



# DIRECTIONS

FAMILIES OF SPINAL MUSCULAR ATROPHY



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Nora Gooden on her sheepskin blanket, in her Radio Flyer Wagon, with her FSMA Care Package.

We are highlighting some amazing progress on SMA research in this issue of Directions, including breaking news on all three of the leading SMA therapeutic development programs that Families of SMA funds:

- Licensing of the Quinazoline program to an industrial partner, and the first ever Orphan Drug Designation for a new therapy specifically designed for SMA. See page 7;
- Completion of the preIND meeting with the FDA for the motor neuron replacement program. See page 17; and
- A multi-million dollar award from the NINDS to Paratek Pharmaceuticals and FSMA to advance a drug candidate for SMA. See page 22.

This edition of our community newsletter is focused though on the patient services and family support that FSMA provides. This is an increasingly important area to our organization and community. FSMA now provides services and support to 70% of all newly diagnosed families each year! You can read reviews of the highly important Annual SMA Conference, and details of the plans for the 2010 meeting in California, throughout this newsletter.

We will continue to improve services to support all newly diagnosed SMA families, and add new programs to assist all patients affected by SMA as they navigate through life.

Thank you for all your support and dedication to Families of SMA and the goals we are trying to accomplish.

Sincerely,



**Kenneth Hobby**  
President, Families of SMA

This year we are celebrating FSMA's 25th anniversary. The Board of Directors took this milestone as an opportunity to look to the future and see what opportunities lay ahead. Now with an updated mission, Families of SMA has a clear map for the future and how we need to enhance both the research and support programs that we provide to our community.

We have moved forward aggressively to implement new programs that Families of SMA will need to provide in the coming years. These new initiatives include special programs to assist newly diagnosed families, improving and adding to our patient services and family support programs, launching the new Medical Advisory Council and producing new publications under our SMA Care Series.

We have been here for 25 years because of the strength of all our families! We know we are making significant strides in developing a treatment and a cure for SMA, and we have accomplished this by working and supporting each other. Thank you for all your hard work and continued support of FSMA!

Sincerely,



**Paula Barrett**  
Chair, Board of Directors



## Mission Statement

Families of Spinal Muscular Atrophy is dedicated to creating a treatment and cure by:

- Funding and advancing a comprehensive research program;
- Supporting SMA families through networking, information and services;
- Improving care for all SMA patients;
- Educating health professionals and the public about SMA;
- Enlisting government support for SMA;
- *Embracing all touched by SMA in a caring community.*

**Our vision is a world where Spinal Muscular Atrophy is treatable and curable.**

## Families of SMA Patient Services and Family Support.

The FSMA services and support programs assist patients and families affected by SMA at all stages of the disease: from first diagnosis; to managing the disease; to improving daily life with SMA.

We are here to help all SMA patients and families with:

- understandable information and emotional support.
- resources to live a healthy and comfortable life with SMA.
- connections with other families.

## New Items Available in the Equipment Pool

The FSMA equipment pool includes many items suitable for SMA children such as bath chairs, car seats, car beds, strollers and manual wheelchairs.

### New Items Recently Added to the Equipment Pool:

- New bath chairs.
- Car seats for Type II children.
- Therapy feeding chairs.

This service is available free of charge to all members of Families of SMA. E-mail [equipment@fsma.org](mailto:equipment@fsma.org) for more information.

## Information Packets for Newly Diagnosed Families

Every time that a newly diagnosed family contacts Families of SMA, we express mail them a packet of information. These packets include up to date medical care information such as copies of the Care Series Booklet Breathing Basics.



The packets also include general information on understanding the disease with background on SMA genetics and the current state of research. Information is provided that is relevant to the specific Type of SMA that the patient has been diagnosed with.

Last year, Families of SMA mailed over 600 of these informational packets to newly diagnosed families and their relatives.

**Cindy Bobolz, Grandmother of Nora Gooden, has been busy making beautiful quilts to send in the care packages.**





# Families of SMA Newly Diagnosed Type I Care Packages

**A**di and Shaina Rappoport and the Jacob Isaac Rappoport Foundation recently awarded Families of SMA a \$15,000 grant to fund the Type I care packages for the next year. These care packages are sent to every new family with a Type I diagnosis that contacts Families of SMA.

These care packages were originally created by Donna and Alex Abraldes in memory of their daughter Deirdre. The Abraldes family started making these care packages in 2001. In 8 years, they created over 450 care packages for newly diagnosed families.

In 2007, Mary Jane and Tim Utzat began making care packages. They would make 5 – 10 care packages at a time and ship them to the Families of SMA National Office to send to families the day that they make first contact. These packages included a hand knit blanket and rattle that Tim’s mother, Marianne Utzat, had made.

With the help of the grant from the Jacob Isaac Rappoport Foundation, we have been able to add many new helpful items for newly diagnosed Type I families. Some of the items in the care

packages now include:

- Fisher Price outdoor swing
- Sesame Street Sing Along DVD
- Several toys for an SMA infant
- Lightweight balloons on a stick
- Small pinwheel on a stick
- Feathers
- Wind chimes
- Books
- Pacifiers
- Plaster hand print molds
- Several links
- Finger puppets
- An infant bath pad
- Sheepskin blanket
- Fisher Price Ocean Wonders Projector
- Motorized bubble wand

Many of the items include a label with a few sentences explaining the item, summarized by Mary Jane and Tim Utzat. They give other toy ideas as well as include several pictures of their daughter Samantha using some of the toys.

The packages include books donated by Donna Taylor (in memory of her daughter Hannah Jean Campbell), and Jessica Picinich (in memory of her daughter Loren), as well as sheepskin blankets funded by The CAM Fund and the Munson family. Cindy Bobolz, Grandmother of Nora Gooden, has also been busy making beautiful quilts to send in these care packages.

Families of SMA currently sends over 200 Type I care packages each year to newly diagnosed families.



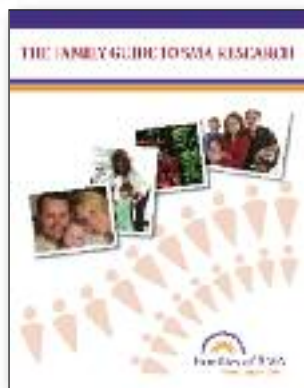
**Hannah Willingham with a feather from the care package.**



**Nora Gooden**

Families can also receive a Radio Flyer Wagon from Families of SMA through the Tumbleweed Wagon Fund. The wagons are beneficial for transporting type I infants around the house, yard, doctors’ offices, hospitals, stores, etc. The babies love to be pulled around in the wagons and may be more comfortable to be moved from one location to another in them as opposed to being picked up and carried frequently for feedings, diaper changes, etc. Many families also add attachments to the wagon to hang toys, tie balloons, or hang bolus or feeding bags. The sides of the wagon can be removed so the babies can lie in them to nap. Many families line their wagon with the Sheepskin Blanket, also sent by FSMA.



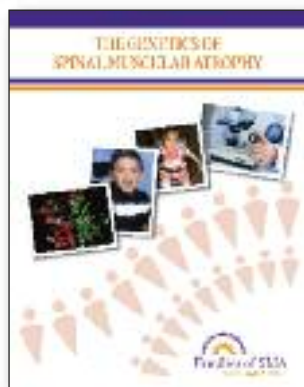


### Family Guide to Research

This guide helps to answer questions such as what are the key areas of SMA research and what are the costs for conducting SMA drug development and clinical trials. Clear definitions and graphics are included to help explain and illustrate how SMA drugs begin and the key steps involved in developing new therapies for Spinal Muscular Atrophy.

The topics in this new booklet cover the following important areas of SMA research:

- SMA Researchers.
- Drug Research.
- Gene Therapy.
- Stem Cells.
- Clinical Trials.
- Government Research and The FDA.



### The Genetics of Spinal Muscular Atrophy

Confused about genes, proteins, DNA, and how SMA is diagnosed?

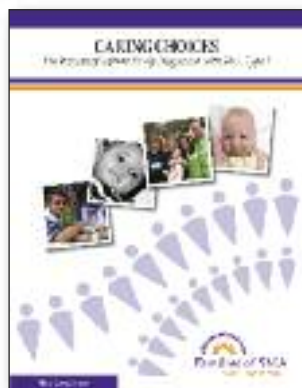
Read this helpful pamphlet. It includes definitions, explanations, and diagrams from genetics expert, Louise Simard, Ph.D. and the FSMA Medical Advisory Council.



### Patient Services & Family Support

This booklet has details on the following Families of SMA programs:

- 1) Programs For Newly Diagnosed Families:
  - Including our special Type I programs such as: Care Packages; Sheep Skin Blankets; Radio Flyer Wagons; Dinners; and Home Support.
- 2) Families of SMA Equipment Pool.
- 3) Medical Care.
  - Including the "Ask the Expert" Service.
- 4) Daily Living.
- 5) Local Support.
- 6) How to Keep Up to Date.
- 7) The Annual SMA Conference:
  - Including the FSMA Newly Diagnosed Conference Program.



### Caring Choices

This booklet is focused on caring choices for parents of infants newly diagnosed with Spinal Muscular Atrophy Type I.

The topics review the basics of the main care options for newly diagnosed SMA Type I:

- What is Non-Invasive Respiratory Care.
- What is Invasive Respiratory Care.
- What is Palliative Care.

And, where you can go for support and guidance.



### Breathing Basics

This new booklet is focused on the critical aspects of respiratory care for children with Spinal Muscular Atrophy. The booklet was authored by Mary Schroth, M.D., a member of the Families of SMA Medical Advisory Council, and a leading expert on respiratory care for SMA patients.

This booklet reviews the following important topics:

- Why is respiratory care so important in SMA.
- What are common respiratory problems in children with SMA.
- Elements of respiratory care management in SMA:
- What are special needs of children with SMA Type I.
- What are special needs of children with SMA Type II.
- What are special needs of children with SMA Type III.
- What respiratory equipment will you need at home.

If you would like a hard copy mailed to you please email us at [info@fsma.org](mailto:info@fsma.org) or call (800) 886-1762.

## 2010 Annual SMA Conference June 24th – 27th. Santa Clara, California

We are excited to announce that the 2010 Annual SMA Conference will be held in Northern California!

The Annual SMA Conference will be in Santa Clara, CA from June 24th to June 27th, 2010 at the Santa Clara Marriott. Santa Clara is located just 45 minutes from San Francisco. We will be offering family attendees an amazing room rate of just \$99 per night, along with free parking!

The hotel is just 4 miles from the San Jose airport, and just 30 miles from both the San Francisco and Oakland airports, and has an incredible pool for families to enjoy.

This year, we will be rolling out exciting new focused workshop tracks including for newly diagnosed families, repeat attendees, and adults with SMA. We are expecting a record attendance of over 1,000 attendees next year. As always, the Family and the Research conferences will be running alongside each other.

Registration for the 2010 conference, as well as for the hotel, is now available on our website at [www.curesma.org](http://www.curesma.org).

Please call the Families of SMA National Office at (800) 886-1762 or email [info@fsma.org](mailto:info@fsma.org) if you have any questions about the conference.

Since 1988, Families of SMA has hosted an annual conference so that families can hear about the latest research, gain an understanding of the genetics behind the disease, share the latest in disease management techniques and



therapies, and network with other families and professionals.

For the last 14 years this conference has been held in conjunction with the

annual International SMA Research Group Meeting (sponsored by FSMA). This gives the opportunity for families to actually meet the researchers who are looking for a treatment and

cure for SMA, as well as with some of the most outstanding doctors and clinicians in the field of SMA.

The conference provides many opportunities to interact and get first hand updates from the leading SMA researchers and clinicians from around the world who are working to develop a treatment and cure for SMA.

This is the largest conference in the world for those affected by SMA, and also for those involved in providing support and care for SMA patients. There is no other program like this for those affected by SMA in the world. The aspect of interactions between the researchers and families at one conference is extremely special. The annual conference also provides our children an opportunity to make new friends and have a great time.

The weekend will be filled with networking opportunities with other families, medical and care workshops, a memorable kids program, a family and professionals banquet, and a kids carnival.

## Newly Diagnosed Conference Program

Families of SMA will be again offering an amazing support program for all newly diagnosed SMA families to attend the annual conference.

Through generous donations and sponsorships, Families of Spinal Muscular Atrophy will cover the registration fees for the SMA Conference for all families newly diagnosed since the previous meeting in 2009. Our goal is to allow as many newly diagnosed SMA families as possible to attend and experience the benefits of this amazing conference.

The meeting gives families the opportunity to gather critical care and daily living information early after diagnosis, learn directly from experienced SMA physicians, and network with other families.

### Details of the Program:

- Families of SMA will cover the conference registration fees for any newly diagnosed SMA family for up to 4 immediate family members.
- To apply for this new program please email [info@fsma.org](mailto:info@fsma.org) or call (800) 886-1762 and mention “Newly Diagnosed Conference Program”.

(We also provide a limited number of financial need based scholarships to families.)



## FAMILIES OF SPINAL MUSCULAR ATROPHY LICENSES POTENTIAL TREATMENT FOR SMA TO REPLIGEN CORPORATION

ELK GROVE VILLAGE, IL (October 22, 2009) – Families of Spinal Muscular Atrophy (FSMA) announced today that it has entered into a groundbreaking exclusive license agreement with Repligen Corporation for the development of a potential treatment of Spinal Muscular Atrophy (SMA).

Families of SMA was started by a small group of parents in 1984 who wanted to raise funds for SMA research and support all affected families. FSMA has funded \$50 million for SMA research, raised from individual donations and through fundraising events held by volunteer families and Chapters.

FSMA made investments of \$13 million during the last decade to bring this specific program to the cusp of clinical development. Through FSMA's leadership, the research has resulted in a drug candidate that treats the underlying cause of SMA. In preclinical studies, the drug has been shown to efficiently cross the blood brain barrier - a critical feature for a neurological drug - and prolong survival significantly in two different mouse models of SMA.

Often an "Orphan Disease," such as SMA, will not be "adopted" by the pharmaceutical industry because there is limited financial incentive to make new medications to treat smaller patient populations. This license agreement marks a significant milestone for the SMA community by securing the commitment of an industrial partner to develop potential

treatments through the highly expensive human clinical trial phases.

"At this point in the program, joining forces with a corporate partner to advance into clinical studies is the best way to meet our objective of accelerating drug development for SMA," said Kenneth Hobby, President of Families of SMA. "Repligen is an ideal partner for this program with the necessary resources and expertise to invest in and focus on successfully developing this new treatment for SMA. We are very excited to partner with Repligen and look forward to making our combined efforts successful in delivering an effective treatment to our patients."

"Families of SMA has made remarkable progress in defining a series of highly potent compounds which may be clinical candidates for SMA," stated Walter C. Herlihy, President and Chief Executive Officer of Repligen Corporation. "We look forward to working with FSMA and their collaborators in the development of what we hope will be an important new treatment for SMA."

Through the agreement with Repligen, Families of SMA has the potential to recover its investments through a series of milestone payments if the program successfully progresses through clinical stages and eventually reaches market approval.

## Families of Spinal Muscular Atrophy Receives FDA Orphan Drug Designation For Quinazoline495 For The Treatment of Spinal Muscular Atrophy

August 28, 2009.

Families of Spinal Muscular Atrophy announced today that the Office of Orphan Products Development of the Food and Drug Administration (FDA) has granted Orphan Drug Designation to Quinazoline495 for the treatment of Spinal Muscular Atrophy.

This is the first time a new therapy specifically designed for Spinal Muscular Atrophy has ever reached the important stage of being awarded orphan drug status by the FDA. Reaching this key milestone for the first time is a significant step forward for the entire SMA community and signifies the rapid progress being made to develop an effective treatment for this terrible disease.

The US Orphan Drug Act is intended to assist and encourage the development of safe and effective therapies for the treatment of rare diseases and disorders. In addition to providing a seven-year term of market exclusivity upon final FDA approval, orphan drug designation also provides advantages through a wide range of financial and regulatory benefits.

"We are extremely pleased that the FDA has awarded orphan drug status to this promising drug for the treatment of SMA", said FSMA Research Director Jill Jarecki, Ph.D. "Orphan Designation will allow us to utilize all the opportunities provided by the Orphan Drug Act, including working closely with the FDA Office of Orphan Products Development throughout clinical development."



**3D Structural Model of a Quinazoline Compound Bound to the Human Enzyme DcpS, from Singh et al 2008.**

## The 25th Anniversary SMA Conference.

Hyatt Regency Cincinnati • Thursday, June 18 – Sunday June 21, 2009.

The 25th Anniversary Spinal Muscular Atrophy Conference in Cincinnati was a fantastic success as Families of SMA marked 25 years of research progress and service to all those affected by SMA. We had over 900 families and researchers in attendance this year - a record number for the annual SMA conference.

The Ohio – Kentucky – Indiana Chapter of Families of SMA, which is headquartered in Cincinnati, hosted the conference this year. Approximately \$300,000 is invested by Families of SMA every year to carry out the event, the largest meeting in the world for the SMA community.





There were 30 workshops offered during the conference covering the latest information on critical topics in SMA care including:

Respiratory Care for SMA Patients; SMA Physical Therapy and Occupational Therapy; Transition to Adult Care for SMA Patients; Nutritional Care for SMA Patients; Palliative Care; Orthopedic Management for SMA Patients; Tracheotomy.

Also provided were important sessions and demonstrations pertaining to daily living topics including:

Toy Adaptation; Yoga Therapy for SMA; Adaptive Sports; Aquatic Therapy for SMA; Computer Adaptations; Coping as a Family; Grieving.

#### Highlights of this years conference included:

- Keynote speeches given at the main Conference Banquet from:

Paula Barrett, Chair of the Board of Directors of Families of SMA.

Dr. Jill Jarecki. Research Director of Families of SMA.  
The Honorable Steve L. Driehaus. US House of Representatives.

Dr. Hans Keirstead. World famous stem cell scientist and Co-Director of the Sue and Bill Gross Stem Cell Research Center and Associate Professor of Anatomy and Neurobiology at the University of California Irvine.

Dr. Mary Schroth. Leading pediatric pulmonologist at the University of Wisconsin American Family Children's Hospital. She also chairs the FSMA Medical Advisory Council and is a researcher with Project Cure SMA.

- HBO filmed a documentary during the conference.
- The latest Developments on three drug discovery programs funded by Families of Spinal Muscular Atrophy were announced at the Conference.
  - 1) Quinazolines to boost SMN2 expression;
  - 2) Tetracyclines at Paratek Pharmaceuticals to correct SMN2 splicing;
  - 3) Motor neuron replacement program at California Stem Cell and UCI.

These three programs represent an investment of over \$16 Million so far to build a SMA drug pipeline. You can read about these programs in the pull out edition of Compass in this newsletter.



### Goals of the SMA Conference:

1. To allow networking between researchers and families and patients;
2. To educate researchers on the latest research advancements;
3. To attract the best researchers to the SMA field and encourage collaborations;
4. To educate patients and families on the basic facts of SMA;
5. To update families and patients on the latest medical care and research progress;



At the 25th anniversary FSMA conference in Cincinnati, OH this year, a generous donor bought a Standing Dani and donated it to FSMA to raffle off as a fundraiser. Shaina and Adi Rappoport drew the winning ticket from their \$1000 donation ticket entry and then graciously donated the \$16,000 Standing Dani to Madison Smith who is 2 yrs old and has SMA type 2. Jennifer Smith said "Madison and our family were overwhelmed with joy when they presented it to us, as we have been wanting a Standing Dani for 2 years and could not afford one. We are so blessed to know the Rappoport and be the recipient of this generous gift. They are wonderful people and do wonderful things for Families of SMA."

## A Very Special Thank You

**Barbara Trainor –**  
*Conference Coordinator.*

While we are celebrating 25 years of progress in the SMA research field, it is also a perfect time to look at the critical part that the annual FSMA conference plays in our community.



Through the tireless efforts every year of Barb Trainor, the conference has grown into an annual highlight for the SMA community. However, it is more than just a friendly get together each year. This is a conference that has a real impact on the lives of families and patients through the many support and medical programs. The meeting has also been a major influence on bringing researchers into the SMA field and establishing successful collaborations.

Barb is the central point of contact for the conference. She organizes all of the speakers, schedules the workshops, and coordinates all the details of our food, audio-visual, and rooming. With no exaggeration, Barb does the job of several people – and amazingly all as a volunteer!

Barb is also the Families of SMA Chapter Coordinator. She spends hundreds of hours of volunteer time guiding families to help them form new chapters, and leading the existing 26 FSMA Chapters to raise incredible funds for research and helping local families. All of this is done on top of her very busy schedule raising her two daughters, running the very successful FSMA Chesapeake Chapter, and being a member of the Families of SMA Board of Directors.

Thank you Barb. You have been directly responsible for so much progress in SMA, and have reached and had a positive effect on thousands of families. *Thank you.*



Many of the presentations which were given during conference general sessions and workshops are now available online. Visit our website to download.



## Results of Patient Services and Family Support Programs:

FSMA has invested over \$2 Million to date in the annual SMA conference which brings families, patients, researchers and medical professionals together. A third of the attendees receive assistance to attend from our financial need scholarship and newly diagnosed conference programs.

We currently answer 40 questions a month for patients and families through our “Ask the Expert” service, and have loaned over 4,000 pieces of medical equipment to SMA families in need through the FSMA Equipment Pool.

Through our newly diagnosed information packets and Type I care packages Families of SMA now provides services and support to 70% of all newly diagnosed SMA families each year!

FSMA reaches 7,000 families every quarter with this Directions newsletter. We also provide information and resources to over 300,000 people each year through the FSMA website.

## Families of SMA is Pleased to Announce its New Medical Advisory Council, the “MAC”

The Families of SMA Medical Advisory Council is one of the most highly respected bodies of SMA medical and clinical experts in the U.S., setting the agenda for proactive, creative, and collaborative leadership on issues that improve the quality of medical care for those affected by SMA.

The new Council will focus on educating families, health care providers, and the public about SMA; expanding SMA standards of care; and translating positive research results into clinical practice.

Dr. Mary Schroth, a leading pediatric pulmonologist will serve as Chair for the new Council. Mary is Associate Professor of Pediatrics and Director of the Pediatric Pulmonary Center Grant at the University of Wisconsin Children’s Hospital.

The MAC is composed of experts in the following fields; Neurology (pediatric and adult); Pediatrics; Pulmonology; Pediatric Orthopedic Surgery; Pediatric Critical Care (NICU specialist); Psychiatry (rehab medicine); Psychology; Genetic Counseling and/or Medical Ethics; Physical Therapy; Occupational Therapy; Diet and Nutrition; Respiratory Therapy; Nursing



## New SMA Care Booklet Now Available

Patient Services and Family Support available from Families of SMA.

The booklet has details on the following Families of SMA programs:

- 1) Programs For Newly Diagnosed Families:
  - Including our special Type I programs such as: Care Packages, Sheep Skin Blankets, Respiratory DVD’s and Radio Flyer Wagons.
- 2) Families of SMA Equipment Pool.
- 3) Medical Care.
  - Including the “Ask the Expert” Service.
- 4) Daily Living.
- 5) Local Support.
- 6) How to Keep Up to Date.
- 7) The Annual SMA Conference:
  - Including the FSMA Newly Diagnosed Conference Program.

If you would like to download a copy please visit our website.

If you would like a hard copy mailed to you please email us at [info@fsma.org](mailto:info@fsma.org) or call (800) 886-1762.

## Highlights:

# The 2009 International Spinal Muscular Atrophy Research Group Meeting

The SMA International Research Group Meeting is the biggest SMA research conference in the world. Families of Spinal Muscular Atrophy organizes the conference, and financially underwrites the meeting by covering all food and logistical expenses and by waiving registration fees and covering travel and lodging costs for all first author presenters.

250 researchers from around the world attended the 13th Annual SMA Research Group Meeting in Cincinnati, Ohio in June. These attendees represented over 60 academic organizations and 20 companies. 95 presentations were given by the leading experts in the SMA research field.

