



Families of SMA *Research. Support. Hope.*

COMPASS

A Publication Dedicated To Research Updates

Families of SMA Awards Clinical Care Research Grants.

In this issue of "Compass" Families of SMA (FSMA) reviews four new Spinal Muscular Atrophy (SMA) patient care research grants. These projects were selected from applications received in response to our Clinical Care Research Project Request for Proposals (RFP).

Overview of the Clinical Care Research Program:

Since 1984 FSMA has been funding critical research to develop a treatment and cure for the disease, along with providing important resources and support for families affected by SMA. In 2013 we launched our Clinical Care Research Program to fund care research and drive improvements in patient care of SMA. The goal of this new program is to improve patient care and living with SMA.

Funding for this program will build upon the Consensus Statement for the Standard Care of SMA with data driven results on specific areas of SMA care. The results of these funded projects will help build an evidence base and demonstrate measurable, positive effects on the clinical management and lives of SMA patients. Key project information will then be used to provide the following:

- Educational programs for professional medical providers, such as the FSMA Continuing Medical Education Conference;
- New family-focused care publications, such as the FSMA Care Series Booklets; and
- Peer reviewed journal publications to influence insurance coverage.

This initial round of funding was designed to fund pilot studies which will demonstrate the feasibility of data collection, and which will then support submission of a larger study for FSMA or government funding.

Our RFP Solicited Projects on Key Areas in Clinical Care Research:

In order to fund the most critical research, our 2013 RFP solicited pilot studies that would help build an evidence base and demonstrate quantifiable, positive effects on the clinical management and lives of patients with SMA. These studies were in the following key areas:

- Respiratory interventions and breathing issues.
- Nutritional interventions and the impact of diet.
- Spinal deformity and surgical interventions.
- Psychological interventions to address mental health issues facing SMA patients and care givers, quality of life and the impact of the disease on the family.
- Studies to evaluate the impact of standing.
- Studies to evaluate the impact of medical devices.
- Studies that address the gaps in the delivery of care.

FSMA Clinical Care Research Grants are Selected by Our Medical Advisory Council:

Our Clinical Care Research Program is overseen by the FSMA Medical Advisory Council (MAC), to ensure that we fund important care research on SMA. The MAC is comprised of 16 highly recommended, leading SMA medical and clinical experts.

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Announcements!

New 2014 Clinical Care Research Funding

In addition to the funding announced in this edition of *Compass*, FSMA will invest in future care research in 2014. The new Clinical Care Research RFP will be available soon and announced on the FSMA website.

New Care Series Booklet

A new Orthopedic Care Series Booklet will be coming soon! We will announce when this is published on www.curesma.org. This is a great patient care resource.

FSMA has the following Care Series Booklets (also available in Spanish):

- Caring Choices
- The Genetics of Spinal Muscular Atrophy
- The Family Guide to SMA Research
- Breathing Basics
- Families of SMA Family Support & Patient Services
- Nutrition Basics
- Understanding Spinal Muscular Atrophy
- The Family Guide to SMA Research



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The MAC sets the agenda for proactive, creative, collaborative leadership on issues to improve the quality of care for SMA. The MAC carefully reviews all of the Clinical Care Grant applications to make certain that they have a strong scientific merit with SMA relevance. All projects are considered on the scientific quality and relevance to the FSMA research mission of improving care for all SMA patients.

Importance of Our Clinical Care Research Grants:

The first project by **Dr. Mayer and Ms. Battista, RN, MS, CPNP, CCRC at the Children's Hospital of Philadelphia** will fund a pilot study to assess the decision making process by parents of children with SMA on clinical care options. This work could lead to the most valuable data set on perceptions of parents with SMA Type I and SMA Type II children, and could provide the basis for a larger study. The second project by **Dr. Swoboda at the University of Utah** will fund a study to determine whether children/preadolescents with SMA Type II demonstrate impaired glucose tolerance after glucose loading and to determine whether these same children demonstrate intolerance of fasting. If the study confirms glucose metabolism abnormalities in children with SMA further study will be needed to develop appropriate management protocols and care guidelines. This pilot is needed to further understand glucose, insulin, and other responses to fasting and glucose loads in people with SMA. The third project by **Dr. Halanski at the University of Wisconsin** will fund a study to create a database of patients with SMA from multiple clinical centers. It will be used to compare outcomes across sites and to generally improve patient care. This work could help facilitate clinical management across clinical centers and establish evidence-based best practices for clinical management. The fourth project by **Dr. Lotze at Texas Children's Hospital at the Baylor College of Medicine** will fund a project to enable health care providers to take better care of patients with SMA Type I in emergency centers and hospital settings. This work could allow quality improvement care for the hospital care of SMA Type I children driven by collaborative efforts of the healthcare providers and families to educate primary care providers.

Thank you for supporting these important Clinical Care Research projects.

