospital of Boston)

The role of SMN in axon outgrowth and targeting (*as part of the 2004 Young Investigator Awards co-sponsored with the American Academy of Neurology*)

#### **Anneliese Schaefer, Ph.D.** (Washington University)

In vivo imaging of motor axons and neuromuscular junctions in a mouse model of SMA

#### **Michael Sendtner, M.D.** (University of Wuerzburg)

Characterization of the role of the SMN protein and its binding partners for axon growth, differentiation and function of motoneurons

#### **Shanghai Husker Chemical**

Chemical synthesis of trichostatin A

#### **Jeremy Shefner, M.D., Ph.D.** (The Research Foundation of the State University of New York)

Motor unit number estimation in SMA mice

#### **Brent Stockwell, Ph.D.** (Columbia University; The Whitehead Institute for Biomedical Research)

A high-throughput cytoblast assay for monitoring SMN protein in concentration in mammalian cells and high-throughput screen for compounds that increase SMN protein level

#### **Kathryn Swoboda, M.D.** (University of Utah School of Medicine)

Refinement of outcome measures for clinical trials in infants and children with SMA (*as part of the 2004 Young Investigator Awards co-sponsored with the American Academy of Neurology*)

#### **Wesley Thompson, Ph.D.** (University of Texas at Austin)

The role of muscle fibers in the mouse model of type II SMA

#### **Eduardo Tizzano, M.D., Ph.D.** (Genoma España)

Defining targets for therapeutics in SMA

#### **Marcel van den Heuvel, Ph.D.** (University of Oxford)

The role of SMN in RNA metabolism

#### **Ching Wang, M.D., Ph.D.** (Stanford University School of Medicine)

Pilot clinical trial of hydroxyurea on SMA patients

#### **Brunhilde Wirth, Ph.D.** (University of Cologne)

Search for SMA modifying genes