The conference has many tangible benefits for the entire SMA research community:

- provides the only open venue for annual communication among international SMA researchers.
- enables open communication of early unpublished scientific data among researchers - a key component in accelerating the pace of research.
- creates a vital sense of community among SMA researchers, creating a collaborative spirit and resulting in many productive research collaborations that endure well beyond the close of the conference.
- allows cross-disciplinary dialog among basic researchers, clinicians, and industry representatives that is vital in creating effective therapies.
- motivates SMA researchers by allowing for direct interaction with families and patients living with SMA.

The conference is organized into sessions focusing on major unanswered topics in the field. In 2009, these topics were addressed in five distinct sessions. A leading expert in the field moderates each session in order to stimulate meaningful discussion of the presented results.

Below is a Summary of Important Research Findings Presented At The 2009 Conference:

- 1) Human SMA Studies: Population Screening, Outcome Measures, and Clinical Trial Updates. Session Moderated by Drs. Kathy Swoboda and Tom Crawford. This session included discussion of new outcome measures and biomarkers for use in SMA clinical trials. It also provided updates from SMA clinical trials. Dr. Howell from NICHD and who leads the federal committee advising on newborn screening gave an update on the feasibility of newborn screening for SMA. The effectiveness of several new clinical trial outcome measures were reported by the Project Cure SMA clinical trial group, Linda Hynan from Dallas, and Petra Kaufman from Columbia University. In the final talk, Dr. Wang of Stanford University presented clinical trial results from a double blind placebo controlled trial of hydroxyurea in Type I SMA patients
- 2) Regulation of SMN Expression and Splicing with Therapeutic Implications. Session Moderated by Dr. Adrian Krainer. This session addressed the regulation of expression and splicing of the SMN2 gene and how this can be exploited

for therapeutic benefit. Most notably a talk was given by Dr. Charles Cho of the Genomics Institute of the Novartis Research Foundation on large genome-wide screens for proteins and genes that influence SMN2 levels. This work should lead to new “druggable” protein targets for SMA.

- 3) SMN Protein: Functional Domains and Temporal Requirements. Session Moderated by Dr. Louise Simard. During this session two notable talks were given. In the first, Dr. James Allen of Arizona State University reported on his work to determine the three dimensional structure of the SMN protein. This will be invaluable data for the SMA community, allowing directed drug discovery projects to be initiated to modulate SMN. Excitingly, Dr. Burghes gave a talk in this session proving for the first time that post-natal delivery of SMN protein can correct the disease course in SMA mice. This is a very important finding. While not directly applicable to therapy development, his data indicates that the development of SMN enhancing drugs and gene therapy drugs for SMA are valid approaches. His data also suggests early intervention is most effective, highlighting the importance of newborn screening for SMA. His new mouse system will yield many important results in the coming years on when and where SMN is needed to correct SMA.
- 4) SMN Neuronal Specific Functions. Session Moderated by Dr. Arthur Burgher. This session focused on





presentations regarding the role of SMN in motor axons and at the neuromuscular junction. Generally, these talks focused on results generated in a number of SMA animal models, including in mice, zebra fish, worms, and drosophila. These studies help address whether the disease is motor neuron specific in nature and to determine what the function is of SMN in motor neurons involved in disease pathology. An interesting talk was given from the Dr. Kothary's lab at the University of Ottawa describing the role of SMN in motor axons and how this perhaps can be exploited for therapeutic use. Two talks were given on data generated in the Sumner lab at Johns Hopkins University and the Tabaras lab in Spain, describing electrical signaling defects at the neuromuscular junction in SMA mice. These are important signals from the motor neuron telling the muscle to move. Three talks were given on data generated in the Talbot lab at Oxford University, the Dreyfuss lab at U. Penn, and the Pellizzoni lab at Columbia University on changes in mRNA expression of other genes in

the spinal cord cells of SMA mice due to deficient levels of SMN protein. Determining the consequence of these alterations in the expression of other genes could lead to new targets for drug development and to a greater understanding of the molecular deficits in SMA.

- 5) SMA Therapeutic Development. Session Moderated by Dr. Brian Pollok. During this session updates were given on ongoing therapeutic programs for SMA, including small molecules, stem cell therapy, and gene therapy. Genzyme Corporation and the Kaspar lab at OSU gave two promising talks on gene therapy for SMA. One large hurdle for gene therapy for SMA has been finding a way to get SMN protein made in motor neurons, which are on the other side of blood brain barrier. Both groups have shown this is now possible at least in newborn mice with the use of new viral vectors, and it leads to substantial correction of the disease course. A critical next step to show the approach could be viable in humans will be to determine if blood brain barrier crossing and resulting SMN expression is possible

in older and larger animals. An interesting talk was given on a research collaboration between the Krainer lab at Cold Spring Harbor Laboratory and Isis Pharmaceuticals on the utility of anti-sense oligonucleotide (ASO) approaches as a potential SMA therapy. Several talks were given on potential small molecule drugs for SMA. These included a talk about HDAC inhibitors in SMA from the Wirth Lab., a talk regarding the ongoing SMA drug programs at PTC Therapeutics, and a talk focusing on the Families of SMA-funded drug program at Paratek Pharmaceuticals to correct SMN2 splicing. Finally, an update was given on the Families of SMA Quinazoline program, indicating that the clinical lead compound increases survival by 400% in a new mouse model of SMA. The compound has undergone the battery of IND-enabling safety studies required by the FDA, and preparations are now underway for a fall 2009 pre-IND meeting with the FDA to discuss the next steps for the drug program.

# Health Care Reform: A Political and Policy Overview

The current debate on health care reform is arguably the most complex and contentious issue to be considered by Congress in recent memory. In order to comprehend the direction of the debate, it is important to understand the role of the five Congressional committees with jurisdiction over health matters and the differing rules governing debate in the House and Senate. (Please visit the Families of SMA website to read a more detailed description of the health reform debate).

## The Committees of Jurisdiction

There are five committees of jurisdiction in Congress that oversee and create health care policy, two in the Senate and three in the House.

In the Senate, the Finance Committee and the Health, Education, Labor and Pensions (HELP) Committee share jurisdiction over health care; the two committees are separately writing health reform bills. The HELP Committee approved its version in a party-line vote this summer while the Finance Committee is still working on its draft as it tries to forge a bipartisan compromise. Eventually, the two committees' versions will need to be combined into one bill prior to it being considered on the Senate floor, likely sometime in September or October.

In the House, the Energy & Commerce Committee, the Ways & Means Committee, and the Education & Labor Committee share jurisdiction over health care. These committees cooperated in preparing a "Tri-Committee" draft bill that subsequently has been passed by each of the three committees on party-line votes. The bill likely will be considered on the House floor in September.

## Looking Ahead

In general, bipartisanship is more important in the Senate. Due to arcane rules, a simple majority of 218 votes is all that is necessary to approve a bill in the House while a three-fifths

supermajority of 60 votes is needed in the Senate. Currently, Democrats have 256 votes in the House but only 59 seats in the Senate.

Despite its historically large majorities, Democrats are having trouble forging consensus on the health reform bill. It has been difficult to craft a compromise that will attract the votes needed for passage in either chamber.

In the Senate, Democrats need at least some Republican support to move a health reform bill, but no Republican has pledged support at this time. Additionally, some conservative Democrats are wary of the proposals. Senate Democrats may explore other options, such as the use of "reconciliation", an arcane parliamentary procedure that essentially lowers the threshold for passage from 60 votes to 51 votes. However, reconciliation is governed by several parliamentary rules that could make it very difficult for Democrats to enact major components of the health reform measure. Another option being considered by the Democrats is splitting the health reform bills into two or more measures.

In the House, too, passage of health reform is not ensured, despite Democrats holding a large majority. No Republican has been willing to support the House measure and House Democratic Leadership is facing competing threats from conservative Blue Dog Democrats and the liberal Progressive Caucus. With Blue Dogs claiming 52 Members and the Progressive Caucus claiming 81 Members, if either group were to oppose health reform en bloc, they could bring down the entire bill in the House.

The entire process remains very much in flux due to its complexity. Even if a bill can be passed in the House and the Senate, these bills must be reconciled into one measure and a House-Senate compromise must be able to pass both the House and the Senate with their differing political structures and rules. The next several months promise to be exceeding chaotic and interesting.

## Spinal Muscular Atrophy Researcher Named Director of NINDS Office of Clinical Research.

The National Institute of Neurological Disorders and Stroke (NINDS), part of the National Institutes of Health, has named Petra Kaufmann, M.D., M.Sc., as Director of its Office of Clinical Research.

Dr. Kaufmann is among the foremost experts in the design and management of clinical trials for neuromuscular disorders, including spinal muscular atrophy (SMA), amyotrophic lateral sclerosis (ALS), and mitochondrial diseases. In her new role, Dr. Kaufmann will lead the Institute's efforts to increase the effectiveness of clinical studies by addressing issues such as optimal trial design, ethical safe conduct of trials, and challenges in patient enrollment.

"Dr. Kaufmann has experience in all phases of clinical research, from conducting laboratory investigation and studies on disease mechanism to serving in key leadership positions on several major multicenter trials," said Story C. Landis, Ph.D., director of NINDS. "Dr. Kaufman's outstanding skills and expertise will allow us to make the most of the scientific opportunities ahead and to have a significant impact on clinical neuroscience."



## The Senate Appropriations Committee Encourages Increased Federal Support for SMA Research



The Senate Appropriations Committee on July 30, 2009 approved the fiscal year (FY) 2010 Appropriations Bill for the Departments of Labor, Health & Human Services, and Education. The Committee included in its Report accompanying the legislation specific language encouraging the National Institutes of Health (NIH) to continue to make research for SMA a priority. The Committee Report urges the Director of the NIH to establish a SMA “working group” comprised of the three primary NIH Institutes with jurisdiction over SMA research, the National Institute of Neurological Disorders and Stroke (NINDS), the National Institute of Child Health and Human Development (NICHD), and the National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS), to support ongoing research, drug development, and clinical research efforts in SMA. The language further encourages each of the three aforementioned Institutes

to pursue specific research priorities relative to SMA:

- NINDS is encouraged to plan for each successive stage of SMA research, including preclinical testing of multiple compounds and the development of clinical trials infrastructure necessary for a national, coordinated clinical effort.
- NICHD is encouraged to support large-scale pilot studies that facilitate the development of a national newborn screening program for SMA.
- NIAMS is encouraged to take an active role in research that will provide a better understanding of the effect of SMA-linked genetic mutations on muscle and research that could provide therapeutic benefit through actions on muscle.

The Senate Appropriations Committee has included language in its annual Labor, Health & Human Services, and Education Appropriations Committee Report for several continuous years. Families of SMA is grateful for this support and all it has done to expand federal involvement in SMA research and to advance research to identify a treatment or cure for SMA.

*Spinal Muscular Atrophy (SMA) – Given the near-term scientific opportunity for an effective treatment, the Committee encourages the Director to establish a trans-NIH working group on SMA composed of NINDS, NICHD, and NIAMS, as well as other relevant institutes, to ensure ongoing support of SMA research and drug development, including vitally needed support for clinical research efforts in the field. In particular, the Committee encourages the NINDS to plan for each of the successive stages of SMA research, including preclinical testing of multiple compounds and the necessary clinical trials infrastructure that is needed on a national and coordinated level to ensure effective treatment studies; it encourages the NICHD to support large-scale pilot studies that support the development of a national newborn screening program for SMA; and it encourages NIAMS to take an active role in research that would provide a better understanding of the effects of SMA-linked mutations on muscle as well as research that could provide therapeutic benefit through actions on muscle.*

Senate Report 111-66, Departments of Labor, Health & Human Services, and Education, and Related Agencies Appropriations Bill, Fiscal Year 2010.



## Join the FSMA E-List!

With each research breakthrough or other important event on the road to a cure, Families of SMA sends an e-mail to everyone on its e-mail list. If you would like to join our e-list please e-mail [info@fsma.org](mailto:info@fsma.org) (Use “subscribe FSMA” in the subject line.) with the following information: Name, E-mail address, City and state or country of residence.

# SMA Treatment Acceleration Act is Reintroduced in Congress

Legislation designed to enhance federal support for SMA clinical research and spur the identification of a treatment or cure for SMA has been reintroduced in the United States Congress as “**The SMA Treatment Acceleration Act of 2009**”. The new bill has been modified slightly from a similar measure introduced in the previous (110th) Congress to ensure that federal resources dedicated to SMA clinical efforts and infrastructure are maximized. The legislation is supported by Families of SMA, Fight SMA, the Muscular Dystrophy Association, and the SMA Foundation.

Like the previous iteration of the SMA bill, this new measure is sponsored in the U.S. House of Representatives by Congressman Patrick Kennedy (D-RI) and Congressman Eric Cantor (R-VA). Companion legislation introduced in the United States Senate once again is sponsored by Senator Debbie Stabenow (D-MI) and Senator Johnny Isakson (R-GA), who also joined forces in the 110th Congress.

The new version of the SMA legislation builds upon the success of the SMA Treatment Acceleration Act introduced in 2007, which garnered 85 cosponsors in the House and 21 cosponsors in the Senate, including then-Senator Barack Obama. Due to a strong grassroots effort by the SMA community throughout the summer, the new House bill (H.R. 2149) already has 32 cosponsors and the new Senate measure (S. 1158) currently has 9 cosponsors. A complete list of cosponsors is provided below.

## Bill Details

The SMA Treatment Acceleration Act of 2009 is designed to aid the efforts of the investigators, clinicians, and families

who have been striving to find a treatment or cure for SMA. It provides federal funding to complement the efforts of national non-profit organizations like Families of SMA that are investing substantial private funding into SMA research, clinical trials, and drug development. Passage of this landmark legislation will enable investigators to mount national clinical trials to demonstrate that identified therapeutics are safe and effective for SMA patients.

## Changes to the Bill

- We have worked closely with the bill’s sponsors, the congressional committees with jurisdiction over the legislation, and the National Institutes of Health to improve and streamline the legislation prior to its reintroduction.

## Outlook

At present, Congress and the congressional committees with jurisdiction over health care policy are focused solely on the President’s health reform legislation. The effort is so monumental that Members of Congress and congressional staff simply do not have the time or manpower to address any other health-related matters. Consequently, all “other” health-related issues (including the SMA Treatment Acceleration Act of 2009) are on hold, likely through the end of 2009.

By the end of calendar year 2009, work on the health reform bill will be completed. Congressional staff has indicated that health-related bills such as the SMA Treatment Acceleration Act will be addressed at that time.

Looking ahead to 2010, our initial goal is to secure a hearing or a markup of the SMA bill in the two committees of jurisdiction: the House Energy &

Commerce Committee’s Health Subcommittee and the Senate’s Health, Education, Labor and Pensions (HELP) Committee. We have been working closely with staff for the two committees for over a year to try and facilitate a hearing or markup. While we are reasonably well positioned to prove successful in this endeavor, more work is needed, especially in terms of continuing to grow congressional support for the bill.

It is important that families continue to reach out to their Members of Congress and urge them to cosponsor the SMA bill. An active grassroots campaign is crucial to forging a critical mass of congressional support for the bill and moving it through the legislative process towards passage and enactment into law. Legislation rarely moves through Congress without broad demonstrated support from several Members of Congress, and Members of Congress are significantly more willing to cosponsor legislation if their constituents are actively lobbying on the bill’s behalf.

## How to Help

Visit the Families of SMA legislative action website for instructions for how to identify your Members of Congress, tips for contacting them, and a sample letter and talking points. We strongly urge you to take the time to participate in this important grassroots effort and to ask your family, friends, and colleagues to join you.

Please do not hesitate to contact Spencer at [spencer@fsma.org](mailto:spencer@fsma.org) if you have any questions about the bill or contacting your Members. With your assistance, we can continue to build support for the SMA Treatment Acceleration Act of 2009.

## California Stem Cell, Inc. and Families of Spinal Muscular Atrophy Announce Completion of Pre-IND Meeting with FDA for Stem Cell-Derived Therapy for the Treatment of Spinal Muscular Atrophy Type I.

### Significant Progress made Towards Moving Novel Therapy into Human Clinical Trials.

California Stem Cell, Inc. (CSC) and Families of Spinal Muscular Atrophy (FSMA) announced today that they have completed a formal pre-Investigational New Drug (Pre-IND) meeting with the Food and Drug Administration (FDA) for guidance on the clinical and regulatory pathway and requirements for submission of an IND to initiate human trials for a stem cell-derived motor neuron replacement therapy for Spinal Muscular Atrophy (SMA) Type I.

SMA is the leading genetic cause of death of infants. It is a disorder that results from a chronic deficiency in the production of the SMN protein, which is essential to the proper functioning of the motor neurons in the spinal cord. SMA is typically marked by the deterioration of the muscles that control crawling, walking, swallowing and breathing. There are no approved therapies for the treatment of SMA.

Approximately 1 in every 6000 babies born is affected. 1 in 40 people are genetic carriers, indicating approximately 7.5 million carriers in the United States.

CSC, a leading stem cell therapeutics company, has developed a stem cell-derived motor neuron replacement product, for the treatment of SMA Type I. Pre-clinical studies, completed

in collaboration with professor Hans Keirstead of the University of California, Irvine, have shown clinical proof of concept through the demonstration of functional benefit in animal models treated with CSC's motor neuron replacement product, MotorGraft™.

MotorGraft™ has the potential to provide benefit for SMA by two mechanisms: 1) direct replacement of the motor neurons lost during the disease course, resulting in new muscle innervation, and 2) providing a nursing support function to remaining motor neurons.

"We are greatly encouraged by the feedback we received from the FDA" said CSC COO Chris Airriess, "It will greatly assist us in preparation of our final IND application. Completion of this key milestone is an important step towards moving our SMA program to the clinic.

"We are very pleased with the outcome of the Pre-IND interaction with the FDA" said Kenneth Hobby, President FSMA. "FSMA has invested almost \$2 Million over the last 8 years to develop this motor neuron replacement therapy for SMA. This meeting was a significant accomplishment towards the filing of the IND and ultimately bringing this therapy to patients.



Dakin Lovelace

## August is Spinal Muscular Atrophy Awareness Month.

August is National Spinal Muscular Atrophy (SMA) Awareness month. We are making great progress in developing potential treatments and a cure for this disease. This is the time to come together with your communities to raise knowledge of the disease to new heights.

Awareness is the beginning of change. The majority of people do not know about SMA until it directly affects their family. Even the letters SMA don't ring a bell with many doctors, nurses and community members. This is where you can help.

Remember to send us copies and links to any press or proclamations that you may get. And we will include them on the website.



# CHAPTER updates

www.curesma.org

## Western NY Chapter

The Western New York Chapter had the perfect day on the **1st Annual Walk for a Cure**. The weather couldn't have been more beautiful at Beaver Island State Park. It was warm with a bit of a breeze coming from the Niagara River. We had over 400 people enjoying the day and the hot dogs, the clown and the music.

Over \$45,000 was raised from our generous family and friends. Participants came from the South towns and Central NY to enjoy the day and contribute to the basket raffles and 50/50 split. Tim Walker, from National, joined us and talked about the latest accomplishments that are making news. Everyone applauded the news. He then joined in and helped pass out hot dogs and thank you plaques to our corporate sponsors.

On August 22, a comedy night was held at a local club. The night was organized by Diane Blair, our VP. Tickets were sold and a basket auction was held. Great fun and many laughs were had by all that attended. Many thanks to all the families and friends that have helped in our fundraising and awareness raising events.



## Chesapeake Chapter (Maryland/Virginia)

Our summer and fall fundraising season are always quite busy. The Chesapeake chapter held its **16th Annual Crab Feast**. The hall was again filled with over 300 people enjoying Maryland's best crabs! Participants enjoy crabs, buffet of food and a silent auction. The silent auction has been a fun and important addition to the event. Special thanks to Beverly Venedam who coordinated the silent auction. The event raised over \$15,000.

The Chesapeake Chapter hosted their first Wine tasting event. Over 130 guests attended **A Vintage Affair to Uncork a Cure for SMA** at the Castle at Maryvale. Live Jazz by Faux Minx made for an enchanting evening. Riedel Crystal cosponsored to create a unique experience in a Riedel Stemware Seminar and Wine Tasting. All guests went home with their own set of Riedel crystal and opportunity to purchase the wine they were tasting. The event raised over \$16,000.

The beginning of October will mark the 15th annual **CRM Golf Classic** to benefit FSMA at The Woodlands/Diamond Ridge Golf Club. Two courses and 38 foursomes are a wonderful testament to the many companies and individuals who continue to support the work of FSMA and its goal - Advancing Research and Supporting Families.

The chapter welcomes new families to get involved. Please contact Barbara Trainor at 800-762-0113 or email her at [fsma-chesapeake@comcast.net](mailto:fsma-chesapeake@comcast.net)



## Greater NY Chapter

The Greater NY Chapter has recently been expanded to include the five boroughs of NYC, Nassau, Suffolk, Westchester and Rockland Counties. We are very excited to welcome the new families into our chapter.

The Greater NY Chapter was awarded with On Field Spirit Awards from both the New York Mets and the New York Yankees. Chapter members were invited for on field ceremonies from both teams. Many thanks to Carol Laurenzano of the New York Yankees and to Matthew Gulotta from the New York Mets for working so hard to help raise awareness for SMA. Chapter Board Members were also invited to attend a BBQ at Gracie Mansion with NYC Mayor Michael Bloomberg and Commissioner Matthew Sapolin in August to participate in a celebration for the anniversary of the Americans with Disabilities Act.

The Greater NY Chapter is also proud to announce that they have been selected as the Local Charity for the Annual Rockville Centre St. Patricks Day Parade which will be held on Saturday, March 20, 2009.

Also there have been some extraordinary efforts from some of our local families. The Erwin Family raised over \$80,000 in the NYC Half Marathon and the Gaynor Family has raised over \$75,000 on behalf of Sophia's Cure.

There will be a Gala in NY on Thursday, November 12, 2009 at Bridgewater at the South St Seaport. Tickets will be \$150 per person. The event is hosted by Kiley & Dylan's Sweet Dream Foundation and Sophia's Cure. For more information on the Gala please contact Debbie at [greaterny@fsma.org](mailto:greaterny@fsma.org).



Dani at CCK KY Walk



## New England Chapter

On Sat. May 16, 2009, we had our 9th Annual Cure SMA Walk-n-Roll, hosted by Silvia & Brian Murphy. It was held at Wompatuck State Park in Hingham, MA. There were over 700 registered participants who enjoyed having family fun, refreshments and a chance of winning raffle items. With the help of the Farrell family, the Johnson family and the Norton family \$80,000 was raised. Over the last 9 years, the Murphy family has raised over \$500,000 with their walks!

On the weekend of June 13 & June 14, 2009, the Johnson family held their first Hunt For A Cure Charity Disc Golf

Tournament at the Norfolk Hunt Club in Medfield, MA. There was a World Class Frisbee Show & Playshop enjoyed by all ages along with silent auction items, carnival games, lunch and tips from the Pros! This event raised \$11,000!

On Monday, August 3, 2009, we had our 12th Annual Golf Tournament and Barbeque hosted by the Barrett family with help from the Gaudreau family. It was held at Bear Hill Country Club in Stoneham, MA. There was a putting contest, raffle items and a silent auction, helping to raise over \$30,000 this year and over \$400,000 over the last 12 years!



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alabama@fsma.org

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## Northern California Chapter

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## Southern California Chapter

(since 1999)  
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Burbank, CA 91501  
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Rosemary Roope, President—  
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(877) 591-4023 (toll free)  
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## Chesapeake Chapter (since 1995)

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## Michigan Chapter (since 2004)

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Ken Armbrustmacher, President—  
michigan@fsma.org

## Minnesota Chapter (since 1992)

PO Box 32813  
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Wanda Wosika, President—  
minnesota@fsma.org

## FSMA Carolinas— North & South Carolina

More info to come.