Descriptions of Newly Funded Clinical Care Research Grants



Decisions Related to Goals and Limitations of Care and the Challenges in Making Them for Parents of Children with SMA

Oscar Mayer, MD, PhD and Vanessa Battista, RN, MS, CPNP, CCRC



Children's Hospital of Philadelphia

Objective: The aim of the proposed study is to assess the possibility of conducting research with parents of children with SMA, as has been done in populations of parents of children with other serious illness, and to assess parents' decision-making processes, perceptions about parenting, and levels of depression and anxiety as changes in their children's medical status occur.

Research Strategy: We propose to conduct a study involving 50 parents of children (<18 years of age) with SMA Type 1 or SMA Type 2, by conducting interviews at the start of the study and again three months later.

Significance of the Project: To our knowledge, this proposed study will provide the richest data set available involving perspectives of parents' of children with SMA Type 1 and SMA Type 2, and will thereby provide the basis for a larger study and ultimately the design of a supportive care intervention trial.



Pilot Study of Glucose Load Tolerance and Fasting in SMA Type II

Kathryn Swoboda, MD
The University of Utah

Objective: The aims of this pilot study are to: determine whether children/preadolescents with SMA Type II demonstrate glucose tolerance after glucose loading and to determine whether these same children demonstrate intolerance of fasting in a well state.

Research Strategy: Participants with SMA Type II will be given a glucose drink. Blood sugar, insulin, and other markers will be measured up to 3 hours after to measure the body's response to the drink. In a separate visit, the same participants will fast under medical supervision and similar markers will be monitored.

Significance of the Project: This pilot is needed to further understand glucose, insulin, and other responses to fasting and glucose loads in people with SMA. This information can be used to guide fasting limitation protocols and intake guidelines for patients with SMA and may also serve as preliminary information for glucose loading and tolerance in SMA.



Spinal Muscular Atrophy: A Multicenter Multidisciplinary Assessment

Matthew Halanski, MD

The University of Wisconsin

Objective: To create a database of patients with SMA from multiple clinical centers focused on 3 clinical care areas: breathing support, nutrition, and spine care. The database will be used to compare patient outcomes with the goal to improve the standard of care for individuals with SMA.

Research Strategy: To enroll >150 patients from the University of Wisconsin plus patients from 5 other care centers using an electronic database called REDCap and the common data elements identified by the NINDS. The data will be compared and analyzed across 3 clinical care areas to identify associations with care management and outcomes such as decreased hospitalizations and survival.

Significance of the Project: This project will facilitate the comparison of clinical management across clinical centers around the country to better establish evidence-based best practice clinical management strategies.



Quality Improvement Project to Reduce Gaps in Care in the Hospital Setting for Children with SMA Type I

Timothy Lotze, MD

Texas Children's Hospital Baylor College of Medicine

Objective: The objective of this study is to identify gaps in care for children with SMA Type I while they are in the emergency center, hospital or with their primary care provider, and improve the standard of care provided.

Research Strategy: Parents will be invited to participate in a focus group in either English or Spanish to discuss their family's experience in the emergency center, hospital and with primary care. Healthcare providers will be asked via survey to (anonymously) identify deficits in their knowledge related to SMA Type I and to share their perceptions about barriers to care for children with this disease. The information from these groups will be used by the investigators to develop interventions to improve healthcare. Interventions will include: 1) Education about SMA Type I to a broad spectrum of hospital providers, 2) Development of a medical template that parents can carry with them to the emergency center / hospital that is individualized and addresses urgent care concerns related to the child with SMA Type I, and 3) An electronic medical record of the same medical template.

Significance of the Project: This project is significant because it allows quality improvement for the hospital care of children with SMA Type I to be driven by the collaborative efforts of both families and healthcare providers and includes education to primary care providers. It also provides information to create a hard copy medical template (tailored to each child) that can serve as a practice alert or emergency card to alert providers who are not as familiar with SMA Type I, to immediate disease concerns.

The 2014 Continuing Medical Education (CME) Conference Held on June 13 at the Gaylord National Harbor Hotel in Washington, D.C.

This one day CME Conference for medical professionals is a great step toward broadening our support programs by educating medical providers. Families of SMA partners with the University of Wisconsin School of Medicine and Public Health, who are accredited to provide CME credit for medical professionals.

Upon completion of this course medical professionals will learn how to:

- Identify when to refer children with delayed motor milestones for further evaluation.
- How to apply best practices to the coordinated care of individuals with Spinal Muscular Atrophy.
- Understand and promote care coordination between the community and specialty care for children with Spinal Muscular Atrophy.

Target audience: Physicians, nurses, nurse practitioners, physician assistants, physical therapists, occupational therapists, nutritionists, respiratory therapists, speech therapists, allied health, residents, fellows, and other health care professionals.



The 2014 Family Medical Workshops at SMA Conference

- Breathing Basics and Care Choices for SMA Type I
- Breathing Basics and Care Choices for SMA Type II & SMA Type III
- Genetics and Reproductive Options for SMA Families
- Hands on Physical Therapy
- Medical Management For Adults with SMA
- Good Nutrition Principles and Round Table for Oral Feeders
- Nutrition for G-tube Feeders
- Orthopedic Management
- Aquatic Physical Therapy for Fun and Function
- Recognizing and Managing Pain in SMA
- Ethics in Research

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