## South Jersey/Delaware Chapter

(since 2002)  
PO Box 538, Medford, NJ 08055  
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Jessica Moyer, President—  
southjersey@fsma.org

## New Mexico Chapter (since 2009)

Natasha Abruzzo, President

**NEW**

## Greater New York City Chapter

(since 2006)  
PO Box 322  
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## Western New York (since 2003)

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## Texas Chapter (since 2005)

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**NEW**

**Are you interested in starting a  
new chapter for Families of SMA?**

Please contact our Chapter Coordinator, Barb  
Trainor at chapters@fsma.org

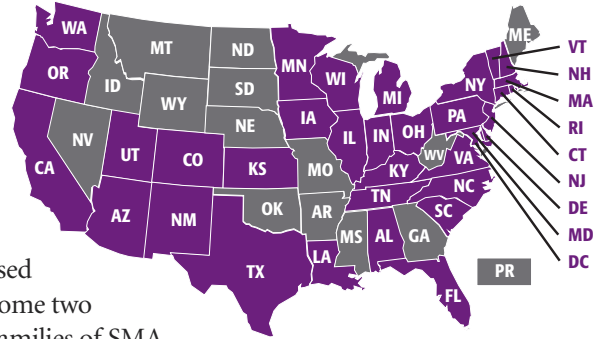


**Utah Chapter** (since 2009)  
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 Shane Barber, President—  
 utah@fsma.org

**Pacific Northwest** (since 2006)  
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 (206) 284-1888  
 Rick Jones, President  
 pacwest@fsma.org

**Wisconsin Chapter** (since 1999)  
 PO Box 320516  
 Franklin, WI 53132  
 (414) 324-0499  
 wisconsin@fsma.org  
 Contact: Kate Vogedes, Laurie King,  
 Jenny Imhoff

## New FSMA Chapters



We are extremely pleased to announce and welcome two new Chapters to the Families of SMA community.

**New FSMA Chapters were recently formed in New Mexico, North Carolina and South Carolina.**

Our Chapters make it their mission to support families and fundraise for SMA, giving hope to families in their community. Chapter fundraisers include Walk n Rolls, golf tournaments, and gala events. Chapter support includes providing resources to families affected by SMA, linking families together for mutual support, and providing public awareness in their communities for a wider awareness of SMA.

*Have you ever thought about starting a chapter?*  
**We want to hear from you.**

CHAPTERS ARE REACHING OUT TO COMMUNITIES ALL ACROSS THE COUNTRY. Families of SMA currently has over 28 chapters in the United States.

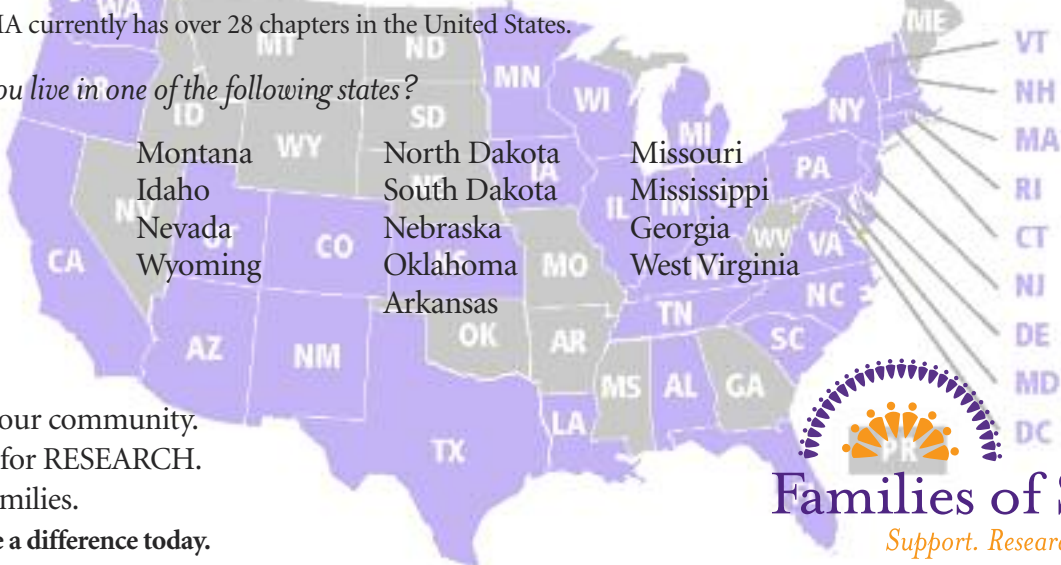
*Do you live in one of the following states?*

Montana  
 Idaho  
 Nevada  
 Wyoming

North Dakota  
 South Dakota  
 Nebraska  
 Oklahoma  
 Arkansas

Missouri  
 Mississippi  
 Georgia  
 West Virginia

SUPPORT your community.  
 Fundraising for RESEARCH.  
 HOPE for families.  
**Begin to make a difference today.**



Contact Barbara Trainor at [chapters@fsma.org](mailto:chapters@fsma.org) to receive more information on how to start a chapter in your state.

## Paratek Pharmaceuticals Inc. and Families of Spinal Muscular Atrophy Announce a Multi-Million Dollar Award from the National Institute of Neurological Diseases and Stroke to Advance a Drug Candidate for Spinal Muscular Atrophy.

**P**aratek Pharmaceuticals (Boston, MA) and Families of Spinal Muscular Atrophy (Elk Grove Village, IL) announced today that a jointly funded drug development program for Spinal Muscular Atrophy has been awarded a multi-million dollar cooperative agreement from NINDS.

The five-year cooperative agreement encompasses pre-clinical drug development up to the time of an Investigational New Drug Application (IND) to the FDA. The program is focused on developing a novel small molecule of the tetracycline family from within Paratek's propriety compound library. The potential for success of the drug candidate is evaluated at key stages of the development using quantitative go/no-go milestones that have been established in the agreement between Paratek and the NINDS. The Krainer Laboratory at Cold Spring Harbor Laboratory and the Hastings Laboratory at Rosalind Franklin University are also key collaborators in the program and will also receive funding under the NINDS cooperative agreement.

Spinal Muscular Atrophy (SMA) is the leading genetic cause of death of infants. It is an often-fatal genetic disorder resulting from the loss of both copies of the Survival Motor Neuron (SMN1) gene. This causes a chronic deficiency in the production of the SMN protein, which is essential to the proper functioning of the motor neurons in the spinal cord to the control of muscles in the limbs, neck and chest. SMA is typically marked by the deterioration of the muscles that

control crawling, walking, swallowing or breathing. There are no approved therapies for the treatment of SMA. Approximately 1 in 6000 babies born is affected. 1 in 40 people are genetic carriers, indicating approximately 7.5 million carriers in the United States.

The compounds under investigation at Paratek are intended to correct RNA splicing of a low functioning back-up gene to SMN1 called SMN2, which will in turn increase SMN protein levels. The leading drug candidate has been shown to accomplish this in a number of tissues in animal models of the disease. The focus of the project with this new grant funding will be to optimize the drug-like properties of this compound class through directed medicinal chemistry.

"At Families of SMA we are excited that our \$2 Million initial investment at an early stage of this project has provided the preliminary data to leverage larger funding amounts from NIH. We feel this grant award is wonderful validation of the Families of SMA research funding program and more specifically of Paratek's promising drug program for SMA." said Jill Jarecki Ph.D., Research Director at Families of SMA.

"The Families of SMA drug discovery strategy is to invest funds to enable companies to begin early-stage programs for this orphan disease, and as programs progress to later stages, we look for funding to transition from non-profit to commercial and government sources. Clearly Paratek's program is a successful demonstration of that approach", said Kenneth Hobby, President at Families of SMA.

Dr. Stuart B. Levy, M.D., Paratek's Vice Chairman, Chief Scientific Officer and co-founder, stated, "We are very grateful for the support of FSMA, which has helped get us to this exciting moment. We are encouraged that through the NINDS funding, we shall identify a drug candidate for this devastating disease."

John D. Porter, Ph.D., Program Director for the Paratek award at NINDS, stated, "This award was made through the NINDS Cooperative Program in Translational Research—a novel, milestone-driven program that solicits ideas for therapies from disease communities, evaluates those ideas through peer review, and both optimizes and assesses candidate therapies at each stage of development. This cooperative project combines the corporate resources and expertise of Paratek, the research experience of the academic investigators, the focus, insights, and funding of the Families of SMA, and the funding and project management experience of NINDS staff. It represents a powerful strategy for developing new therapies for SMA and other diseases."

Families of SMA and SMA Europe (through SMA Trust), a consortium of European SMA advocacy groups, have also co-funded a recent grant award to Paratek Pharmaceuticals for their Spinal Muscular Atrophy drug program. This award will cover aspects of the program not funded by the NINDS allocation, including funding to the laboratory of Dr. Louise Simard at the University of Manitoba.



Families of SMA *Research. Support. Hope.*

# COMPASS

A Publication Dedicated To Research Updates

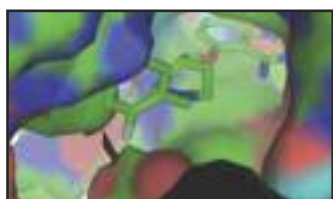
## FSMA Quinazoline Program Update: Preparations Now Underway for a pre-IND Meeting with the FDA.

By Jill Jarecki, Ph.D., Research Director FSMA

In June 2009, the IND-enabling safety studies were completed for the FSMA-directed Quinazoline program. This is the program previously worked on by Vertex Pharmaceuticals and deCODE genetics, and fully funded by Families of SMA.

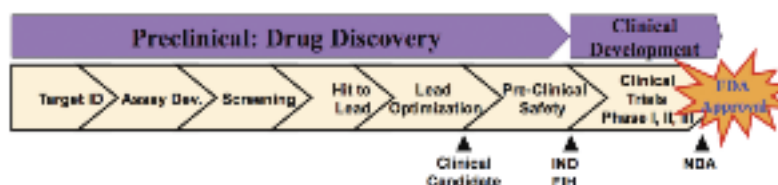
The next step for this program is to request a pre-IND meeting with the FDA and to prepare the required data package. Families of SMA will be working over the next several months with toxicology and regulatory experts to prepare for this meeting.

**This meeting will be an exciting milestone for the project, and an essential step in the path towards human clinical trials of the drug candidate.** During the pre-IND meeting, the FDA carefully assesses the existing data on the drug candidate, providing essential feedback on the feasibility of human clinical trials, and specific information on any additional experiments that may be required by the FDA for the Investigational New Drug (IND) Application.



X-ray crystallography reveals the nano-sized drug binding site of DcpS (Source: deCODE chemistry & biostructures)

### Drug Development Process



IND = Investigation New Drug Application needed for FH  
FH = First In Human  
NDA = New Drug Application for FDA approval

An IND application essentially seeks permission to begin human safety testing of a particular drug therapy. We anticipate our pre-IND meeting will occur in September of 2009.

An IND filing would be the culmination of almost 10 years of pre-clinical drug development work, which started at the earliest stage of drug discovery in assay development. This project has successfully progressed through each stage of the pre-clinical process shown in the figure above.

Many groups have collaborated with us to reach this point, including Vertex Pharmaceuticals, deCODE genetics, Invitrogen (Life Technologies), Paracelsus, the Burghes lab at Ohio State University, the Didonato lab at Northwestern University, the Kiledjian lab at Rutgers University, the Chung lab at Columbia University, and the SMA Project at NINDS.

**Families of SMA has invested over \$12 Million in this particular program to discover and develop a new drug treatment specifically designed for SMA.**

### Families of SMA Therapeutic Pipeline

FSMA is funding three distinct drug discovery programs:

- 1) Quinazolines to boost SMN2 expression;
- 2) Tetracyclines at Paratek Pharmaceuticals to correct SMN2 splicing;
- 3) Motor neuron replacement program at California Stem Cell and UCI.

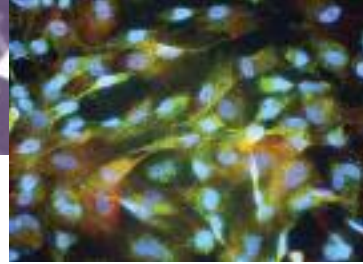
**These three programs represent an investment of over \$16 Million so far in our pursuit to build a SMA drug pipeline.**

FSMA plans to continue to invest and further expand the pipeline of drug programs. This pipeline will enable us to move multiple drugs forward at the same time. When one drug candidate drops out of consideration, another one will always be there. In addition, it allows us to tackle the treatment of SMA with several distinct approaches. Diversifying our approaches increases the chances of success.





**SUPPORT OUR DRUG DISCOVERY  
AND DEVELOPMENT PROGRAMS:**  
Please visit [curesma.org](http://curesma.org) and  
make a donation.



## FSMA-Funded Project at Paratek Pharmaceuticals Moves into Next Phase with a Multi-Million Dollar NINDS Grant Award.

By Paul Higgins, Ph.D., Director, Inflammation Drug Discovery, Paratek Pharmaceuticals.

**F**SMAs has supported the research at Paratek for the previous three years for our work on tetracycline derivatives as a potential treatment of SMA. These compounds are intended to correct SMN2 splicing and in turn increase SMN protein levels.

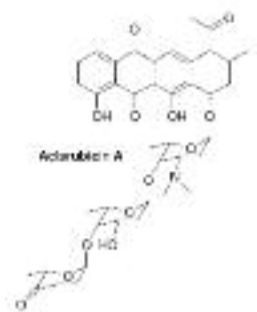
We have generated and tested many new tetracycline compounds for their ability to influence SMN2 splicing and to increase SMN levels. Importantly, several of the new compounds that can increase SMN protein levels have shown good capability to cross the blood-brain barrier (BBB) in mice, which will be a critical feature for a SMA drug. In addition, experiments were performed in SMA mice models to confirm compound ability in correcting SMN2 splicing in animals, not just in cells.

### Future Plans:

Using funding from FSMA to generate the preliminary data for a grant application, Paratek has been awarded a multi-million dollar U01 grant from the NINDS to continue our research that FSMA funded for the past 3 years.

With this funding, we will continue to synthesize new tetracycline derivatives, test them for SMN2 splicing modification and for BBB penetration in mice. Importantly, we will test additional compounds for efficacy in SMA mice. Two compounds

are particularly interesting candidates as both exhibit both splicing activity and BBB penetrability. In addition, we will test compound administration via the intrathecal route (the injection of a therapeutic agent into the sheath surrounding the spinal cord) in order to test brain-specific efficacy and to explore the possible clinical utility of this means of drug administration in humans.



Our overall goal for this project is to develop a drug candidate for SMA resulting in an Investigational New Drug (IND) application with the Food and Drug Administration (FDA) within 4 to 5 years.

For the past 3 years, FSMA has invested \$2 Million in research at Paratek Pharmaceuticals, and with collaborators in the Krainer Laboratory at Cold Spring Harbor, and in the Hastings Laboratory at Rosalind Franklin University, to discover new drugs for the treatment of SMA.

## High Purity Human Motor Neurons for Treatment of SMA Type I.

By Chris Airriess, Ph.D., Chief Operating Officer, California Stem Cell, Inc.

**C**alifornia Stem Cell (CSC) is preparing for a final FDA pre-IND meeting to take place in mid 2009. This is a critical step on the track to submitting a formal Investigational New Drug (IND) application to the FDA to begin clinical trials in SMA Type I.

CSC has developed methods for the manufacture of clinical grade human motor neurons from human embryonic stem cells. It is the intent of CSC to gain approval to begin FDA-approved clinical trials for the use of these cells in development of a cell replacement therapy for SMA Type I.

Numerous pre-clinical efficacy studies have been completed, demonstrating that the cells work. These include:

- 1) Correct localization of CSC motor neurons in the spinal cord;
- 2) Cell growth and extension out of the spinal cord toward the limbs;
- 3) Contact and synapse formation with target muscle;
- 4) Functional re-innervation of muscle, leading to restoration of limb function in animal models of motor neuron loss.

**The pivotal animal safety study, required to support an application to begin clinical trials, was completed in October 2008.** Data analysis is in the final stages and should be complete by the end of June 2009. There were no negative outcomes of this safety study, leading to the conclusion that motor neuron replacement should be a safe strategy in the treatment of diseases such as SMA characterized by motor neuron loss.



Numerous and comprehensive medical community focus groups have been held to develop the clinical strategy for SMA clinical trials. Topics included clinical site selection, inclusion criteria, route of administration, immunosuppression to prevent rejection after transplantation, and outcome measures to assess both benefit and safety of the treatment.

Manufacturing facilities and procedures have been audited for compliance with FDA guidelines for clinical manufacturing and the full Quality Assurance System is in place.

**CSC is now preparing for a final FDA pre-IND meeting to take place in mid 2009, keeping us on track for a formal IND application to begin a Phase I/IIA clinical trial in SMA Type I.** This stage of the project has included the hiring of a Medical Director and a Clinical Research Coordinator at CSC. The two new positions will be responsible for coordinating the preparation of the above IND application and for overseeing and monitoring the resulting clinical trials. The financial support for these positions came from a grant to CSC from Families of SMA through the generous support of the Dhont Family Foundation.

Families of SMA has invested over \$1.5 Million so far in a collaboration with the University of California, Irvine, California Stem Cell, Inc., and Johns Hopkins University to progress stem cell therapy for SMA to clinical trials.

# 2009 H1N1 Flu and Spinal Muscular Atrophy

## What is influenza?

Influenza (the flu) is an illness caused by a virus. It usually happens in the fall and winter but people can get the flu at other times of the year. The flu is easily spread by direct contact, coughing, sneezing, and when an infected person touches a surface that others then use, like doorknobs and railings.

## What is 2009 H1N1 flu?

2009 H1N1 (referred to as “swine flu” early on) is a new influenza virus causing illness in people. This new virus was first detected in people in the United States in April 2009. This virus is spreading from person-to-person worldwide, probably in much the same way that regular seasonal influenza viruses spread. However, it has the potential to cause more severe disease than seasonal influenza especially in individuals with SMA.

## Why is 2009 H1N1 virus sometimes called “swine flu”?

This virus was originally referred to as “swine flu” because laboratory testing showed that many of the genes in this new virus were very similar to influenza viruses that normally occur in pigs (swine) in North America. But further study has shown that this new virus is very different from what normally circulates in North American pigs. It has two genes from flu viruses that normally circulate in pigs in Europe and Asia and bird (avian) genes and human genes. Scientists call this a “quadruple reassortant” virus.

## What are the signs and symptoms of 2009 H1N1 flu in people?

The symptoms of 2009 H1N1 flu in people are similar to the symptoms of regular human influenza and include:

- fever and chills,
- cough,

- sore throat,
- runny or stuffy nose,
- body aches,
- headache,
- fatigue,
- vomiting and diarrhea are also possible.

## How does 2009 H1N1 flu spread?

Spread of the 2009 H1N1 influenza is thought to occur in the same way that seasonal flu spreads. Flu viruses are spread mainly from person to person when someone with influenza coughs or sneezes. Sometimes a person may become infected with the flu by touching something that has flu viruses on it and then touching their mouth or nose.

## For how long is 2009 H1N1 flu contagious?

Infected people may be able to infect others beginning 1 day before symptoms develop and up to 7 or more days after becoming sick. That means that you may be able to pass on the flu to someone else before you know you are sick, as well as while you are sick.

## How can I avoid 2009 H1N1 flu and other germs?

The seasonal influenza vaccine is currently available to protect against seasonal influenza.

The 2009 H1N1 vaccine is in production and should be ready for the public soon. When the 2009 H1N1 vaccine is available the following are target groups for vaccination:

- pregnant women,
- persons who live with or provide care for infants aged <6 months (e.g., parents, siblings, and daycare providers),
- health-care and emergency medical services personnel,

- persons aged 6 months--24 years, and
- persons aged 25--64 years who have medical conditions that put them at higher risk for influenza-related complications, such as SMA.

In addition, there are everyday actions people can take to stay healthy:

- Clean your hands with alcohol-based hand gel or soap and water, especially after you cough or sneeze.
- Cover your nose and mouth with a tissue when coughing or sneezing, throw it away, then clean your hands.
- Avoid touching your eyes, nose or mouth since germs spread that way.
- Avoid close contact with sick people.

## What should I do if I think I or my child has the flu?

If you or your child has SMA and you think you or they may also have the flu, call your doctor. If you get sick, the Centers for Disease Control and Prevention (CDC) recommends that you stay home from work or school and limit contact with others to keep from infecting them. If you have 2009 H1N1 flu your doctor may prescribe an antiviral drug like oseltamivir (Tamiflu®) or zanamivir (Relenza®) that can modify the severity of your illness. If you or your child with SMA are exposed to a confirmed case of 2009 H1N1 influenza A (H1N1) virus, contact your doctor for consideration of medication for 10 days to prevent the symptoms of H1N1 influenza.

For up-to-date information on 2009 H1N1 influenza, visit:  
<http://www.cdc.gov/h1n1flu/qa.htm>.

## Families of Spinal Muscular Atrophy Announces New Clinical Trial Site for SMA at Duke University in North Carolina.

Families of Spinal Muscular Atrophy is pleased to announce that Duke University in Durham, North Carolina has been added as a new site to the clinical trial network Project Cure SMA.

Dr. Priya Kishnani, Professor of Pediatrics, Chief Division of Medical Genetics, will serve as the site director for the study. Dr. Edward C. Smith, Medical Instructor in the Department of Pediatrics, Division of Neurology, will be assisting at the site. Dr. Kishnani has critical experience conducting clinical trials in infants with neuromuscular disease, having been the lead principle investigator during the clinical trials of the Genzyme Corporation drug Myozyme® for Pompe Disease.

The Duke site is participating as part of the multi-center trial of Valproic Acid and Carnitine in infants with Type I Spinal Muscular Atrophy ongoing at seven centers across North America.

The addition of the Duke University site is critical to this trial and to the future success of clinical studies in the SMA patient population. Because Type I infants have a difficult time traveling, multiple regional trial sites will be key to conducting clinical trials for SMA. Establishing a Project Cure SMA site in the Southeastern United States is another step in our long-term goal of growing the clinical network to encompass regional coverage across the country in preparation for pivotal trials of novel SMA drugs.

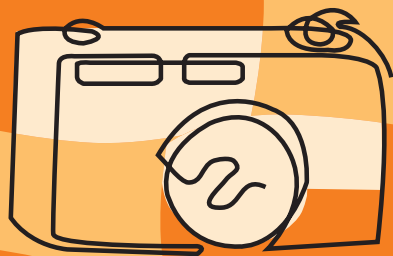
## VALIANT SMA Study Clinical Trial Enrollment Complete

Project Cure SMA clinical trial network is pleased to announce that enrollment for the Phase II VALIANT SMA Clinical Trial has concluded. Enrollment began in September 2007 at The Ohio State University Medical Center and at this time we have enrolled more than 32 participants, which exceeds our goal of 28. Twenty participants have completed the study and we expect all participants to finish the study by November 2010.

“This has been an extraordinary clinical trial. While enrollment was challenging at times due to participant travel and work scheduling conflicts, we are thrilled with the data we have collected and believe it will provide valuable insights into treatment and evaluation methodologies for SMA researchers and clinicians. Participants in the study also felt it was an invaluable experience.” According to, Sharon Chelnick, study coordinator, “For many, it was the first time they were able to talk to physicians and other professionals who truly understood their disease and some of the challenges they face”.

Results are expected to be published in early 2011.

## Be a “Directions” Contributor



Photos, poems, articles based on your knowledge and experiences, summaries of great FSMA fundraisers...we want them all! This is your chance to share. Your contributions will help to make this publication even better. Please email text either in the body of an email or attached as a word document. Photos submission requirements are:

- dimensions 1600 x 1200 pixels
- jpg format
- 2 megapixel
- Approx. Print Size 4" x 6"

All materials can be sent to [newsletter@fsma.org](mailto:newsletter@fsma.org) or via mail to the National Office, Newsletter, Families of SMA, 925 Busse Road, Elk Grove Village, IL 60007.



# New Paper Published on Spinal Muscular Atrophy Clinical Trial Results

**P**roject Cure SMA Group Publishes Open Label Valproic Acid Trial Results. The paper entitled “Phase II Open Label Study of Valproic Acid (VPA) in Spinal Muscular Atrophy” was published in the online Journal PLoS ONE. This clinical trial was fully funded by Families of SMA.

The paper can be obtained at <http://www.plosone.org/> and the “Open Label Study of Valproic Acid in Spinal Muscular Atrophy” Trial was registered was ClinicalTrials.gov as NCT00374075.

The paper presents the data from an open label trial of VPA in 42 subjects with SMA to assess safety and explore potential outcome measures to help guide design of future clinical trials. The results indicated that VPA was well-tolerated and without evident hepatotoxicity. Carnitine depletion was frequent, and temporarily associated with increased weakness in two subjects, indicating a need for co-administration of carnitine with VPA. Clear decline in motor function occurred in several subjects in association with weight gain. Mean fat mass increased without a corresponding increase in lean mass, suggesting that weight gain is likely to be significant confounding factor in future VPA clinical trials. A significant improvement in motor function, as measured by the Modified Hammersmith Functional Motor Scale (MHFMS), was observed in participants younger than 5 years of age.

The authors concluded that the study provides good evidence that VPA can be used safely in SMA subjects over 2 years of age in the setting of close monitoring of carnitine status. However, they also indicated that further studies of VPA in infants and young children are needed to better assess safety in this more vulnerable cohort, since children under 2 were not included in the current study.

The results presented within the PLoS ONE publication suggest that while there may be a potential treatment benefit in a subset of younger non-ambulatory type II children, conversely, older subjects may be at risk due to excessive weight gain. Given the uncontrolled nature of the study, it is unclear whether the improvement in some younger subjects reflects a therapeutic drug effect, maturation, or increased cooperation leading to improved functional measurement scores. These data underline the importance of randomized, controlled efficacy studies to assess the impact of therapies in



SMA, and that the same drug could have differing results in select subsets of patients.

“These results are significant not only because they are a first step to determining the therapeutic benefit of VPA for SMA, but also because they inform us about proper clinical trial design. These results demonstrate the need for randomized controlled trials in subsets of patients, since much weaker (Type I) and much stronger (Type III) patients may respond quite differently to the same intervention. In addition, the clinical outcome measures used to examine patients over such a wide range of strength and function will differ. Using the correct measure for each population will be critical to prove efficacy”, says Dr. Kathryn Swoboda, lead author on this paper.

The data presented in the paper indicates that several additional clinical studies are warranted in order to assess the efficacy of VPA for SMA. These studies, funded by Families of SMA, include:

- 1) A double-blind placebo controlled trial of VPA and carnitine in a non-ambulatory group of Type II children.
- 2) An open label safety study in Type I infants, an extremely vulnerable group of patients.
- 3) A double-blind placebo controlled crossover study in ambulatory adults with SMA.

“Families of SMA is pleased to have the first clinical trial results published from the work of the Project Cure SMA Clinical Trial Network. Funding clinical trial initiatives allows our community to achieve multiple goals. It gives us the means to develop the required outcome measures to test drugs in all SMA populations, to conduct trials to test repurposed drugs for safety and efficacy in SMA patients, and to build the necessary infrastructure, including adequate regional clinical trial site representation across the US for future new drug trials”, says Kenneth Hobby, Executive Director of Families of SMA.

# ACTIVE families

www.curesma.org



**Haley and Addison Kuester with the Easter Bunny**



**Trevor Broton petting a sheep**



**Kale Shiesley at the beach**



**Jack Freedman with Philadelphia Eagles cheerleaders**



**Sarah Barber skiing**



**Kiley and Quinn McClay in a snowball fight**



**Mary Nelson and her Dad on the Hudson**



**Sprinkler Time for Charlie Sykora**



**Lizzy Hallam and Charlie Sykora swimming**



**Malorie Fox and Sydney Potjer golfing**



**Jacob Slaymaker with the Simpsons**



**Andrew Murray and Dad on a Roller Coaster**



**SMA Families at Faith's Lodge**



**Ally Krajewski at the Sesame Street Show**



## Multi-Center Trial of Valproic Acid and Carnitine in Infants with Type I Spinal Muscular Atrophy Has Completed Enrollment in North America.

A clinical trial designed to evaluate the combination of Valproic Acid (VPA) and L-Carnitine for the treatment of SMA in infants with Type I SMA, called Carni-Val Type I, is being conducted by the Project Cure SMA Group. This trial, which is fully funded by Families of SMA, will assess the safety of VPA and L-Carnitine in infants and develop improved methods to assess the strength and motor abilities of severely affected infants.

The following seven North American sites have now completed enrollment.

- Salt Lake City, Utah
- Durham, North Carolina
- Detroit, Michigan
- Baltimore, Maryland
- Montreal, Canada
- Columbus, Ohio
- Madison, Wisconsin

Enrollment at the German site in Cologne is ongoing.

**More details about the trial can be found at [www.clinicaltrials.gov](http://www.clinicaltrials.gov) and [www.projectcuresma.org](http://www.projectcuresma.org).**

Families who are interested in being contacted for clinical trial participation should register with the International SMA Patient Registry at Indiana University (<https://smaregistry.iu.edu/>).

Thirty-six infants with SMA Type I, ages 2 weeks to 12 months have been enrolled in the trial at our North American sites, and trial recruitment is closed at this time. Enrolled infants receive drug treatment for 12 months. Therefore we anticipate the trial will be completed in the fall of 2010 and results released in 2011.

Dr. Kathryn J. Swoboda, Principal Investigator for Project Cure SMA said, “We are very pleased to have successfully reached our target enrollment in the SMA CARNI-VAL Type I trial. This achievement represents a monumental effort on the part of coordinators, investigators and families, who worked closely together to meet the needs of these fragile infants and enable their participation. This milestone should prove invaluable in helping us to effectively measure meaningful change in outcomes for those most severely affected by this devastating disease. We hope that the successful completion of this clinical trial will reduce barriers to future studies in SMA infants for new therapies currently under development”.

### About Project Cure SMA

In 2001, Families of Spinal Muscular Atrophy established and single-handedly funded a clinical trials network called Project Cure SMA. This network has conducted natural history studies that increase our understanding of Spinal Muscular Atrophy disease progression, built models for designing SMA clinical trials, and now runs clinical trials with existing drugs. FSMA’s investment of over \$6 Million to date in five multi-center clinical trials is helping to test existing drugs that may lead to a treatment for Spinal Muscular Atrophy. In addition, as novel drugs currently being designed for SMA become available, having a fully operational clinical network with a sufficient number of sites to conduct pivotal SMA drug trials will help attract and encourage biotech and pharmaceutical companies to invest in SMA drug development.

Funding for all support staff and for all sites in the United States is being provided by Families of SMA. Funding for the site in Cologne, Germany is being provided by the “Initiative Forschung und Therapie für SMA”. Funding for the site in Montréal, Canada is being provided by Families of SMA Canada.

### Families of Spinal Muscular Atrophy Awards \$400,000 in New Contracts to Support Ongoing Clinical Trials

Families of Spinal Muscular Atrophy awarded \$400,000 for 6 new grants to support the clinical trial network Project Cure SMA for 2009. These new awards are for the functions that support ongoing trials being conducted by the network. This new funding is in addition to that awarded directly to each of the project cure clinical trial sites.

The new awards will provide continued funding for the infrastructure portion of the clinical trials network. This includes the Clinical Trials Manager, the Data Coordinating Center, the Lead Evaluator Physical Therapist, the Lead Occupational Therapist, the Data Analysis Statistician, and the Laboratory performing gene expression analysis.

The Project Cure SMA clinical trials network currently includes 7 sites in North America, 2 International sites, and the entire infrastructure needed to complete a clinical trial. Over thirty professionals are involved.





the International SMA Patient Registry

**uniting families with researchers to find a treatment and cure for SMA**

**Researchers**

Visit the site to learn how the registry can help connect you to the people and data your research needs.

**Participants**

Visit the site for more information or to learn how you can join the registry.

This project is supported by the Patient Advisory Group of the International Coordinating Committee for SMA Clinical Trials, which includes Families of SMA, FightSMA, the Muscular Dystrophy Association, the Spinal Muscular Atrophy Foundation, and other SMA advocacy groups.

For more information call 1-866-482-0248

e-mail [smareg@iupui.edu](mailto:smareg@iupui.edu)

<https://smaregistry.iu.edu>

Memberships can be made online at [www.curesma.org](http://www.curesma.org)

# MEMBERSHIP form



## Suggested Annual Donation for Membership

Family           \$30  
Professional   \$35  
International   \$40

\* We ask for a donation to help cover the costs associated with the printing and mailing of our newsletters. You should receive quarterly editions of both the Directions newsletter and Research Compass. We hope these publications provide valuable information and useful support.

## Payment Method

- Check
- Money Order
- VISA
- Mastercard
- Discover

\$

Amount enclosed or to be charged

Credit Card #

Expiration Date

Name on card

Signature

Family Name

Email

Address

City, State, Zip

Home Phone

Work Phone

Mobile Phone

Mother/Wife/Guardian

Father/Husband/Guardian

Relationship to affected person

Affected person name

M / F      Date of Birth

SMA Type

Current Status

Date of diagnosis

Date of death  
(if applicable)

Other persons NOT affected by SMA (siblings, children, parents)

Name

M / F      Date of birth

Name

M / F      Date of birth

Can we add your name/address to our family contact list?

YES

NO

Return form to **FSMA Membership**, 925 Busse Road, Elk Grove Village, IL 60007 or FAX to 847.367.7623

09DIRFA

This section has been designed to enable you to pull it out. It includes membership forms, donation forms, as well as a memorial section.

## Zane Suzanne Schmid

January 2, 2009 – June 18, 2009

*Diagnosed with SMA- 2/19/09*

Our beloved daughter Zane was called to heaven on June 18th 2009 at 3:37 p.m. She was being embraced by her parents and was surrounded by family and friends when she passed. Zane had big, beautiful brown eyes in which she used to communicate. They were so expressive! Her smile was infectious and made you feel good. She was such a happy baby and an excellent patient. She only cried when she was hungry, and when the cough assist was being used on her. During her short 5 ½ months, she created so much awareness about SMA and made a lasting impression on thousands of people with her story. Here's our story...

My husband Keith and I were blessed with twin girls on January 2nd, 2009. We named the girls Avery and Zane. We were elated by their arrival and so excited to become parents. Around one month of age, we began to notice a lack of motor movement and low tone in Zane's body. At the girls' six-week check-up, I talked to the pediatrician about our concerns. After a physical assessment of Zane, he directed us to A.I. DuPont Hospital for Children in Wilmington, Delaware. Zane was admitted for four days and put through a battery of tests. On February 19th, we received the devastating diagnosis. Zane had Spinal Muscular Atrophy Type I. We had never heard of this disease. The doctors explained to us what the disease was, and the prognosis for children who were afflicted with it. Keith and I were scared, angry, heartbroken, and sad..... Yet determined for Zane to beat this disease. Avery was tested and is thankfully, SMA free.

We, along with family and friends began researching the disease. Families of Spinal Muscular Atrophy, we learned was a valuable resource for education, contacts, and research. We sprinted into action. During the last four months our family, friends, and community have gone to great lengths to help Sweet Baby Zane.



### Some of the things we have done are posted below:

- Created a website [www.sweetbabyzane.com](http://www.sweetbabyzane.com)
- Enrolled Zane in a clinical trial through John's Hopkins Medical Center in Baltimore, Maryland
- Created a video to help create awareness of SMA and what Zane's and our daily lives incur
- Raised over \$39,000 to help pay for Zane's medical care
- Established Zane in The National Transplant Assistance Fund (NTAF)
- Established prayer chains throughout the United States
- Designed and sold Sweet Baby Zane t-shirts
- The children in the community have organized lemonade stands and bake sales
- Have contacted the local media, and had Zane's story out to the public
- Established a Sweet Baby Zane Facebook page

We were lucky to have Zane healthy for three months. As the tumultuous journey began, we visited specialists weekly. We had machines delivered to our home, did G-tube feedings, implemented respiratory and Early Intervention therapies. All of this while doing our best to have Avery and Zane develop their social, and developmental skills. On May 19th, Zane became instantly pale, and she began breathing quicker than normal. After taking her Oxygen level and discovering it was very low we put her in the car, and drove to the emergency room. Zane was admitted to DuPont Hospital where she "lived" for one month in the Pediatric Intensive Care Unit. She had contracted para-influenza, a type of flu. As she battled this illness, it was heartbreaking for our family to be apart. It was difficult for the girls not to see each other. It was also difficult for Keith and I not to see each other. Between Keith, family members, friends, and myself,



# LOVING memories

Zane always had someone by her side. She was not going to go through this alone. The doctors and nurses took excellent care of our daughter. After a few ups and downs, and almost a full month, Zane became healthy again and was finally sent home on June 15th.

We were so excited to have our family whole again. We had to prepare for Zane's arrival. We had to create another nursery, due to all the machines and equipment that was necessary for her daily needs. Schedules were posted, directions to machines were visible, and the medications were organized. After 12 hours of being home, Zane's oxygen level dropped again. She was very congested, and we tried several therapies before calling 911 at 2:30 in the morning. She was taken to Bryn Mawr Hospital in Bryn Mawr, Pa, where she was stabilized. After stabilization, we went via ambulance to Dupont Hospital..... again into the PICU. The staff was so shocked to see us back so quickly. It hadn't been 24 hours. This time, Zane had mucous plugs in her lungs. For the second time, the staff did an excellent job of caring for Zane. They were very aggressive in the therapies, hoping she would react quickly. Zane wasn't herself this time. Previously, she had tolerated all of the poking and prodding with a smile. This time was different. The spark in her eyes was gone. She rarely smiled, even to the sight and sound of familiar faces and voices. She looked so unhappy, and it was heart wrenching for us to watch her suffer. After two days, Zane's progress was deteriorating.

On June 18th, our world was shattered when Zane peacefully passed away in our arms. Although we find solace in the fact that she is no longer limited by her disability, our beautiful baby was taken from us. Avery will always have a twin- sister. I will always be the mother of twins, and Keith a father of twins.

We are determined to bring awareness of this disgusting disease. How is it that so few people know about it when it takes the lives of so many children? Education and awareness are the keys in ending SMA. We will continue to spread awareness and raise money for FSMA.

Zane, we love and miss you so much. We can't believe you are gone, but hope that you are able to move freely in Heaven. You are forever in our minds, hearts, and souls. We will share your story and courage with Avery.

Sincerely,  
Hillary Schmid



**Jaelyn Michelle Martins**

January 22, 2008 – June 9, 2009

God saw that she was getting tired  
And a cure was not to be,  
So He put His arms around her and  
Whispered "Come with Me"  
With tearful eyes we watched her suffer  
And saw her fade away.  
Although we couldn't bear to lose her,  
We could not bid her stay.  
A golden heart stopped beating,  
Loving hands laid to rest.  
God broke our hearts to prove to us  
He only takes the best.



**Matthew James Troland**

January 5, 2007 – June 27, 2007

*Please accept this donation in memory of our grandson Matthew James Troland. It was the happiest day of our lives when he was born. And it will always be the saddest day of our lives the day he died. I hope and pray they will find a cure soon.*

Thank you,  
Mary & Jim Troland

## Madelyn Lake

January 1, 2002 – September 13, 2002

*Dear Families of SMA,*

Seven years ago, my family and I became aware of SMA when my 6 month old daughter, Madelyn Elizabeth Lake, was diagnosed with Type I. Two months later, Madelyn lost her battle to this disease, but we will always appreciate the knowledge, guidance, and dedication shown by your organization. Seven years ago, my dad created another acronym for SMA and posted it anytime Madelyn was hospitalized. For our family, SMA stands for Someone Mighty Awesome!

Nineteen months ago, I gave birth to a healthy, little boy! Two days later, my father, John M. Cook was diagnosed with small cell carcinoma (lung cancer). My dad fought a long, hard battle and endured numerous rounds of chemotherapy and radiation while continuing to work in law enforcement.

On numerous occasions, our dad would always comment on the number of lives Madelyn touched in her short time here. He dearly loved all of his grandchildren, but Madelyn always held a special place in his heart. Last week, we learned that he touched many lives in the 30 years he served our community. He earned the respect of his peers, community figures and citizens. The Sheriff's Department Chaplain was stopped by a woman who shared that she and her daughter would not be alive without the help of our dad. A letter was written by Family Services, on behalf of the children of Grant County, thanking our dad for his assiduous work in protecting abused and neglected children.

In the final week of my father's life, I had the privilege of caring for him at home along with my brother, mother and close family friends. He was able to meet my 5 day old, niece Leah. As I sat at his bedside, I was 39 weeks and 5 days pregnant. My father continued to fight his battle, but we encouraged him to let go. We sensed he was waiting on the last grand-child so I whispered in his ear that she maybe waiting on him. One hour later, I went into labor.

My mother stood in the delivery room and called home to announce the arrival of Sophia Marie. Although my dad was non-responsive, his eyes opened when he heard the cry of his newest grand-daughter. Throughout the day, he began responding to our family and friends. Our dad wanted to make sure he was here for both grand-daughters births and he met his goal.

I called home late Sunday night (Aug. 9th) and learned my dad was non-responsive. My mother placed the phone next to my dad; I told him I loved him and would be by his side tomorrow morning. The next morning, while I was preparing to leave for the hospital, my mom called stating my dad passed away very peacefully (while holding Madelyn's blanket) at 10:15 AM.

Please accept these donations in memory of Captain John M. Cook. He would be honored to help your organization and the families affected by Spinal Muscular Atrophy.

**Sincerely yours,  
Amy (Lake) Rendel on behalf of the family of Captain John M. Cook**



## Connor Jumper

August 24, 2006 –  
July 14, 2009

Connor was a very sweet loving little boy and for the families who may be newly diagnosed I'd like them to see that these children are so happy and no matter what they may go through, it never gets them down. I'm sure they can see that through Connor's pictures. He was always a happy child.

**Thank you,  
Jessica Jumper**

**Dear FSMA,**

This small check is being sent in memory of little Oliver Mastin, who was such a joy to his parents, grandparents and great-grandparents (to all that knew him).

Such a short life, but loved so much. I appreciate everyone of you who donate your time and effort to learn and help people with this terrible disease.

**Sincerely,  
Thelma Gentry**

**Skylar Bahrenburg**  
 April 24, 2001 – June 21, 2008

*Dear Family and Friends,*

It is with a heavy heart that we write our 2008 holiday letter. As many of you know, our precious son, Skylar, who has been battling Spinal Muscular Atrophy, passed away in June at the tender age of 7. We had just spent an amazing week at a beach house in Kailua, Hawaii with Jim's mom and two of his brothers and their families. Skylar went to sleep in the car as we returned from the airport and did not wake up. Although we knew Skylar's time with us would be limited, we were not prepared for the suddenness of his passing.

We are so thankful to our family and friends and the Sonoma and Presentation School community. Our entire Amlicke, Bahrenburg, Clark, Quade, and Matthews clan flew to Sonoma immediately. Skylar's first grade class organized a beautiful tribute in our neighborhood park. Over 400 people attended his Memorial Service which was held at the Presentation School, the private school that both Spencer and Skylar attended (You can view his Memorial Video at [www.skylarsfight.com](http://www.skylarsfight.com)). A tree was planted at school for Skylar; a Gazebo at a new Children's Home in Hawaii is being dedicated to Skylar as well as a wing of a new orphanage in Nairobi, Kenya. Skylar has an amazing ability to touch so many lives and bring so many people together. We are so thankful to everyone for their cards, kind words, flowers, meals, wine, donations and loving support. All of you are helping us get through these heartbreaking days.

We are so thankful to have Spencer (9) and Sawyer (15 months) to focus our energy. Sawyer is a joy who is growing every day. His smiles and giggles warm our hearts. He is just on the verge of walking and his vocabulary skills are constantly improving. The current favorite words are "cat", "cheese", "cracker", and most recently, thanks to daddy, "tackle".

Spencer is in 4th grade. He is a good student who is very talented in math and language arts but prefers recess, art, and music. He recently adopted an orange tabby cat named "Rowland" for who he enjoys caring. Spencer is playing CYO basketball and scored in his first game. We were very proud. Spencer is also turning into an avid chess player.

And so we must close this letter before we run out of space... Remember each day how precious life is. Don't forget to give your kids one more hug, one more song, one more toss of a ball, one more game of chess... This past year has reinforced how important it is to live in the now.

Our wish for you is much love, health, and happiness. Thank you to all of you who have supported our family through our journey.

**All Our Love,**  
**The Bahrenburgs**  
**Jim, Liz, Spencer, Sawyer and our angel Skylar**



**All we really need to know we learned from Skylar**

- Have a positive attitude at all times.
- Laugh from your toes up.
- Sing loudly. Smile often.
- Love your family.
- Be protective of your friends, and let them be protective of you.
- Zoom around and if you run over someone's toes say you're sorry.
- Be silly. Don't let anyone name your baby brother Slugo.
- Even if you can't play, jump in and try.
- Read as much as you can – and if you can get away with it read Pokemon or Captain Underpants during silent reading.
- Hold hands in class. Touch each other.
- Cherish your siblings.
- Take risks. Insist on fairness.
- Be good to everyone.
- Push the rules. Challenge yourself.
- Don't let anyone tell you you can't do something.
- Squawk when you want to be heard.
- Always bring something for sharing.
- Live life to the fullest.

**-Jennifer Hainstock-**





**Andrew Glenn Butler**  
*January 8, 2009 – June 4, 2009*



## Andy's Story

*Written by Audra Perry Butler*

On January 8, 2009, my husband and I welcomed our firstborn son, Andrew Glenn Butler, into the world. A beautiful baby boy that we called "Andy" with thick brown hair and bright blue eyes, he was the best thing that ever happened to us. After 10 years of marriage, our family finally was complete.

At the time of his birth, we thought that Andy was a healthy, thriving baby boy. He kicked, punched and cried like any other newborn for the first month of his life. However, when he was just weeks old, we noticed that his movements were getting weaker, instead of stronger.

We took him to our pediatrician on March 3, 2009, who immediately admitted Andy to the ICU of University Community Hospital in Tampa. Initially, he was diagnosed as having hypotonia, which is the lack of muscle tone and movement in his limbs, as well as having fluid in his lungs. Andy underwent a battery of invasive, diagnostic tests to determine the cause of these symptoms. We slowly were beginning to realize that something was terribly wrong with our baby.

The doctors said that their working diagnosis was spinal muscular atrophy (SMA). They believed that Andy had the most severe form of the disease, Type 1, also called Werdnig-Hoffmann Disease. We immediately searched the Internet for this disease and found that SMA was the number one genetic killer of children under the age of two. It was every parent's worst nightmare.

We turned off the computer in shock. We cried as we counted all of the symptoms of this insidious disease that Andy already was showing – lack of movement, floppy arms and legs, no neck strength, feeding issues. How was this happening to us? How could we suddenly go from having a thriving baby to one on the brink of death? How had we never heard of this disease?

On March 17, 2009, our worst case scenario became our reality when our son's diagnosis of SMA Type 1 was confirmed. Andy was just nine weeks old. His doctors told us that his prognosis was six months. We couldn't believe that our baby might not live to see his first birthday.

While we were in the depths of despair, our friends and family reached out to Families of Spinal Muscular Atrophy for help. Every day it seemed another box arrived on our doorstep, filled with information about how to cope with the progression of the disease, how to deal with our grief, and how to make Andy's life as happy and comfortable as possible. We received a lambskin for his bed as well as a lovely handmade blanket and many light toys. The local Greater Florida FSMA chapter offered much needed guidance and support. We also received e-mails, letters and pictures from other affected families, extending their friendship. We are so thankful for the love and support of the SMA community.

Although we were heartbroken, we pushed our feelings of loss aside and committed ourselves to celebrating each day of Andy's life. We showered him with love and affection. We danced with him and sang to him every day. We put a portable DVD player in his crib so he could watch videos and adapted toys so he could play. And, we had birthday parties for him every month, because each month we feared he wouldn't be there for the next.

In the weeks following his diagnosis, we had countless doctor's visits and therapy sessions. Andy was put on several medications to help reduce his reflux and to promote his muscle strength. But, with no real treatment protocol available, the disease still caused his muscles to weaken a little more each day, eventually impacting his ability to suck, swallow and breathe. His symptoms were progressing very quickly, regardless of what we did to try to slow them.

But, through it all, Andy still laughed and smiled. He was a cognitively advanced baby with an attentive look in his eyes and an unbelievably long attention span. No matter how weak he was, he still cooed along with music, was captivated by his Baby Einstein videos, and flirted with his nurses. A cuddler, he loved to be held and rocked. He even learned to give kisses. That Andy was such a happy baby kept us going as we continued our fight against his SMA.

Between March and June 2009, Andy was hospitalized four more times at All Children's Hospital in St. Petersburg for respiratory and gastrointestinal issues, including one surgery and another stay in the ICU. And, it was in the hospital on June 4, 2009, that Andy lost his battle with this insidious disease. He was just 20 weeks old.

Andy was physically weak, but he was strong in spirit. He was the bravest person my husband and I have ever known. He taught us to smile in the face of grief and proved the power of laughter. He made us realize how important it is not to take time or family for granted. He renewed our faith, because we have to believe that there is a greater plan for our family.

And, with his passing, Andy gave us a mission. We will do everything we can to prevent another family from going through this horror. With the support of our friends, family and FSMA, we are committed to sharing Andy's story and spreading awareness about this terrible disease. We created "Andy's Army" on Facebook ([www.causes.com/andysarmy](http://www.causes.com/andysarmy)) to share information about SMA and how people can help stop it as well as to raise funds for FSMA. We now are active members of the Greater Florida FSMA chapter with several fundraisers underway or being planned for the upcoming months.

Our son's life, while brief, was profound, and his legacy is everlasting. We will miss him every day and look forward to the day that we meet again, somewhere over the rainbow. Until then, we will continue his fight against SMA as Andy's Army, and together we will find a cure.

*Poem by Alan Butler for his son, Andy:*

### **My Dream Boy**

Some dream of things that you can do  
Some have dreams of old, others new  
Some dream of things that you touch  
A few of little and others of much  
That was not for me.

I dreamed a dream of love  
A dream that came from above  
My dream was easy and it never got harder  
It was one day to be someone's father  
That was for me.

My little boy came to me this year  
His smile it ran from ear to ear  
A beautiful face with eyes so blue  
From the very first moment his love was true  
He gave that to me.

But soon we learned he had to fight  
We put the gloves on gave a left and a right  
We jumped through hoops and made the right moves  
To fight the disease we had something to prove  
I fought it with him.

On March 17th we learned time was short  
SMA Type 1 was the doctor's report  
We knew we had a limited time  
My poor baby boy was no longer fine  
He would be taken from me.

He fought the hard fight, my brave little boy  
Loved us every minute and loved his toys  
He gave me everything I needed from him  
Even as he weakened and knew he couldn't win  
I loved everything about him.

On June the 4th he lost his fight  
We stayed with him into the night  
He passed away to the angels in peace  
To a better place and to be finally free  
We said he could go.

So here I am standing with a broken heart  
For my beautiful boy who never got a start  
My dream was cut short and taken away  
But I know we will meet again someday  
I will miss him every day of my life  
Until we meet again in paradise.

**I love you Andy, My Dream Boy.**



**KATHERINE MABEL VANCE**

*November 19, 2008 – April 27, 2009*

A huge THANK YOU to FSMA for the nice gestures, blankets, gifts, kind thoughts that came with this horrifying diagnosis. The toys and blankets and support helped a lot.... Thank you for the work that you do. I just wish you did not have to do it.....

**Chip Vance**

**In Loving Memory of Katherine Mabel Vance**



*IN MEMORIAM*

**AMBER BURKHOLDER**

*SEPTEMBER 16, 2008 - JANUARY 10, 2009*



*IN MEMORIAM*

**KE'AJA CHISHOLM**

*JANUARY 29, 2002 - MARCH 8, 2002*

## Sarah Ann McColl

December 24, 2008 – August 20, 2009



**S**arah Ann McColl was born on Christmas Eve at 2:57 PM. She came into the world screaming as she was taken out of the warm confines of Heather's womb. Sarah was a beautiful creature that lit up

each room that she entered or every time you walked into see her. She wrapped this dad around her finger the first time her tiny hand grasped my finger. Sarah had this striking beauty that drew you to stop and linger with her and to stare into her beautiful blue eyes.

Heather and I knew for a while that Sarah was considered a "floppy baby" and was later diagnosed with a general term called Hypotonia, which means that she had low muscle tone. So we started working with an amazing program called First Steps, thanks to a friend's suggestion. Sarah had two amazing Therapists, Gina and April. They taught her to turn her head to the right and left, to hold her head in the middle of her body, to reach out and grasp things with both hands, and to roll over with assistance. In the couple of months that they worked with Sarah, she made great progress and loved her time with them.

Heather, Sarah and I got to do a few trips together in her time with us. We got to go to Pittsburgh for a wedding in April where we went to the Zoo and Aquarium and she escorted me into the reception. Then in May we travelled to North Carolina to see family and to watch her dad and Aunt Laura, along with other friends run a Tri at White Lake, while there she got to dip her toes into White Lake. She also got to travel to Christ mount, her Grandparents' Houses in North Carolina and Indiana. She also attended her first General Assembly in Indianapolis where she met her Cousin Amanda.

Sarah went to the neurologist on August 7th and was tested for various things including a genetic disorder called Spinal Muscular Atrophy. SMA is a degenerative disease that has different types. Sarah was tested for Type 1, which is for infants and is fatal.

On Friday, August 14, Sarah had a cold and was struggling to breath so Heather took her to the emergency room at UK Hospital and she was admitted to the Pediatric ICU. She was on oxygen and an IV drip and Antibiotics for possible Pneumonia. Saturday and Sunday

she was doing better and we were working to bring her home. On Monday, she had a set back and the middle lobe of her right lung had collapsed. She was put back on High Flow oxygen to help reinflate her lung.

At this point we started to talk about keeping Sarah comfortable and not in pain. So on Wednesday morning we met with the UK Palliative Care Team and Daniel's Care, a part of Hospice for children. At this point, our doctors were pretty sure that she had SMA and we were looking for the best care possible for Sarah.

On Wednesday afternoon, around 12:15 PM she went into Cardiac Arrest. The PICU team did CPR and brought her back to life. At that point we knew that it was time to make her as comfortable as possible. We removed her oxygen and her NG tube and we got to hold her and talk to her and she was passed from family member to family member. Heather and I got to lay down with her and talk with her and she slept on my chest all night long. This morning about 7:00 AM we made the decision to bring her home and she calmed right down. Heather and I got to walk with her out of the hospital and feel the breeze blowing across her face and brought her home where we were able to give her a bath, put on an outfit from Costa Rica and a Lady Bug Dress, she loved her Lady Bugs. We also got to put her in her swing and to see her dog Jake and some of our cats. Daniels Care came to our house and started the process of her care. Sarah was hungry and so we offered her some milk and after that small taste she quietly and quickly with no struggle and no pain walked over to sit in God's Arms where she can sit up, crawl and run.

Our little Sarah bear, left this world to God's world at 12:50 PM on Thursday, August 20th.

Our angel is now home where she can be the baby that this world would not let her be. She is smiling, crawling, sitting up, and saying Hi to all because our beautiful baby never knew a stranger. She was and is love, pure and simple. And we blessed to have her choose us to be her mother and her father.

**Mike McColl**





**Morgan Jane Sitzman**

*December 9, 2008 – June 27, 2009*

*To All Parents*

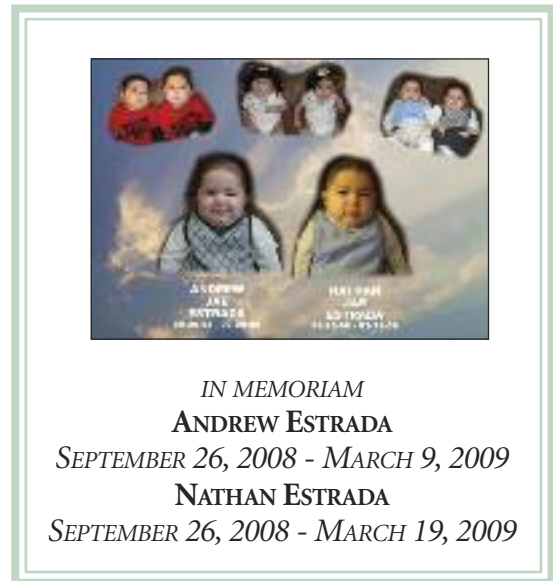
“I’ll let you have, for a little while, “a child of mine” He said,  
 “For you to love the while she lives and mourn for when she’s dead.  
 It may be six or seven years or twenty-two or –three.  
 But will you, ‘till I call her back, take care of her for me?”  
 “I’ll bring her charms to gladden you and should her stay be brief  
 You’ll have her lovely memories as solace for your grief.  
 I cannot promise she will stay since all from earth return,  
 But there are lessons taught down there I want this child to learn.  
 I’ve looked the wide world over in my search for teachers true  
 And from the throngs that crowd Life’s lanes, I have selected you.  
 Now hate Me when I come to take her back again?”

I fancied that I heard you say “Dear Lord, Thy will be done.  
 For all the joy Thy child shall bring, the risk of grief we’ll run.  
 We’ll shelter her with tenderness; we’ll love her while we may  
 And for the happiness we’ve known, forever grateful stay.  
 But should your angels call for her much sooner than we’ve planned,  
 We’ll brave the bitter grief that comes and try to understand.”

**Author Unknown**



IN MEMORIAM  
**ALEX COLE DEVINCENZI**  
*OCTOBER 22, 2008 - APRIL 12, 2009*



IN MEMORIAM  
**ANDREW ESTRADA**  
*SEPTEMBER 26, 2008 - MARCH 9, 2009*  
**NATHAN ESTRADA**  
*SEPTEMBER 26, 2008 - MARCH 19, 2009*



IN MEMORIAM  
**BLAKE ELLIS HILL**  
*MAY 31, 2002 - AUGUST 2, 2009*



*IN MEMORIAM*  
**LIZETH OROZCO DAVILA**  
*JUNE 23, 2008 - NOVEMBER 18, 2008*



*IN MEMORIAM*  
**MIKEY UNDERHILL**  
*DECEMBER 14, 2007 -  
DECEMBER 17, 2008*



*IN MEMORIAM*  
**KEIRA SWEENEY**  
*APRIL 5, 2008 - JANUARY 26, 2008*



*IN MEMORIAM*  
**JIREH LENNOX WHITE LINTON**  
*OCTOBER 19, 2000 - JUNE 4, 2009*

## **Brendon Dupree**

July 26, 1990 – February 2, 2009

*Families of SMA,*

Thank you for the beautiful card and figurine, it is a beautiful reminder of my son. We are hoping there will be lots of money coming to you all in Brendon's memory - so others will not have to watch their children die of this horrible disease. Thank you for your support, we will continue our support of you all as well.

**Thank you again,**

**Ron, Shana, Rachael, Jake & Evan Dupree**

Donation Form

09DIRFA

I want to make a donation in the amount of \$

In honor of \_\_\_\_\_

Donor Name \_\_\_\_\_

Donor Address \_\_\_\_\_

Notice of donation—Name & Address \_\_\_\_\_

Payment Method

- Check  Money Order
- VISA  Mastercard  Discover

Credit Card # \_\_\_\_\_

Expiration Date \_\_\_\_\_

Name on card \_\_\_\_\_

Signature \_\_\_\_\_

Return form to **FSMA Donations**, 925 Busse Road, Elk Grove Village, IL 60007 or FAX to 847.367.7623

Gift Certificate Order Form

09DIRFA

Gift Certificate vendor list available online or by calling 800-886-1762.

Retailer	Qty	\$ Amount	TOTAL
_____	_____	_____	_____
_____	_____	_____	_____
_____	_____	_____	_____

Payment Method

- Check  Money Order
- VISA  Mastercard  Discover

Credit Card # \_\_\_\_\_

Expiration Date \_\_\_\_\_

Name on card \_\_\_\_\_

Signature \_\_\_\_\_

Ship order to

\_\_\_\_\_

\_\_\_\_\_

\_\_\_\_\_

Phone \_\_\_\_\_

**Shipping** Up to \$499 -\$8 • Over \$500 -\$10

Subtotal \$ \_\_\_\_\_

Shipping \$ \_\_\_\_\_

**Total \$** \_\_\_\_\_

Return form to **FSMA Gift Certificates**, 925 Busse Road, Elk Grove Village, IL 60007 or FAX to 847.367.7623

Merchandise Order Form

09DIRFA

Item	Size	Qty	Description	\$ Amount	TOTAL
_____	_____	_____	_____	_____	_____
_____	_____	_____	_____	_____	_____
_____	_____	_____	_____	_____	_____

Payment Method

- Check  Money Order
- VISA  Mastercard  Discover

Credit Card # \_\_\_\_\_

Expiration Date \_\_\_\_\_

Name on card \_\_\_\_\_

Signature \_\_\_\_\_

Ship order to

\_\_\_\_\_

\_\_\_\_\_

\_\_\_\_\_

Phone \_\_\_\_\_

**Shipping** Up to \$25 -\$4 • \$26 to \$50 -\$7 • \$51 to \$75 -\$10 • Over \$76 -\$13

Subtotal \$ \_\_\_\_\_

Shipping \$ \_\_\_\_\_

**Total \$** \_\_\_\_\_

Return form to **FSMA Merchandise**, 925 Busse Road, Elk Grove Village, IL 60007 or FAX to 847.367.7623



# FSMA merchandise

www.curesma.org

**NEW Items!**



**Baseball Cap | \$10**  
New FSMA Logo.



**FSMA Car Magnet | \$5**  
1 design.



**Travel Mug | \$10**  
New FSMA Logo.



**Sport Bottle | \$5**  
New FSMA Logo.

**Mail your Orders to  
FSMA National Office  
or fax to 847-367-7623  
Order form is on Page 42**



**Families of SMA T-Shirt | \$12**  
1 design on short sleeve white shirt.  
Kid sizes: 2/4 6/8 10/12 14/16  
Adult sizes: S M L XL



**Bag | \$10**  
New FSMA logo.



**25 Year Grocery bag | \$5**  
Large bag with logo on either side.



**25 Anniversary T-Shirt | \$12**  
1 design on short sleeve grey shirt.  
Kid sizes: 2/4 6/8 10/12 14/16  
Adult sizes: S M L XL

**NEW Items!**



**FSMA Keychain | \$5**  
New FSMA Logo.



**FSMA Lunch Bag | \$10**  
Insulated with bottle holder.



**Umbrella | \$20**  
1 design.



**Spring Note Cards | \$6**

Alyssa Silva's "Working on Walking" note cards. 6 different cards.



**American Cards | \$6**

Alyssa Silva's "American" note cards. 6 different cards.



**Thank You Cards | \$6**

Alyssa Silva's "Thank You" note cards. 6 different cards.



**FSMA "Moving Forward" T-Shirt | \$12**

Sizes: S M L XL



**Wipe Out SMA | \$12**

1 design on short sleeve shirt. Kid sizes: 2/4 6/8 10/12 14/16 Adult sizes: S M L XL

**Donation Gift Card \$10ea. or 5 for \$40**

Have you ever needed a gift for a teacher or a birthday gift for a "hard to buy for" person? These elegant cards are a perfect gift for when you want to thank someone for their kindness, honor someone who makes a difference in your life, or mark a celebration. Instead of buying teachers, doctors and anyone else another scarf or trinket, consider giving a FSMA gift card.



**Merchandise can be ordered online at [www.curesma.org](http://www.curesma.org)**



**Angel Wing Pins | \$10**



**Cure SMA Bracelet | \$2**

Available in small or large. Created in honor of Steven Potter.



**License Plate Frame | \$7**

Please note that as of **October 1st**, Families of SMA has a new mailing address. The address is:



**Cookbook for a Cure | \$14**

The FSMA cookbook contains almost 400 recipes from SMA families all over the world!



While overall giving to charity was down by a full 2% in 2008, online giving grew by 44% to more than \$15.4 billion dollars in the US!



Events from Birthday Parties to Backyard Bashes. It is fun and easy to fundraise online. Help cure SMA and plan an event today. For help, email the office at [fundraising@fsma.org](mailto:fundraising@fsma.org)



**Get your FSMA fundraising event listed on the FSMA Website.**

Submit your FSMA event information via email to: [fundraising@fsma.org](mailto:fundraising@fsma.org)





## Celebrate a Quarter Century of Dedication and Determination!

Host your own Families of SMA fundraiser to support research and other critical support programs. Our support programs include the equipment pool, new caring booklets, the annual Family & Professionals Conference, and much more...

### We can help!

- Online Registration and Fundraising
- Send eblast or Postcards to members in your area
- Signs, Posters, & Banners
- Your own unique event page on the FSMA website
- Sell FSMA Merchandise

*We're All Ears!*



Give Us Your  
Ideas &  
Feedback...

Families of SMA is always looking for new fundraising ideas to help us continue funding research and offer support programs. If you have an idea or feedback, we would love to hear from you. To submit an idea, please fill out the form below and mail it to the office at FSMA – **Fundraising Idea, 925 Busse Road, Elk Grove Village, IL 60007**, or email your name, address, phone number and idea to [fundraising@fsma.org](mailto:fundraising@fsma.org).

### FUNDRAISING IDEA:

\_\_\_\_\_  
Name

\_\_\_\_\_  
Address

\_\_\_\_\_  
Phone Number

\_\_\_\_\_  
Email

Mary and Joe McHale and Nancy and Andris Dindzan held the **9th Annual Concert for a Cure** on May 19th at Blackhawk Country Club in Danville, CA. The event was a smashing success, raising over **\$100,000** for FSMA. The evening featured incredible silent and live auction items; such as, a golf package at LaQuinta, a Monterey Getaway, a weeks stay in Molokai, Hawaii, and much more! Concert for a Cure has now raised over \$650,000 for FSMA!



On September 19, 2008 the “Friends of Lanie Hannah” held their **8th Annual Lanie Hannah Benefit Scramble Golf Tournament**. The event was held at Canyon West Golf Club in Weatherford, Texas. The day featured, lunch, golf, dinner, live and silent auctions. The Friends of Lanie Hannah generously donated **\$33,000** to Families of SMA.

On February 28th, Glenn and Colleen McWilliams, along with many other volunteers held the first **Rockin’ a Cure 2009: A Benefit for Spinal Muscular Atrophy**. The event was held at the California Aerospace Museum in McClellan, CA and featured live music by Delta Breeze, live and silent auctions, gourmet food and wine, as well as an airplane ride simulator. The evening was held in memory of Luke and Megan McWilliams. The evening was a huge success and raised over **\$28,100** for FSMA. Stay tuned, plans are underway for the 2010 event.



Mike and Abbie Dougherty of Charlotte, NC held their first **Gray’s Gang Walk-n-Roll** on April 25th in honor of their son, Gray. The Dougherty family has done numerous fundraisers in the past. The event brought several SMA families together and a great time was had by all. The walk raised an outstanding **\$25,600** for FSMA.

A Special Thank You to the Following Fundraising Teams:

- Michael, Abbie, & Gray Dougherty - \$11,665
- William Wood, Jr. & Margaret Wood - \$805
- Erin Dougherty - \$295
- Danita Chisholm - \$285
- Lindsay Thalinger - \$165
- Ronetta Newby Butts - \$25.



Dear FSMA,

Thank you for coming out to **Ski Away SMA** this year! Close to 100 people were at Magic Mountain in southern Vermont for a fun day of ski races, tubing, good food, music, and spring skiing fun. So, Taylor and I are thrilled to be able to write a check for **\$25,000** to FSMA this year from the Hailey Mae Foundation.

Hailey continues to touch so many lives and although we miss her terribly, how wonderful that each year so many gather in her memory to have fun and raise money in the hopes that SMA will be a thing of the past someday soon.

**All our best,  
Marie and Taylor Smelser**



Eric and Katie Sitzman of Lincoln, NE held the **Chubby Bubby Benefit: A Celebration of the Life of Morgan Jane Sitzman** on July 25, 2009. The event was held at The Nebraska Club and included an evening of silent and live auctions and music by R-Style. The Sitzman’s first fundraising event raised over an incredible **\$20,000** to benefit Families of SMA.

Rod and Kristi Gellner of Fargo, ND held their annual **Jack Attack on SMA**, Round 5 on Saturday, June 6th in honor of their son, Jack. The event raised **\$16,567** for SMA, bringing their 5 year grand total to nearly \$100,000 raised!

*To Families of SMA:*

Please accept the enclosed donation of **\$13,200** on behalf of my daughter, Joanna Buoniconti. As you are aware, Joanna suffers from SMA, and on Monday, April 20, 2009 I ran the **Boston Marathon** in her honor.

This was the fifth year that I have participated in the race. My official time was 3:38:28; it is my personal best, and I am proud to have done this as a tribute to Joanna.

I am grateful for the support of my friends and family, and I am hopeful that their kindness and generosity will help find a much-needed cure for SMA.

**Very truly yours,  
Stephen J. Buoniconti**



Eric and Susan Kutell and Marcy and Matthew Fantel worked together to host a **Neighborhood Social** in honor of their grandson/son Ray on August 23rd in Monroe Township, NJ. The event featured silent and live auctions and was attended by over 200 people. They raised over **\$12,000**, with more money trickling in every day!



*Dear FSMA,*  
The **Cure SMA: Run, Walk-N-Roll** was held on April 25, 2009 in Stephenville, Texas in honor of Seth Gilley. Also attending were, Lanie Hannah and her family from Weatherford, TX, Hannah Ostermeyer and her dad from Dallas, TX, Savanna Rush and her family from Saginaw, TX, and Jake Stamper with his family and friends from Leander, TX. Morieann Jackson and many supporters came from Ft. Worth, TX to walk in memory of her son Jordan. Keven and Kelly Coggin, along with little brother, Camden, came from Austin, TX to walk in memory of their daughter, Grace. Rosie Echeverris came from Mesquite, TX to walk in honor of her granddaughter, Madison Villegas, from Kansas City.

It was a terrific day! Over 400 people participated in the fundraiser benefiting Families of SMA. After the walk, we had a silent auction, bounce houses for the kids, live music from the DHB Band, and a hamburger lunch. Savanna Rush and her family brought Savanna's Sno-Cones and raised over \$300. Thank you, Rush Family! Cindy Webb, a family friend from Springtown, TX, had a garage sale to benefit FSMA and raised over \$1,500. Thank you, Cindy! Linda

Gilley, Seth's grandmother, made a quilt and raffled it, raising \$1,900. Thank you, GG! All together, the event raised over **\$11,000** for FSMA!

Big thanks go out to all of the SMA families that attended. Other big thanks go out to our friends and family for all of your generous support and hard work. The fundraiser wouldn't have been so successful without you! And, thank you to FSMA for all your efforts in helping find a cure for our kids!

**Lacy Gilley**

A Special Thank You to the Following Fundraising Teams!

Lacy & Chad Gilley - \$1,106  
Renee Griffen - \$345  
Leo & Ione Villegas - \$295

Christy Greene and Evelyn Vasquez organized the **2nd Annual Inland Empire Walk-N-Roll** that was held at Fairmount Park in Riverside, CA on Sunday, June 7th for FSMA. The event raised \$9,601 for Families of SMA.

A Special Thank You to the Following Fundraising Teams!

Yelena Sololova - \$2,810  
Kenneth & Christy Greene - \$1,270  
Cindy Henry - \$585  
Evelyn Vasquez - \$240



Ruthie Henry - \$170  
 Irma Horchstetler - \$160  
 Crystal Estrada - \$100  
 Christie Godinho - \$100  
 Heather Burkhart - \$60  
 Maria Pollack - \$50  
 Lily Steffen - \$25

*Dear Families of SMA,*

Our **1st Bowl-a-thon** in honor of our son, Zachary, who passed away June 24, 2009, was a success. We were able to raise close to \$7,500 to benefit FSMA. The bowl-a-thon took place on April 25, 2009 a few days after Zachary would have been 1 years old. Many family members, old friends, and new friends (that we met through FSMA) attended. It was amazing to see all of the support we had. About 30 bowling teams signed up and we took over two-thirds of Parkside Lanes in Aurora, IL. Many local and chain stores donated goods to use in our raffle and silent auction. My husband and I want to thank everyone who participated and volunteered to help to make the day such a success.

**Sincerely,**  
**Sherri Deutsche**

*Dear Emmy's Crop Supporters,*

On March 28, 2009, the third annual **Emmy's Crop for SMA** was held at the New Oxford Pavilion in New Oxford, Pennsylvania. Over one hundred women scrap booked from 9 AM to 9 PM, while enjoying time with family and friends. We had great food and great prizes. The women took part in

free make and take classes, best page contests, raffles and silent auctions. With help from friends and family, and all our supporters, we raised \$6,500 to help find a cure for Spinal Muscular Atrophy (SMA). The funds raised were donated to Families of SMA in memory of Emmy Rose Baugher. Emmy received her angel wings on December 27, 2006 at six months in age.

This year I am happy to announce that over \$20,000 has been donated to Families of SMA since December of 2006 in memory of our beautiful Emmy Rose. With your help and support "together we will find a cure".

**Thank you,**  
**Brandy Baugher and Sandra Cromer**

Marcy and Matthew Fantel, of Kendall Park, NJ held **Happy Hour for SMA** on May 8th in honor of their son, Ray. The event was held in New York City and was attended by about 70 people. The evening featured a bucket auction and raised a total of \$6,500 for FSMA.

*To Whom It May Concern:*

I am writing to let you know that once again our school, **CA Montessori Children's Center**, is offering a dance class to our families and have decided to donate all of the proceeds to your organization. We felt that the children should be dancing for others who may not have the opportunity to do so in their lifetime.

Enclosed you will find checks from the families in varying amounts, which

were their donations for the dance program. So far we have collected \$3,000 for Families of SMA. This does not include the CA Matching Gifts. Many of the families have attached the Matching Gift letter as our company matches 100% of the donation.

We will continue to send more checks or online donations as they are submitted, but wanted to make sure that you have enough time to process the matching gift, as there is a time frame associated with it.

The grand total raised was nearly \$6,000!

In addition, we would like to make this donation in honor of Greyson Shepherd Erwin, the nephew of Claudine Campanelli (a manager at the Montessori Children's Center) who is a very brave and strong little boy living every day to the fullest with SMA.

**Regards,**  
**Jennifer Fuggini**  
**Islandia, NY**

Amy Allen of Rochester, MN was happy to send along more than \$5,600 raised at this year's **Lindsey Ronningen Pancake breakfast**. The annual event is held in memory of her daughter, Lindsey. Congratulations and pass the syrup!

Allie Mazzella of Short Hills, NJ held the **2nd Annual Walk-n-Roll to Cure SMA** in Millburn, NJ in memory of Katherine Santiago. The walk was a huge success and raised \$5,160 for FSMA.

More than \$3,800 was donated in memory of Will at the **Will Blumensaadt Benefit** from donors like The Jet Express and Weickert Insurance Agency, Inc. Thanks so much for your generous donations.

Rob Young and friends ran the **Boston Marathon** in honor of William Johnson. They raised funds for FSMA in support of their efforts and were able to donate \$3,642 to support SMA research.



Dear FSMA,

Enclosed you will find a donation totaling over \$1,800. This donation was collected recently at Justin's dad, **Dennis Kuester's, retirement party**. As a Grandpa and Grandma to Addison Kuester (SMA Type I), Justin's parents decided that they wanted no gifts but rather that money be donated to FSMA in honor of Addison. Some people gave directly to FSMA. We believe the total donations are over \$3,000. Many people wrote checks. Please put the money to good use to find a cure for this horrible disease. Your organization has done so many things for my family. Thank you for all that you do.

Sincerely,  
**Justin and Shannon Kuester**  
Hartford, WI

Dear FSMA,

Please find the enclosed checks totaling \$2,871. This contribution comes from the **4th Annual Lily Kennedy Golf Outing** in memory of our daughter, Lily Kennedy; sponsored by the St. Francis University Social Work Jr. class, under the supervision of Dr. Mark Lynch and Suzanne Black. Thanks for all that you do.

**Heather Kennedy**  
East Freedom, PA



*Families of SMA,*

Enclosed please find checks totaling \$2,775 for funds raised during our **3rd Annual Ayden's Swim for a Cure**. The swim was held on July 12th in Phillipsburg, KS with children from the area collecting funds for SMA research during the months of June and July. A

swim party and balloon launch was held for all the kids with many prizes awarded. Ayden's Swim for a Cure was organized in honor of Ayden Trammell who was diagnosed with SMA Type I in December of 2006.

Thank you,  
**Donna Studley**  
Phillipsburg, KS

*To Whom It May Concern:*

Please find the enclosed donations from the employees of **Cubist Pharmaceuticals**, in honor of Macarthur Sohl. We were pleased to be able to raise a total of \$2,548 for your cause at our holiday gathering in January. We look forward to more events to raise donations and awareness for your group.

Thank you and best wishes in the New Year,

**Kate Sohl**  
Natick, MA

Jamie Novak was happy to pass along a donation from Robert Pope of Minot Builders Supply Association in Minot, North Dakota and President of the Board of Director of the Western North Dakota Charity Pro-Am Golf, Inc. The generous \$2,500 donation was greatly appreciated.



**Mary Beth Butch** and family held a fundraising coffee at their lovely home in honor of Charlie Sykora. Charlie is 6 years old and has SMA Type I. More than 35 women attended, and Charlie truly enjoyed being the center of attention and surrounded by so many "pretty girls" as he says. The fundraiser raised more than \$2,500 for Families of SMA, as well as raised awareness of SMA in our community.

Dear FSMA,

On February 14th I had a fundraiser for FSMA. We celebrated a life cut short from SMA. Some very good friends of ours, Maggie and Andy Golobic lost their son, Kash to this dreadful disease. Thank you for helping raise awareness of SMA. We hope this donation helps with research and hopefully a cure! We raised \$2,307, not bad for one night!

Thank you for all you do.

**Becky Zang**  
Cincinnati, OH

Dear FSMA,

Our fundraiser was a huge success! The Solonators (the band that I sing in) performed and raised over \$2,000 for FSMA. Enclosed you will find the checks/cc slips, as well as one check that I made out for \$857, which was the total cash contributions.

Let me know if you have any questions. Thanks! We had a blast and I will definitely send you some pictures when I get some!

Warmest Regards,  
**Melissa House**  
Ann Arbor, MI

Kay and Vince Wittman of Jerseyville, IL once again held their **Annual Golf Day for Medical Research** in honor of their daughter, Amy. The event raised \$1,690 to support SMA research.

Chris Cooter and the Calvary Riders TEAM of Motorcyclists for Jesus Ministries of Warminster, PA were happy to send Families of SMA a donation of \$1,688 which was raised at the **4th Annual Ride to Fight Spinal Muscular Atrophy**. Great job and thanks to the Calvary Riders for their hard work at this annual event.





**Olson Family Christmas in July 2009**

From a very early age, my husband Wade, his brother Scott and their six cousins spent Christmas at their (maternal) Grandmother Dee Olson's house. Each year names are drawn and everyone buys/receives one gift. This tradition has continued, long after Grandma Dee passed away. Some years ago, the family decided to celebrate Christmas in July at the lake cabin in Detroit Lakes, Minnesota. Last year, as a group, it was decided that rather than exchange gifts, we would donate the money (intended for a gift) to a charity. Each year, a different Olson sister family will choose the charity.

This year, the Beverly (Olson) Tranby family chose SMA. Our son Jonathan had SMA Type I. He passed away December 16, 2003 at the tender age of seven months, two days. Jonathan was the first great-grandchild of Dee Olson and the first grandson for Beverly & Orville Tranby.

We are blessed with a wonderful, loving and compassionate family. As we continue in our journey of grief, it is comforting to have the love and support from this family.

We decorated a SMA donation box and handed out SMA facts cards. We committed to matching the amount collected. We are so proud of the extended Olson families! Together we raised \$1,540!

**We again want to thank everyone for supporting SMA:**

Beverly & Orville Tranby

Scott Tranby  
 Jackie & Len Evenson  
 Linda, Michael, Alexander & Kristian Sandilands  
 Trevor Evenson  
 Kristin Rutigliano  
 Susan & Tim Wilhelm  
 Hannah Wilhelm  
 Miriam Wilhelm  
 Gloria & Phil Thompson  
 Betty Thompson  
 Monica, Ryan and Maxwell Nelson  
 Mark Thompson

**Sincerely,**

**Wad, Sandra, Wyatt and Jack Tranby**

*Dear FSMA,*

Enclosed are the proceeds from our online auction benefiting FSMA. We raised \$1,304! I am also sending back the unused thank you notes. Thank you for sending them! It was nice to send out my thank you & receipts in them!

Thank you for your help and patience.

**Sincerely,**

**Monica Eberhardt  
 Charleston, SC**

For several years, the ladies of the Phi Tau Omega Sorority – Delta Phi Chapter of Maryland Heights, MO have held their annual trivia night and donate funds to benefit Families of SMA. This year, the ladies raised an amazing \$1,243 for FSMA.

Sherry DeGraffenreid of Eugene, MO was happy to pass along a generous donation of \$1063 raised from selling T-shirts during Change for the Cure. Sherry organized the event in honor of her granddaughter, Grace DeGraffenreid.

The Fairfield Ludlowe Class of 2009 does its best to instill a solid core of morals, values and sense of community in their students and it really shows! This year the Class of 2009 donated \$1,000 in honor of Cubby Wax to fund critically important research!



*Dear Families of SMA,*

My name is Theresa Nelson. My family and I live in North Port, Florida. My husband and I are also the proud parents of four beautiful girls; Kayla-16, Taylor-13, Kristina-8, and Jessica-7. Jessica just happens to be our child affected with SMA. Back on April 4th, North Port had their 50th Anniversary Games Celebration. The city agreed that I could have a hotdog stand to help raise money for SMA. This also enabled me to help spread the awareness. I am enclosing some pictures and a check for the money we were able to raise (\$795). Smoothie King was also a vendor at this event in which they donated a portion of what they received for the day. A big thank you to Publix who also donated money to help pay for the food and drinks. I would also like to extend a big heart felt thank you to all of you who are helping us in our fight to wipe out SMA. Together we will find a cure.

**God Bless all of you.  
 The Nelson Family**

My grandson, Michael "Mikey" James Underhill died from SMA Type I on December 16, 2008, three days shy of his first birthday. SMA robbed Mikey from most of his physical abilities and every breath was a struggle, but it didn't touch his bright blue expressive eyes.



His contagious smile was worth more than a million dollars. With everything he had to endure everyday to survive, he truly taught everyone who came in contact with him to appreciate every breath you take; it may be your last. He also reminded us to never take anyone or anything for granted! Mikey will always be my inspiration to keep raising money to help find a cure for SMA!

So, on May 21, 2009, I held a cookout/raffle fundraiser at my employer, Shaws Supermarket in Rockland, Maine. They donated the food and several businesses donated certificates for the raffle. A local radio station, Real Country 103.3 advertised the fundraiser.

Two special guys, Rob Johnson and Paul Bossie, who work in the grocery department, generously volunteered their time to cook the big juicy burgers. My team of angels, Barbie, Doreen, Kevin and Jeanette also helped and supported the cause.

Liz Chase made pins for us to wear, with a picture of Mikey wearing his Elmo hat that I made for him. So, I made a similar hat for me to wear in memory of Mikey. The fundraiser was a huge success! Enclosed is a check for \$700. I'm so thankful for everyone who contributed to the cause. Hope to do it again next year!

**Sincerely,  
Sandi Fogg  
Union, Maine**

*Hello Families of SMA,*

Hope this finds you doing well. Please be advised I'll be forwarding a check in the amount of \$635. Our IT Department had a luncheon near the end of the year and raised that amount for charity, and chose FSMA as the recipient. Even in these tough economic times it's amazing how generous people are!

**Thanks very much,  
Douglas and Michelle Erwin  
Massapequa, NY**

Blaise and Melanie Fritsche pooled the money from their graduation and confirmation parties to send Families of SMA a donation of more than \$635. The generous donation was in memory of their cousin Michael James Underhill.

Andy and Heather Simmons of New Bedford, MA donated \$500 to FSMA after holding their first fundraiser on May 7, 2009 in honor of their son, Owen Reid Simmons, who was diagnosed with SMA in December 2008. They hope to make their event an annual one, helping to find a cure for SMA.

The School District of Osceola County Florida was pleased to send along a donation of \$419 from donations collected from students. Donations were collected for SMA at St. Cloud Middle School in St. Cloud, FL.

The Cheltenham Township Police Association of Wyncote, PA was happy to send along a donation in memory of Gavin Patrick Crews. Members from the Association, including Officer Kevin O'Donnell (husband of Sue Crews O'Donnell/Gavin's aunt) took a trip to watch the Philadelphia Phillies whip up on the Cincinnati Reds 8-7 in extra innings. Despite a bit of rain, Association members were able to donate \$350 and are planning another trip to watch the Phillies play the Marlins... Go Phillies!

Michael and Joann Campanelli of Levittown, NY, in celebration of beginning a new chapter of their lives together requested in lieu of gifts, donations could be made to Families of SMA in honor of Michael's grandson, Greyson Erwin. Donations for the wedding celebration totaled \$300 to benefit FSMA.

ATDC and Executive Director Bob Policastro were pleased to announce their fundraiser to support children's disease research was a great success. ATDC donated \$250 in honor of Brianna

Carlino, Dylan Cuevas, Kily McDonnell, Alex Rodriguez, Michael Brodsky – the children from Long Island that ATDC assists.



Matthew Shepard was very appreciative of his classmates donating a portion of the 50/50 raffle to Families of SMA at their Tiffin Columbian High School Class of 1994 Reunion. Lisa Bivens Click who won the raffle donated her winnings as well – generating \$225 to support research to find a cure. The donation was made in honor of Matthew's 19 month old son Thomas who has SMA Type II. Matthew wanted to thank Janice Keesey Brunner and Heather Detillion Staib co-organizers of the reunion, as well as, say thank you for all of the hard work everyone does on behalf of Thomas and all of the other children with SMA.

Anthony and Charisse Pulkrabek of Angus, MN along with their family members held a small fundraiser in honor of their son, Cole for FSMA. Their efforts and generosity raised \$220 to support SMA research.

Margie Fetter, member of the Greater Clark County Schools Spirit Committee is proud to pass along \$202 in honor of Colby Russ. The donation was collected at the 2008 Casual-for-Charity Days. Dressing casual at work and donating to families of SMA – a win/win situation!

Dorothy Shuler, Maureen Gill and Lauren Lundy O'Conner all participated in the San Diego Half Marathon in honor of Owen Shuler to raise money for Sophia's Cure. \$189 was raised and donated to Families of SMA at this great event.

Duska Hepner and the rest of the staff of Tolland Middle School in Tolland CT hold a Dress Down Day each payday. In May they collected a total of \$147 and are sending that along to Families of SMA. Thanks for being comfortable!

*Dear Families of SMA,*

I am writing you this letter somewhat delayed, so I apologize. For the past three years I have held a Pampered Chef Fundraiser in the month of December. I do this in honor of my twins Owen and Grant who were born December 28, 2004. It is hard to believe they would have been four this past year. Unfortunately, SMA Type I took their lives way too soon (Owen at 11 weeks; Grant at 15 weeks).

Jeanette Arnold, Pampered Chef Consultant has once again donated her profit from the sales totaling \$145 this year. My Aunt and I have donated \$50 each as our Christmas gifts to each other.

I know nothing will bring back my beautiful Owen and Grant, but I know the struggles and pain that SMA families face. One day, God willing, we will no longer have to lose our loved ones. Thank you for all the work that you all do for the SMA cause!

**Sincerely,  
Vanessa Burns  
Marietta, OH**



The ladies of Curves in Allentown, PA raised funds for FSMA in honor of Griffen Kingkiner. Their fundraising efforts totaled \$125 to benefit Families of SMA.

*To Whom It May Concern:*

Please accept the enclosed donations (\$115) in the names of Scott and Linda

Buttrey on behalf of their grandson, William Scott Blumensaadt, who died of SMA. These donations were given as a 40th wedding anniversary gift to Scott and Linda. They have been very blessed through their years of marriage, except for the loss of Will. We feel this is the best gift we can give them.

**Sincerely,  
Cheryl Abrams**

*Families of SMA,*

Each Christmas my 5th grade class picks a project to contribute to instead of exchanging individual gifts. This year's class chose to raise money for FSMA. One of our class members has SMA, Brandon Cavendish. Brandon is a great inspiration to all who know him. He is such a bright, happy child. Brandon has faced and will face many challenges. We hope that this small gift (\$100) will help Brandon and all children with SMA.

**Sincerely,  
Linda Sparks  
5th grade teacher  
Summersville Elementary School**

*Hi FSMA,*

My name is John Hamilton, and I am a student from the University of Illinois at Chicago. I talked to you over the summer about the student organization of which I am president, Medical Research Funding Society, raising money for Families of SMA. We had our first fundraising event for FSMA, and raised \$100. All the money came from UIC students over a four hour period who participated in our event, which was a 3-point contest on the Playstation 3. We had hoped for sponsorship from local businesses around UIC; however, we were told that many, many student organizations try to get donations from the local UIC businesses, so we had an uphill battle.

**Regards,  
John Hamilton  
President, Medical Research Funding Society  
University of Illinois, Chicago**

Parrilla Grill, Johannes Ariens, and Mick & Deb Sprague were all part of the \$100 donation from RACE – thanks so much for your generous gift.

*To Families of SMA,*

Our office has implemented a “Casual Day” program which enables employees to dress casually on Friday of each week in exchange for a \$1 donation. When the amount collected reaches \$50, one participant is drawn and the “winner” is allowed to pick the charity to which the \$50 amount is donated.

Recently, Beth Hinkley's name was drawn, and she requested that a donation to Families of SMA be made.

Please accept this contribution of \$50 to assist you in your work.

**Sincerely,  
The Laskoff & Associates staff  
Lewiston, ME**

Christopher Foy of Holtsville, New York was happy to pass along the NLF Playoff pool winnings for his Uncle Bob Ferguson in memory of their niece Deirdre Abrales. Congrats on winning the pool and thank you for the generous donation.

*Hello,*

The Home Laureates Extension Homemakers Club each year sells holiday candies and nuts. All proceeds raised are given to non-profit groups. This year the club voted to donate to the Families of Spinal Muscular Atrophy in honor of Katie and Betsy Swetnam of Muncie, Indiana.

**Best Regards,  
Jackie Johnston, Treasurer**

Please note that as of **October 1st**, Families of SMA has a new mailing address. The address is:







Trevor Broton



Tambryn and Braelyn Campbell



Zachary Span



Sarah Barber



Jake Saxton



Savannah Rush and her Dad



Stella Turnbull



Jake Saxton and his Dad



Stella Turnbull with Dr. Schroth and Lisa Pharro



Malorie Fox and Syney Potjer



Sophia Salva and Lizzy Hallam



Sara Greene



Carol Swenson was glad to send along the donations from the 8th Grade class at Dakota Hills Middle School in Eagan, MN. Their teacher, Tera Runquist had the students plan and sell products for a good cause as part of a class project.

This year the students wanted to donate in support of their fellow student Abbey Partridge. Carol says the students did all the work and the Parents Booster just deposited the funds and wrote the check for \$625! Congrats to the 8th Grade class for a job well done!

Cindy Koons, Principal at St. Albert the Great School in Huntingdon Valley, PA was excited to announce a donation of \$478 to Families of SMA. The students had a "dress down" day where students paid \$1 each to be contributed in memory of Colin Lynch. Great job and gold stars all around!

*Dear Families of SMA,*

This past Saturday we threw another joint birthday party for our daughter, Madison Herrera and her best friend, Daniel Chavez. Just as we did last year, we asked that our guests bring a donation in lieu of gifts and we raised \$305 for FSMA. So please accept the enclosed checks as donations made in honor of Madison and Daniel's 5th birthdays.

**Best,  
Brandy Herrera**

Josh Ubben, of Bloomington, MN had a birthday party and instead of gifts, he requested donations be made to Families of SMA in honor of his cousin, Charlie Cowan. Josh's party raised \$221 to benefit FSMA.

The students from Mrs. Wittman's class at Delhi Elementary School in Jerseyville, IL donated \$200 to FSMA in lieu of a Christmas gift for their teacher.

*To Whom It May Concern:*

The Jr. Beta Club at Lost Mountain Middle School sold lollipops for Valentine's Day and would like to contribute some of the proceeds to your organization in honor of one of our members, Chessa Rose Birrell. Please accept this donation, \$119.

**Sincerely,  
Karen Glisson  
Jr. Beta Club Sponsor  
Kennesaw, GA**

Thirteen year old Taylor Ruisch was assigned a group project in school. The project called *Imagine It*, is normally a college-aged assignment where you have to figure out how 100 post it notes can better the world. Taylor and her group took it on full force. They decided to do something for charity and chose SMA in honor of Taylor's cousin Charlie Sykora who has SMA Type I. They made the sign, filled it with 100

post-it notes reading "Thank you for donating to SMA. Your donation will help children around the world." They set up a table at 5 Guys Burgers and Fries and raised \$115 in two hours. They educated a lot of people about SMA and even won Best Project for their class. Taylor said it was great and that they had a lot of fun!

Olivia, age 11 of Braintree, MA made bracelets and sold them in honor of Owen Norton, SMA Type II, age 6. Olivia is a good friend of Owens's sister Caitlin and neighbor. Olivia's bracelets raised \$26 for Families of SMA.

Kara Jenneker, age 9 of Troy, MI sold Cure SMA bracelets in honor of her sister, Angela and was able to raise \$25 for FSMA. Way to go Kara!

Gaelyn, Braedyn, Matthew and Delia McGuire, along with Caitlin Norton of Braintree, MA sold lemonade in their neighborhood to support a cure for SMA and raised nearly \$20 in honor of Caitlin's brother, Owen, SMA Type II, age 6.

Brooks Bahrenburg of Shelburne, VT held an impromptu lemonade stand this summer in honor of her cousin, Skylar Bahrenburg. Her efforts helped raise \$10 to support Families of SMA.



**Sara Greene raising SMA Awareness**



**Makayla Adams bike decorating contest**



**Bri Johnson on the bus on her first day of school.**



**Brett Koehler and friends**



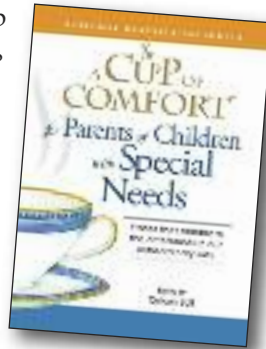
**Student of the month**

## Book for parents of special needs kids features child with SMA

A new book in the national bestselling Cup of Comfort series is being released May 12, 2009 and may interest FSMA families. "A Cup of Comfort for Parents of Children With Special Needs," published by Adams Media, a division of F+W Media, Inc., Avon, MA, contains a story written by Deb Wuethrich, whose daughter, Michele, had SMA.

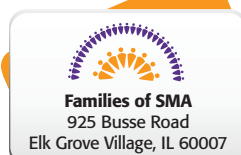
The narrative essay, entitled, "The Gorilla Wore Roller Skates" relates how Michele, through her own ingenuity, once turned her wheelchair into a motorcycle and herself into "The Fonz" and won a costume competition at a local mall one year. Michele passed away in 1982 at the age of 11. Her mom, who is a staff writer for The Tecumseh Herald, a weekly newspaper in Tecumseh, Michigan, says that she has been told that the story of Michele and the roller-skating gorilla has given some readers a new and more positive perspective about Halloween. "As a writer, I have the privilege of sharing memories of Michele and her courage and zest for life, even today," said Deb. "Michele would be so proud to know that her encounter with the gorilla is still being shared more than 25 years since it took place." Deb and Michele's dad, Gordy, live in Adrian, Michigan, and sometimes drop in at the Michigan SMA chapter's Walk and Roll event.

The book, retailing at \$9.95, will be available at bookstores and online stores, including amazon.com and barnesandnoble.com, and also at [www.cupofcomfort.com](http://www.cupofcomfort.com).



**Bella Andrade and Congressman Eric Paulsen**

Please note that as of **October 1st**, Families of SMA has a new mailing address. The address is:



## Student Places 3rd at National Tournament

Michael Pudlow a 2009 graduate of Munster High School in Munster, IN, who has SMA Type II, placed third in the Student Congress House of Representatives at the 2009 National Forensic League (NFL) National Tournament. Student Congress encourages individual debates in group format as students write legislation and speak on the legislation using proper parliamentary procedure. Over 3500 students from all 50 states competed in 10 main events. This year's NFL National Tournament was held in Birmingham Alabama, June 14-19.

Michael also earned the "Show Me Excellence Award" bestowed to students who qualified for the National Tournament during each of their four years in high school. This year, only 18 students received this honor. Michael was coached by Glen Percifield an English teacher from Munster High School who coaches both Student Congress and Public Forum Debate. Mr. Percifield's dedication to his students and hard work really paid off this year, qualifying two students for the National Tournaments. For those of you that are unfamiliar with the National Forensic League, it is the nation's oldest and largest speech and debate honor society. **Congratulation's Mike on a job well done.**



Dear FSMA,

My granddaughter, Stella Turnbull, has Type I SMA. I appreciate the support you've given their family. Thank you so much for helping the families who experience SMA to make heartfelt connections with one another. I appreciate the encouragement and hope you've given to everyone who has been touched by this devastating disease.

When I think about Families that experience SMA, I think of this quote: "Courage doesn't always roar. Sometimes courage is the quiet voice at the end of the day that says, 'I will try again tomorrow'."

Sincerely,

**Maureen McLaughlin  
Sperry, IA**

*Hi Guys,*

I live in Scotland UK and have SMA II – I am also age 31 and have a lovely fiancée and together we have 2 kids; Jai (5) & Jada (17 months).

A few years ago I started a care company as my care needs weren't being met by local organizations – I spotted a niche in the market and grabbed it. My Doctor went on to advise the regulators I was physically unfit to manage a Company.

I proved them all wrong and 2 years later I have won a business innovation award and was a finalist in Best Company – I have over 70 staff and also provide 1800 care hours per week.

I write today to let People know that SMA doesn't mean the end, according to Docs I should have been dead 29 years ago – I hope this gives families and kids a boost to look forward and positively at the future.

Thank you,

**Jon Fleming  
Glasgow, Scotland**

*Dear Families of SMA,*

My name is Chloe, I'm 18 and I live in England. I suffer from SMA severe intermediate, as does my older brother, Tom.

I am studying at college and as part of my course I felt it necessary to bring to light some of the issues I have continually found with the lack of adapted clothing available for disabled people. I have found it almost impossible to find clothes that fit me, that are comfortable and most importantly that look good!

Being a teenage girl I obviously want to look my best and achieve as much normality as I can with my physical disabilities and to "fit in". All my friends look immaculate with their skinny jeans and nicely fitted tops, however I'm stuck wearing baggy trouser (pants) and tops to fit comfortably whilst sitting all day in my chair and that are not to bulky under my belt. Whilst shopping in Topshop and all the usual high street stores I find myself embarrassingly having to look at the maternity department in order to find trousers to fit. The problem I find is that trousers do not have enough room in them to pull up fully and tops are not long enough to tuck down my back. Also tops that are fitted do not have the right shape in them to accommodate for the curvature in my spine.

England is generally quite cold and we hardly ever have a summer. I dread the winter as I can never find a coat or jacket suitable for my needs. Jackets are either too thick and restrict my already limited movement or are inconvenient due to having to undo my belt to take them off and on.

As my condition develops and physical implications of SMA are now more visible I have become very self conscious about the way my head tilts and the position of my neck. This is why I turned to the internet for a solution to find anything that could disguise it and I found that all adaptive clothing was not only extortionate in price but also quite insulting in the range that they had. Admittedly the clothes would fit and may be comfortable to wear however I would not feel comfortable in myself. To me they seemed to accentuate the physical disabilities that I already have and not minimize them.

So surely there must be another answer?

This is why I am asking for all people who suffer from the same problems to email me. I would be interested to hear your personal experiences (both male and female), your opinions and any tips or solutions you may have. I feel by receiving such information, steps forward can be made to widen the range of resources available for those who find this a big issue in their life.

Many thanks,

**Chloe Bellotti  
Suffolk, England  
clo\_1990uk@yahoo.co.uk**

*Dear Families of SMA,*

We are writing to thank the Strong family for submitting the article, Gwendolyn's PVC Play Apparatus in the Spring 2009 issue.

The play apparatus is a hit with our daughter Abigail, who also has Type I-SMA. As a surprise for her 11 month birthday in June, Abby's grandpa made her one following the basic dimensions provided in the article. And her Grandma added Strawberry Shortcake (her favorite) fabric to decorate the PVC pipes. She just LOVES her play gym. It is the one thing besides her Strawberry Shortcake DVD's that keeps her engaged for a long period of time. She especially enjoys ringing the wind chimes and jingling the bells that we suspended from the first bar. As parents we love it too, because she is playing and learning independently!

So thank you very much for sharing this wonderful idea so other families could benefit.

With appreciation,

**Steven, Laurie & Abigail Judge**





Dear FSMA,

Roman was born on May 11th 2007; he was 8lbs 4ozs and 21.25 inches long. When he was just 4 weeks old I noticed he was more floppy than most babies so I spoke with his pediatrician. The doctor sent a referral to Cincinnati Children's neurologist and we made an appointment for July 12th. So we just waited for our appt.

Sunday July 8th I was feeding Roman at about 3:45 and something was different, Roman lost his latch completely which had never happened. When I pulled him back he was not breathing and started to look blue, first instinct I freaked out, screamed and handed him to my mom in hopes that she would know what to do. In that situation you can't completely comprehend what is going on. She wasn't quite sure what to do either so I took him back and put my mouth over his mouth and nose and began to give him breaths. As my mom was calling 911 it was not working like I had hoped it would at first but after about the 6th breath he kind of coughed and he started to breathe again. Never in my life did I ever think that I would have to save my own child's life. In that moment I learned never to take life for granted. The ambulance showed up, as they are questioning what had happened I just held him, he was like a puddle in my lap, I don't even recall everything they were asking, all I could think of was to call Matt (my husband) and getting to a hospital to find out why this had happened. Not even prepared for what we were going to find out within days of this. We got to St. Luke West and they had him on oxygen and took all kinds of blood once he was stable they called Cincinnati Children's transport team. Once they showed up they got him situated in a baby gurney and had this itty bitty nasal canula to oxygenate him, and all kinds of monitors on the way to the hospital. Once we got to Children's they did x-rays, took blood and continued monitoring. Monday they continued to run tests, take blood, and come up with



some sort of explanation. There was Dr. Wong who kept on talking about SMA, Spinal Muscular Atrophy, she was very pushy on this diagnosis. Matt and I cried at the thought that our son had this genetic disorder that is the leading genetic killer in children up to 2 years old.

They took more blood to check for this and it would take 2 weeks to get a definite answer. He would have a feeding tube put in through his nose and through his stomach into his intestines. We had to learn so many things in these 5 days just to be able to take care of our baby.

My son was then pretty much diagnosed with Spinal Muscular Atrophy (SMA), a motor neuron disease, during that week. The motor neurons affect the voluntary muscles that are used for activities such as crawling, walking, head and neck control, and swallowing. It is a relatively common 'rare disorder': approximately 1 in 6000 babies born are affected, and about 1 in 40 people are genetic carriers.

Dr. Wong said that Roman was more than likely to be a SMA type 1. Type 1

SMA is also called Werdnig-Hoffmann Disease. The diagnosis of children with this type is usually made before 6 months of age and in the majority of cases the diagnosis is made before 3 months of age.

Due to this, the lungs may not fully develop, the cough is very weak, and it may be difficult to take deep enough breaths while sleeping to maintain normal oxygen and carbon dioxide levels. We went home on the 13th, and had our definite answer that my son had this horrifying disease.

On the 26th Roman's O2 sats went down to the 60s and required more oxygen than we were told to use, even after doing the Cough Assist and suctioning several times so I called Dr. Wong's nurse and pulmonary nurse, they called me back and said definitely to come back in so they could check him out. While in the ER they ran a couple of tests and did a couple of x-rays. The x-rays looked ok but showed some atelectasis (collapse), checked his CO2 levels they were in the high 60s and they should be somewhere between 35 and 45. So they put him on high airflow with oxygen. We were then admitted to the PICU. Dr. Wong came up and told them they needed to get him on bi-pap, the protocol for SMA children. So they found a really small mask to fit his face. It was big and we had to tweak it just a little bit but it would work for the time being. We were waiting for his g-tube surgery on August 12th. We had one week to go when things started to get bad again. They had maxed out on the pressures of the bi-pap and his CO2 level hit 82. They gave us a choice to either let him go or trach him so we went for the trach and he had both surgeries on August 17th. That ended up being the best decision ever! We stayed in the hospital to learn how to take care of our son and wait to get nursing to help us with Roman once we were home. We were terrified at what we would have to learn and the thought that we were going to have to learn how to do a trach change and basically become nurses and

respiratory therapists other than just being parents, was very scary.

No one ever prepares you that when you become a parent that there is a possibility of all of this. I was so amazed that it all seemed to come naturally for my mom, my husband and for myself. We had pretty much done within the first month and the rest of the time was just a waiting game. If it were not for the staff at the hospital I don't know if I could have made it for the total of 96 days straight that we had to stay at that hospital. On October 30th we got to take our now 6 month old son home! That was the best feeling ever. We had a great holiday season despite a 2 day stay just before Christmas, Roman had gotten an infection that most trached kids get at some point but that stay was so much better than the last. Roman only had one other stay through the winter which was so amazing after what we have heard from all the other families we were so grateful. Roman's first birthday was great and of course we did it up big! Over the summer we got the chance to go to Boston for the FSMA national conference. We met so many other families and kids affected by this disease and I was blown away by the kids and adults that face this everyday they are the most amazing people I have ever met! We had a great summer.

In the end of July we had our follow up appointment with Dr. Wong and we told her we were concerned about Roman's right eye it was almost all the way closed while the other was starting to squint. So she ordered us to have a MRI, we would get the results in a week or so. Roman did great during the test he even fell asleep. We got a call about 8 days later and I was a nervous wreck and the tone of his voice made it even worse. He told us that Roman had fluid around his brain and a slightly atrophied brain; I was mortified how this could happen to him after he had

already been through so much. We had to schedule to meet with a neurosurgeon, which took us a few weeks to be able to get in with. I was so worried about all this I had to keep reminding myself that they had told me that it was not life threatening and he was going to get through this. He had already been so strong to get through the rest I had to be positive about this! When Dr. Maugans came in I think my heart may have skipped a beat. He seemed very worried and more confused than I thought he would be. He wanted to know why it took us so long to come to him after the MRI. I assured him that had we been able to get in sooner, we would have. So he showed us the picture and it was not looking good, he then had us schedule a CT scan and wanted to see us right after, and he wanted us to see an Ophthalmologist sometime before coming back to him to make sure there was no hemorrhaging behind his eyes. We made our appointment for his eyes and the CT scan for the next week and then a follow up with Dr. Maugans. His eye appointment went great and the only thing was that he had very slight nearsightedness, and they would just follow up every 6 months to check it. Roman also did great with the CT and then we went up to neurosurgery. He said there was no change and that was good that it had not gotten worse but that he wanted us to have another CT in two months and then come back right after to see him again. So in December we had another CT and again Roman was great, so back up to neuro and find out the results, this time there was good news. The fluid was now gone. His brain was still slightly atrophied but would hopefully improve over time. We would just follow up in a year with another scan to make sure there would still be no change.

Thank you so much,

**Susan Anderson**  
Elsmere, KY



We are a non-profit, 501(c)3 tax exempt organization. Funds will be specifically directed to scientific, educational or literary purposes in keeping with a charitable organization. The organization is proud to provide funding to the FSMA newsletter and website, which provide information and networking opportunities to its members. We are a volunteer-driven organization.

## Submissions

To submit articles or make other contributions to our newsletter, please contact us at: [newsletter@fsma.org](mailto:newsletter@fsma.org).

Digital images are encouraged! Send your digital pictures to: [newsletter@fsma.org](mailto:newsletter@fsma.org)

## Change of address

Send changes, including ZIP code to: [info@fsma.org](mailto:info@fsma.org) or call 1-800/886-1762 or mail to: FSMA, Membership, 925 Busse Road, Elk Grove Village, IL 60007

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FSMA does not support or endorse any particular treatment or therapy. Information contained in this newsletter should not be used as a substitute for consultation with a qualified healthcare professional.

Please note that as of **October 1st**, Families of SMA has a new mailing address. The address is:





# Courtney's Adventure

By Linda Rosas, Courtney's Grandmother

Courtney Brooke Rosas has added another amazing adventure to her little book of life, she got to go fishing! What a day we had, fishing, riding a boat, meeting new people and being with friends. You wouldn't think that someone who owns a million dollar home would be willing to share their wealth with a bunch of special needs kids and their families that they didn't even know, but they did. The home belongs to Lion Pat McPherson and the Conroe Noon Lions Club sponsors the kids on the lake event that we were blessed to be able to attend. We didn't even have to bait our own hooks or remove the fish from the hooks once they were caught. Lunch was provided, grilled burgers and hot dogs with chips along with all the ice cold sodas and water you could want. There were all kinds of desserts and snacks also. Someone would come and ask us if they could get us something to drink or if we needed anything. We were treated like we were royalty. Sack races were held on the lush green lawn along with face painting and water guns and the lake water came right up to the grass where you could fish or go over to the covered pier to be out of the sun. The United States and Texas flag were flying high on the lawn sort of waving to us as I looked up at the beautiful sky.

For those of you who don't know us, Courtney and her best friend Hayron have Spinal Muscular Atrophy type I. We try to let Courtney experience as many things as she can, at least once. She and Hayron have been friends since Courtney's 4th birthday. The Merida family and our family try to get together pretty often, sometimes just at home and sometimes we take them on outings. We think it is important for Courtney to spend time with Hayron so she will know that she is not the only child in the world like her. Hayron's little sister Gretel and Courtney's little sister Kendall play together too.

Candace was holding Courtney's fishing pole for her and caught a cat fish for her. Courtney was all smiles as the nice man helping us held the fish up so she could touch it. Later on Kendall caught 2 cat fish, she was so proud of herself. Hayron and his family caught a fish

too. We thought Courtney and Hayron would not be able to ride one of the boats because there wasn't enough room for their wheelchair/strollers to roll inside. I had told Candace to turn Courtney around so she wouldn't be able to see Kendall getting on a boat because I knew Courtney would start to cry. For the past few months Courtney has started crying because she can't do things that her little sister can do, like riding a train or a pony. It just breaks my heart when she cries. There was a lady that was at the boats and she told us to stop and wait that there was a bigger boat coming and that Courtney and Hayron will ride that boat, for us not to worry. Just as the boat arrived the lady had gone to get four strong men to lift Courtney and her wheelchair/stroller into the boat, then they lifted Hayron into the boat also. I started to get tears in my eyes because I was so happy for these two special children. Courtney was all smiles as we started our wonderful boat ride, Candace had lifted Courtney's head a little so she could see the water and also look across the boat at her best friend Hayron. We could have stayed on that boat all day long and our wonderful boat driver, Captain Brian, would have been glad to let us but there was a long line of children back on the pier waiting to ride the boat.



As we were leaving the girls each got a pail filled with goodies and a trophy. They were so proud of those trophies, Kendall told us, "See I'm a winner and look there's my name on my trophy." The trophy said kids on the lake 2009 and Conroe Noon Lions Club but I wasn't going to tell her any different. I told her yes I do see your name and I am so proud of you, you and your sissy are such good fisher girls!

I want to thank the Conroe Noon Lions Club for our amazing day, filled with fun, sun, boat rides and fishing, memories we shall cherish forever. During the whole day, our thoughts were all good, filled with joy and happiness. I have to say this was such a relaxing day and we were treated so well, I almost forgot that we have a child with special needs, it was like our children were just like any other kids on the lake!





## The Jeffrey Journey

by Helen Baldwin



In 1997, more than a few of our props were knocked loose upon hearing the news that we'd be welcoming a third baby. They were knocked away two months after his arrival upon hearing the diagnosis (SMA, Type 1) and prognosis.

We had a mere 3½ more months with Jeffrey in the earthly sense, but it is an understatement to say that his impact has continued with gusto ever since. While countless others are organizing wildly successful fundraisers, heading up chapters, and/or birthing foundations, I chose to write a book.

As I was dwindling down on the very first proof of *The Jeffrey Journey* in 2003, I began making a list of those I wanted to include in the dedication. It took only a few minutes to realize it would be impossible to determine a cutoff point, so the thought came to include a special dedication... one that would include everyone in the SMA family.

Over the years, the special dedication has become perhaps more of an obsession than the book itself! The number of names has grown to over 1000 for the upcoming revision this fall, and I have even tiptoed into unlikely territory with a Facebook page to help spread the word to SMA families. I have searched in myriad nooks and crannies for

names and contacts for the past six years, and while I've connected with many, it's disappointing not to connect with all!

The dedication is both emotionally mindboggling and oddly therapeutic. It's not often that so many names are listed together, and while we each struggle with SMA to some extent, it is comforting to see proof that we truly are in this together.

I am always taking names for the dedication. If you are interested in submitting a name, all I need is the name as you would like it listed, including a nickname if desired, and whether it belongs in the 'honor' (living) or memorial section.

There are no restrictions regarding the dedication, such as location, date, age, type, number of names, etc. And if you are the one with SMA, please do not hesitate to include your own name!

I'm also doing a blog with good friend/fellow SMA mom, Cindy Schaefer, whose son, Kevin (Type 2), is a sophomore in high school. We have a link from the blog to the SMA Families page comprised of families' web sites and blogs; in addition, we spotlight SMA folks on occasion in our blog posts. If you're interested in being included on the SMA Families page and/or spotlighted in a blog post, please contact us!!!

**Thanks so much -  
Helen Baldwin**  
jeffreyb@skybest.com  
thejeffreyjourney.com



*Hello to all,*

I just wanted to drop a line and bring you up to speed on Morgan (type 3). She graduated from Scranton Prep this year. Some of her accomplishments include being President of the National Honors Society, being on mock trial 4 years, going to state level competition two times, and being featured in the Scranton Times graduation article. At graduation she received the final and most prestigious award given to a student, followed by a five minute standing ovation. She had a wonderful high school experience.

A new chapter opens in her life as I type this to you. She arrived in Palo Alto, CA on Monday. She is going to major in biology at Stanford University. So we finally have one of our own attending a University like Stanford, who is going to be key on stem cell research and hopefully make a difference in the fight against SMA.

**Kindest Regards,  
Tom Duffy**  
Scranton, PA

## Congratulations



■ Congratulations Mary Jane Utzat on the birth of her daughter Sydney Ann.



■ Congratulations Tonya, David and Grant Willingham on the birth of their twin daughters and sisters Julia and Hannah.

# FAMILIES



**The Murray Family**



**The Imhoff Family**



**The Damon family**



**The Gilley Family**



**The Turnbull family**



**The Cuevas family with  
Derek Geter**



**The Nelson Family**



**The Sykora Family**



**The Campbell Family**



**Jack, Lise Connor and  
Caitlin Murphy**



**Madeleine, Valentina and Andrea Puddu**



**The Gellner family**



# SHARING page

www.curesma.org



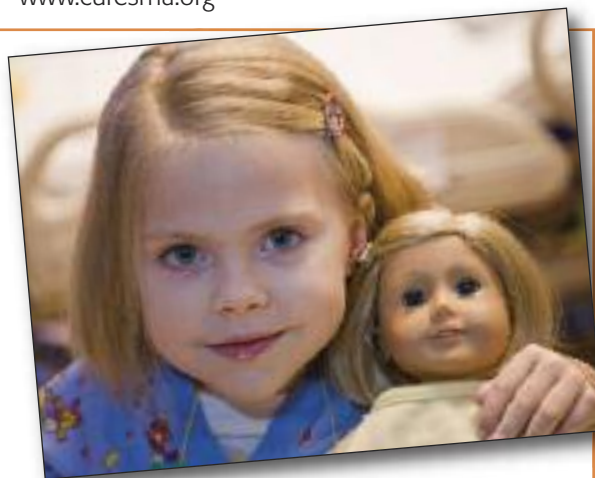
**Ahmad Amsyar Qahhar**



**Amira George**



**Aubriana Navarro**



**Bella Andrade**



**Abigail Judge**



**Dr Schroth, Lizzy Hallam and Lisa Pharo**



**Gabriella Aleman**



**Isabella Nicole Kurek**



**Nora Gooden and her Mom**



**Bri Johnson**



**Gracie Degraffenreid**



**Kayden Wentz**



**Jerika Bolen**



**Jacob Goodson**



## ONLINE FUNDRAISING



### Michele & Douglas Erwin

Douglas and Michele Erwin of Massapequa, NY organized the 2nd Annual NYC 1/2 Marathon for SMA in honor of their son, Greyson. Douglas and over 20 other runners ran the NYC 1/2 Marathon on August 16th to raise money for SMA research and to raise awareness of SMA. To date, Greyson's 2009 NYC 1/2 Marathon Team has raised \$80,147 to benefit SMA research!!

*"Welcome to Greyson's 2009 NYC 1/2 Marathon fundraising Home Page!*

*This year we will be running our 2nd annual event on behalf of Greyson for*



*Families of SMA. Thank you to all of those who supported us last year and made last year's event so successful. We are asking you to consider making another donation to our cause. For those who may be new to our cause, our son Greyson was diagnosed in the winter of 2007 with Spinal Muscular Atrophy (SMA), a genetic illness.*

*An update on Greyson, he is now 2 1/2 years old. This past winter Greyson had suffered two back to back bouts of pneumonia one of which he was admitted to the hospital and stayed 4 days in the PICU. Leaving no choice but for mom to make the best decision and become a stay at home mom and advocate for Greyson. He now is getting all the much deserved and needed TLC. Greyson has received his highly anticipated power chair this spring and is doing fantastically well in learning how to maneuver through the house and community. Greyson has two new friends who visit him every night, his nurses Violet and Mohamed. They have been a wonderful addition to Greyson's routine, Greyson routinely tells them to "go away". Mommy and Daddy can now get some much needed sleep without the worry.*

*Just like last year, Douglas and over 20 other runners will be running the NYC 1/2 marathon on August 16th on behalf of Greyson. All of the runners, along with Phillips Van Heusen (PVH), are fundraising to help find a cure. All money raised will go directly to Families of SMA and Columbia Presbyterians SMA Research Clinic. In addition to personal donations we are also welcoming corporate sponsorship, if your company would like to make a charitable financial donation or a product donation as gifts to our runners for their efforts in raising awareness please contact us for details.*

*This year our run and all proceeds will be donated in memory of Greyson's friend Max Rubenstein who passed away this winter 2 months before his fourth birthday. We miss him.*

*The site is dedicated to Greyson, and the thousands*

*like him that fight everyday against this terrible illness. It is our goal to raise awareness and funds to find a cure. Please help us reach our goal. Thank you! The Erwin Family"*



### Vincent & Catherine Gaynor

Vincent and Catherine Gaynor of Wantagh, NY set up an online Gift Registry on the FSMA website in honor of their daughter, Sophia. Their goal is to raise \$100,000, specifically for stem cell research. In a few short months the Gaynor family has been able to raise \$76,729.44!

*"Welcome to Sophia's Cure 2009 Home Page!*

*We have set up this gift registry page to help support a cause near to our hearts. Sophia was born on February 27, 2009. Shortly after we found out that she has Spinal Muscular Atrophy (SMA Type I), a rare terminal genetic disease that attacks the nerves. There is no cure right now, but Families of SMA is doing extensive research including on Stem Cells for finding a cure. Without a cure Sophia's muscles will slowly degenerate and she will no longer be able to eat or breathe on her own.*

*We are asking our friends and family to make a donation to help Families of SMA. These gifts will be recognized in honor of Sophia and specifically dedicated to stem cell research.*

*Please help us support this extremely worthy cause.*

*Thank you!"*



## Unite for a Cure

Unite for a Cure has raised an amazing \$41,412.34 so far!

*"This is the donation page for Unite For The Cure. This campaign was started by families impacted by Spinal Muscular Atrophy (SMA), the #1 genetic killer of young children, as a way to unite together to raise \$100,000 to help the Families of SMA stem cell program get to human clinical trials."*

Thank you to the following fantastic family fundraising teams who have each raised the following amounts:

Marshall Family \$8,605; Schmid Family \$8,019; Lunt Family \$5,390; Pringle Family \$4,485; Gustafson Family \$4,315; Butler Family \$2,704.60; Lucas Family \$2,610.00; Unknown \$1,663.74; Coleman Family \$1,100; Gooden Family \$870; Wright/Beasley Family \$785; Greene Family \$525; Ragland Family \$200; Lovelace Family \$60; Strong Family \$50; Potter/Reilly Family \$30.



## Gene Trainor

Gene Trainor of Reisterstown, MD ran his first ever Sprint Triathlon on May 31st and asked family and friends to support his efforts by donating to Families of SMA. Gene set up an online Gift Registry through the FSMA website and

raised a remarkable \$14,205!

*"Welcome to Gene's 2009 Sprint Triathlon Home Page!*

*Dear Family, Friends and Work Associates:*

*Over the past 2 months, with the help from my good friend Jeff Handy (Thomas Weisel Partners) I have been on a health and fitness crusade to lose weight and get in shape. My goal has been to participate in my first Sprint Triathlon on May 31st! I am happy to report that I have lost 29 pounds (as of May 8th) with 8 more to go (getting back to pre-marriage weight)!*

*I would like to do this triathlon while also creating awareness for Spinal Muscular Atrophy and raise money for Families of SMA. As many of you know, Barbara and I have spent the past 15 years (after Erin's death) working with this wonderful organization to raise much needed funds for research, family support and education.*

*If I can achieve my fundraising goal, I promise that I will not come in last place and live to talk about my first triathlon!*

*We hope that you will help us support this extremely worthy cause.*

*Many thanks!"*



## Paul Kapellas

Paul Kapellas enjoys running marathons and for the second year he is "Running for Cru", a friend's baby boy who lost his battle with SMA in September 2008. Congratulations to Paul who has raised \$8,472 so far this year!



## Ride for Sophia's Cure for SMA

On September 15th three men Ryan Burkett, Dennis Edison and Michael Grahlf will gear up to ride 3,500 miles across the country to try and raise money to help fund research for a cure. The ride will kick off in Wantagh, NY and conclude in San Francisco, CA. The riders plan to bike approximately 100 miles per day and arrive in San Francisco around early November. In place of gifts Riders are asking their friends and family to make a donation to help Families of SMA.

So far the team has raised \$6,860!



## Melissa House

Congratulations to the House family for raising more than \$6,108 to help Families of SMA find a cure. Sponsors, friends and family put Melissa over the top during this year's wonderful efforts – great job!



**Thank You to the following:****Neal & Rebekka Mastin, \$3,500.**

We began celebrating Ollie's birthday every month on the 5th. Sadly, we will not celebrate his first birthday. Ollie lost his battle with SMA on August 3rd - just two days shy of his first birthday. In lieu of flowers, we are asking friends and family to make donations to help Families of SMA. Please help support this extremely worthy cause. Thank you! Neil & Bekka.

**In Memory of Brendon Dupree, \$1,010.**

We have set up this gift registry page to help support a cause near to our hearts.

**Marla & Lee Marlow, \$1,135.**

We are hoping to see everyone at the Walk-N-Roll on September 12 in Littleton; however, if you cannot attend, we would sincerely appreciate any donations made in memory of Jay Marlow to help Families of SMA reach their goal of finding a cure for this very cruel disease.

**Rita & Todd Lott, \$820.**

We have set up "Jordan's World" registry page to help support a cause near and dear to our hearts.

**Joseph & Jenny Imhoff, \$400.**

In lieu of holding the Rally for Reagan golf tournament due to Reagan's spinal surgery, we set up a gift registry page.

**Joseph Szumigalski, \$320.**

We are asking that you please make a donation of any size to help Joseph and his family along with other Families of SMA.

**Tyler Reed, \$245.**

I am competing in the IronGirl Columbia Triathlon. With these types of events, there are so many causes to support, but I want to support a cause that is a part of my family's life today. I am asking friends and family to make a donation to help Families of SMA.

**Dani Doornerbal, \$225.**

In place of birthday gifts I am asking friends and family to make a donation to help Families of SMA.

**Bradford Flater, \$195.**

In place of gifts we are asking our friends and family to make a donation to help Families of SMA.

**Michelle Moore, \$160.**

On Saturday, August 1, 2009, we are participating in the 6th Annual SMArt Walk for a Cure at Beaver Island State Park in Grand Island along with the rest of our family.

**Sarah Varley, \$95.**

In place of birthday gifts I am asking friends and family to make a donation to help Families of SMA.

**Desiree Goodman, \$45.**

In place of gifts we are asking our friends and family to make a donation to help Families of SMA.

## Families of Spinal Muscular Atrophy Welcomes New Director of Development.

Families of Spinal Muscular Atrophy is pleased to announce that Tim Walker has joined the organization in the role of Director of Development.

As a mission driven development professional Tim has worked for more than 12 years at a variety of nonprofits including Youth Service Project, The Chicago Lighthouse, and the Center for Economic Progress. Tim is a Rotarian who holds a degree from Illinois State University and has received additional training at The Fundraising School at Indiana University and the Center on Nonprofit Management at the Kellogg School of Management in Chicago. In addition, Tim has been an active member of the AFP (Association of Fundraising Professionals) for more than a decade. Tim lives in Brookfield, IL with his wife and two children.

In this new role of Development Director for Families of SMA, Tim will focus on building the volunteer support at the 27 FSMA chapters across the country, supporting individual family fundraising events, as well as growing national and local partnerships with corporations and clubs.

You can reach Tim at (800) 886-1762 or (708) 743-4120, or at [tim@fsma.org](mailto:tim@fsma.org)



# SHARING page

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The Rappoport kids



Ray and Ethan Fantel



Ethan Hammock



Lizzy Hallam

  
**Families of SMA**  
*Research. Support. Hope.*



Malorie Fox



Peter Henkel



Roman Walker  
Anderson



Ryan White



Ryan White and Family



McKenzie Damon with her  
brother and sister



Philip Struble



Kendall Buss



Hannah Willingham



Reagan Imhoff



Jolie Ava Dziubela



Leah Jean Miller



Mary Nelson



Luana Arieira Fernandes de Araujo



Right from the start, Families of SMA was always about community. It was about families and researchers coming together to help everyone. It was always about each one of us making sacrifices for the greater good of the whole community.

**Audrey Lewis, the founder of Families of SMA, created that sense of community in our organization 25 years ago.** The critical research breakthroughs, the drug discovery programs, the annual conference, the clinical trials - would never have been possible without her foresight, her strength and her determination. Her foresight was that the community had a responsibility in carrying the weight of the cost of research.

Early on, Audrey realized that one of the keys to making progress towards a treatment for SMA was attracting talented researchers to the field. Commitments to funding basic research and an open collaborative approach yielded results as scientists began to view SMA as a stable and positive area in which to dedicate their careers.

Additionally, Audrey further understood the crucial component that both researchers and families had to work closely together as a team on a regular basis. To this end the unique annual Families and Professionals Conference was created, and the Chapter structure for the organization formed. Audrey was also an innovator in the field of research foundations with one of the very first non-profit commercial drug discovery collaborations ever attempted.

Her commitment, passion, unwavering strength and focus were a constant for Families of SMA as its Executive Director and Chairman of the Board of Directors from the moment of inception back in 1984.

Through Audrey's commitment, passion and leadership, FSMA's membership has grown to over 65,000 strong, and Audrey continues to be an active member of our Board of Directors.



To help commemorate the 25th Anniversary of Families of SMA we are pleased to announce a new grant award in honor of Audrey Lewis, the founder of FSMA. This new award will be called the

### ***“Audrey Lewis Young Investigator Award”***

The Audrey Lewis Young Investigator Award will be presented to a newly independent principle investigator who is in the first 5 years of running a laboratory.

This grant will be awarded every other year, beginning in 2010. The award will provide funding for a three-year period with a maximum budget of \$100,000 per year. Applications will be accepted during the FSMA grant submission process each September.

One of the significant goals Audrey Lewis had when starting Families of SMA 25 years ago was to attract new and talented researchers to the SMA field. We hope this legacy continues to be built with this new grant award. The award is intended to make a positive impact on the early phase of a talented researcher's career by enabling them to focus on the SMA field.





Adrian R. Krainer, Ph.D.  
As a research scientist, I try to understand the nuts and bolts of how genes are expressed, and to devise effective ways to apply this knowledge towards a cure for SMA.

*Dr. Krainer is the St. Giles Foundation Professor of Molecular Genetics at Cold Spring Harbor Laboratory, in Long Island, NY. His lab studies the basic mechanisms and regulation of human pre-mRNA splicing, as well as the involvement of this cellular process in genetic diseases and cancer. He is developing mechanism-based methods to correct the genetic defect in SMA, and is a member of FSMA's Scientific Advisory Board.*

**Who am I?**



**Why did I become active in SMA?**

I first heard about SMA when I was invited to a workshop at NINDS shortly before the seminal Lorson et al paper in PNAS was published in 1999. The work was discussed there, and showed that a single nucleotide change in *SMN2* caused skipping of exon 7; this meant that correcting the splicing defect could be the way to cure this disease. As it happened, the basic research that my lab was already doing on splicing mutations in other genes, such as *BRCAL*, converged remarkably well with these findings. With much encouragement from Alex MacKenzie, Arthur Burghes, and others, I jumped into the SMA field shortly thereafter. I could sense right away that there was something special about the SMA research, clinical, and patient/family community, which really attracted me to this field.

**What is my contribution?**

The first thing we did was to develop a cell-free system to study how exon 7 is recognized during pre-mRNA splicing. This system has been helpful both to screen and characterize compounds that act directly on the splicing machinery to correct *SMN2* splicing, and to investigate what splicing factors are responsible for the differential recognition of exon 7 in *SMN1* and *SMN2*. We showed that the RNA-binding protein SF2/ASF is a key factor in this process.

We have also put a lot of effort into developing PNA-peptide conjugates and antisense oligonucleotides that can efficiently correct the splicing defect in *SMN2*. This approach is already working remarkably well in mice, and I am very hopeful that before long it will begin to be tested in clinical trials. I am lucky to have had very talented scientists in my group working on SMA, including Luca Cartegni (now at MSKCC), Michelle Hastings (now at Rosalind Franklin University), Yimin Hua, Kentaro Sahashi, and Ying Hsiu Liu, in addition to our close collaborators at Isis Pharmaceuticals and Paratek Pharmaceuticals.

**What do I like about FSMA?**

It's a long list. I love the annual meetings, especially the warm interactions with the kids and families. I have met fantastic people there, and these interactions really help us to stay motivated and focused when we go back to the lab. FSMA generously funded our collaboration with Paratek. It has truly been an honor to serve on the Scientific Advisory Board.

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*As part of our ongoing celebration of the 25th Anniversary of Families of SMA we will be spotlighting "25 Voices of SMA" throughout the year. These will be profiles of families, clinicians and researchers from the SMA community. The first voices are of The Trainor Family, Thomas Crawford, M.D., The Rappoport Family and Hans Keirstead, Ph.D.*

**25 Voices of SMA**  
John Kissel, M.D.



I am a neurologist and neuromuscular specialist with a special interest in all things SMA. I started out seeing mainly adults, but now care for patients of all ages with this disease, and enjoy every minute of it! As a "clinical researcher," I try to bring advances from the research laboratory into clinical practice, and look for the best ways to take care of patients with SMA. This means "leaving no stone unturned."

*Dr. Kissel is Professor of Neurology and Pediatrics at The Ohio State University Medical Center and Nationwide Children's Hospital in Columbus Ohio, where he is Director of the FSMA-funded SMA Clinic. He is a member of FSMA's Medical Advisory Board.*



**Why did I become active in SMA?**

How I decided to focus predominantly on SMA is a funny story! I was in Paris in the early 90s to give a talk (on another disease). At dinner, I sat next to one of the few other English-speaking presenters, a woman from the U.S. She asked if I ever saw adult patients with SMA. When I said yes, she immediately signed me up! The woman, of course, was Audrey Lewis, the founder of FSMA. Within a few months, I was fully recruited and giving talks to adults at the FSMA Annual Conference. From all over the country! Audrey really had a way of recruiting and getting things done! Since that time, my interest in SMA patients and their disease has grown exponentially, along with our knowledge of the disorder. SMA now takes up most of my time and attention, and I couldn't be happier about it! The challenges, rewards, and satisfaction in caring for patients with SMA and their families are special and unique. I'm totally hooked.

**What is my contribution?**

I am a doctor, a clinician who sees lots of patients, so I contribute a great deal on the clinical side of things. I have participated in many clinical trials over the years and am proud of my ability to design effective clinical trials that test various therapies in SMA. We have an extensive group of basic science researchers in SMA here at Ohio State and at Nationwide Children's. I try to "bridge the gap" between the research side and actual patients in the clinic. As a member of FSMA's Medical Advisory Board, I participate actively in the Annual Family Conference and help in any way I can.

**What do I like about FSMA?**

FSMA has simply been indispensable in my work both on the clinical and research level. They have done an incredible job of bringing together hundreds of basic and clinical researchers from around the world for their Annual International SMA Research Meeting. They have funded all of our important clinical trials here at OSU. As a major force in moving SMA forward, FSMA gets the disease on everyone's agenda, including the politicians!

# 25 voices of SMA





**Who are we?**



We're the Lockwood-Merkle Family from Cincinnati Ohio, including Beth and Kevin Lockwood (parents), Elizabeth & Bob Lockwood (grandparents), and Jan & Ron Merkle (grandparents). We work as a team and are devoted to Emma (8) and Nicholas (6) Lockwood, who have SMA Type I.

*Beth is a former research assistant at Cincinnati Children's Hospital and tutor of children with autism. Kevin is a high school band director. They started the Ohio-Kentucky-Indiana "OKI" FSMA Chapter shortly after Nick was diagnosed. Elizabeth works with the Major Donor Committee for FSMA.*

**Why did we become active with FSMA and the chapters?**

In the beginning, this was a race against time. We heard "close to a cure" and "on the verge" and wanted to be part of it. We always wanted a cure to benefit all people with SMA, not just our family. Each of us, along with other families in the OKI Chapter, keeps the fundraisers going. Emma and Nicholas are two beautiful children with SMA, who have been extremely healthy using a non-invasive approach learned from Dr. Schroth in Madison, Wisconsin. The children are bright, proficient on their laptop computers, and drive power wheel chairs. They can get around and into trouble just like other kids! They play in the yard with a power soccer ball and even play baseball on the Miracle League baseball team! Taking care of them is a team effort for their parents and two sets of loving grandparents.

**What is our contribution to SMA?**

The OKI FSMA Chapter has raised over \$1 MILLION since its inception in 2002 through fundraisers and foundation donations. Elizabeth worked with a local TV news personality, Michael Flannery, who connected her with a generous anonymous donor. This year is OKI's 6<sup>th</sup> Annual Walk and Roll. Through the efforts of OKI families and friends, this event gets bigger and better every year! Beth has worked co-operatively with doctors at Cincinnati Children's Hospital to offer the non-invasive protocol and care for children with SMA. As co-presidents for the OKI Chapter, it is not uncommon for Kevin and Beth to be the first line of support to families with newly diagnosed children. Both Beth and Kevin continue to devote hours of time supporting local families.

**What do we like about FSMA?**

We realize now that a cure will take time, but we do not give up hope for a treatment or cure. Small changes and improvements would make a huge difference for Emma and Nick, and other children living with this disease. FSMA turned our hopelessness into hope for the future through new drug trials and stem cell research. Our faith is in FSMA because of their diversified research approach. They don't put all their research eggs in one basket! We choose to raise money for FSMA because 80% of the money we raise goes directly into SMA research. Finally, we are proud to be part of an organization that supports families nationwide who live everyday with the challenges of SMA.

25 voices of SMA

See FSMA Website for Full Profiles

25 Voices of SMA  
The Utzat Family



**Who are we?**



We are Mary Jane and Tim Utzat from New Jersey. Our daughter, Samantha Jane Utzat, passed away from SMA Type I on September 14, 2007, two days before her first birthday. We are expecting another daughter, Sydney Ann Utzat, this September.

*Mary Jane is an R.N., in a surgical intensive care unit and Tim works as a substation technician for an electric company.*

**Why did we become active with FSMA?**

When Samantha was diagnosed with SMA Type I, our goals were to keep her happy, comfortable, and to provide her with fun and playful childhood experiences. We focused on her abilities rather than her disabilities. We were thankful to receive two generous toy packages from the Rappoport and the Abraldes families. These included light toys appropriate for Samantha to play with and enjoy. To these gifts, we added toy ideas of our own and ideas from the FSMA website. We were contacted by and given contact information for, other parents within the FSMA community who had already gone down this path. Jessica Moyer and Shaina Rappoport, in particular, helped to answer many of our questions and provide support. We knew that, someday, we would like to give back to the FSMA community in a similar way.

**What is our contribution?**

Since Sam passed away, we have kept our promise to ourselves. We give back just as others gave to us. We assemble packages for other families that include the kinds of light toys that Sam enjoyed, with a list of play and other creative ideas for each child. We send these packages to FSMA and FSMA sends these out to all newly diagnosed Type I families who contact the organization. Close to one hundred of these packages go out each year. Tim's mother contributes hand-made blankets and rattles. We also donate the infant indoor/outdoor reclining swings, just like the one that Samantha enjoyed.

**What do we like about FSMA?**

What *don't* we like about FSMA? Really, nothing! We appreciate their tremendous support, including the information, contacts, and equipment. These came at a time when our family really needed them. Samantha received wonderful care from her SMA physician, Dr. Finkel, who is on the FSMA Medical Advisory Council. We are in awe of the contribution that families, who support this organization, make to raising funds for research. Last, but not least, the staff of FSMA couldn't be more pleasant and helpful. Our hope is that someday, when a treatment and a cure comes around, that this organization, as special as it is, won't be needed.



We're the Abraldes family. Alex, Donna, our Angel Deirdre and her sisters Maeve (9), Bridget (6) and Shannon (2). We live on Long Island in New York. Deirdre was born on February 5<sup>th</sup>, 1999 and after a year long mission on this earth she passed away on February 13<sup>th</sup> 2000 from SMA Type I with her family holding her gently as she continued on her journey. Her sisters all have a little bit of Deirdre in them and when we catch glimpses of her in them, we know that it's Deirdre's little way of letting us know she's always with us.

We've been members of the NY Chapter since 2000. Donna created the Care Package Program for Type I babies and is busy looking after the girls and all their activities and Alex is an Executive Director for Verizon.

Who are we?



**Why did we become active with FSMA?**

The family at FSMA gave us incredible support from the start when we received Deirdre's diagnosis. They connected us to devoted and compassionate doctors like Tom Crawford who gave so freely of his valuable time to answer all of our frenzied questions. We were connected with other parents who would become life long friends as they supported us through the worst of times. We became active with FSMA after Deirdre passed away to give back what FSMA, the doctors and families gave to us during our greatest need. We knew we wanted to continue Deirdre's mission since she touched so many people and to make her legacy a lasting one. We could not have made it a single day without them and knew we had to be there for all those families who would wake up one day to the horrible news and help them with advice, a shoulder to cry on or just a person to talk to who understood exactly how you were feeling.

**What is our contribution?**

At first, Donna acted as a support line for families who were recently diagnosed with a Type I child. Helping others in this way helped us in our grief and sense of loss. Despite our grief and the fact that we had a new baby to care for (our second daughter Maeve was born two weeks after Deirdre passed away), we resolved to hold a charity event to raise some funds for FSMA. We settled on a Dinner Dance for Deirdre in March 2001 and delivered a check to FSMA for nearly \$70,000. Not too shabby we thought on our first try! After that effort, Donna wanted to focus on a more tangible and immediate impact to Type I babies because their time is so short and we know that parents don't have time to think, only time to react and care for their babies. We created the Care Package Program in memory of Deirdre Abraldes. The care packages contained some of the toys that seemed to stimulate and entertain Deirdre the most, pillows to help the babies prop up and ideas on how to play with your babies. In 8 years, we've sent out over 450 packages with lots of help from family and most recently, Deirdre's own sisters joined in the project.

**What do we like about FSMA?**

Well, even after all these years, from the day we first spoke to Audrey to the gang at the office today, we feel very close to FSMA. They have our gratitude because they have always stayed true to caring deeply about the families who are suffering as a result of this terrible disease. FSMA means so much to us for their ability to show compassion and in recent years we have seen their efforts drive great hope and progress for treatment where there was none previously. Thanks to FSMA for making Deirdre's life touch so many babies and families for so long. We're truly grateful.

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25 Voices of SMA  
Brian Pollok, Ph.D.



I am a scientist and a businessman (two roles that are not always easy to combine) who takes the tools of modern biology, chemistry and engineering and applies them aggressively to disease research. My goal is to use new technologies to solve the challenges of historically underserved diseases like SMA.

*Dr. Pollok is Chief Scientific Officer of Life Technologies, a Fortune 600 company and one of the top 3 global companies devoted to supporting the career scientist. At Life Tech, he guides the Research and Development (R & D) activities of over 1200 scientists and engineers, with a budget of \$300M. He has played an important role in FSMA's first and ongoing drug development program, and is a member of FSMA's Scientific Advisory Board.*

**How did I become active in SMA?**

My first encounter with SMA research came at the Drug Discovery Technologies Conference in 1999 during a chance encounter with Dr. Arthur Burghes, a leading SMA research scientist. He was looking for companies capable of doing early-phase drug discovery programs for SMA, and stopped by a booth run by Aurora Biosciences, the company I worked for at the time. Arthur proceeded to educate and engage me in the genetics of SMA. We talked about how the disease could be altered with drugs and drug therapies. This dialogue ultimately led to a contract, in January of 2000, between FSMA and Aurora Biosciences to conduct the first drug discovery program focused solely on SMA.

**What is my contribution?**

I hand-picked two talented researchers, Dr. Jill Jarecki and Dr. Joan Chen, for this first SMA drug discovery program. As the core research team, they screened and tested over 1.2 million chemical compounds. I provided ongoing support and guidance to the program and made the decision to also screen the Vertex compound collection. This step was not in the original agreement, but yielded excellent results: it turned out to be the starting point for the Quinazolines drug research program at deCODE Genetics. In 2005, Jill and I had a conversation about how to find the protein target (the protein that the Quinazolines regulate to increase SMN2 expression). I suggested we use a special technology, from Life Technologies/Invitrogen, to "fish" for the target. Together with Mark Gurney, a VP at deCODE Genetics, we organized a multi-site team to carry out this expedition. In 2007, we "made the catch"—that is, we identified the human DcpS protein as the protein target. This is a critically important find for SMA drug discovery and provides a strategy for making more effective second generation drugs.

**What do I like about FSMA?**

The personal connection between SMA researchers and SMA families is inspiring. It motivates our scientists to work even harder for SMA. In 2002, I received a "thank you" check from an FSMA family for an Aurora team lunch. This simple gesture of appreciation was so touching. The families draw me in and make it easy for me to work hard for the disease, and for FSMA's Scientific Advisory Board.

25 voices of SMA

## Genzyme Genetics Publishes a Paper Showing Carrier Rates for Spinal Muscular Atrophy.

The paper entitled “Differences in SMN1 allele frequencies among ethnic groups within North America” indicates that although there is a carrier rate between 1 in 25 and 1 in 50 in the general population, this can differ significantly among ethnic groups.

SMA is considered to be a pan-ethnic disease. However, carrier frequencies for many ethnicities, including most ethnic groups in North America, are unknown. To provide an accurate assessment of SMN1 mutation carrier frequencies in African American, Ashkenazi Jewish, Asian, Caucasian, and Hispanic populations, more than 1,000 specimens in each ethnic group were tested using a clinically validated, quantitative real-time PCR assay that measures exon 7 copy number.

The observed one-copy genotype frequency was 1 in 37 (2.7%) in Caucasian, 1 in 46 (2.2%) in Ashkenazi Jewish, 1 in 56 (1.8%) in Asian, 1 in 91 (1.1%) in African American, and 1 in 125 (0.8%) in Hispanic specimens. Additionally, an unusually high frequency of alleles with multiple copies of SMN1 was identified in the African American group (27% compared to 3.3% - 8.1%). This latter finding has clinical implications for providing accurate adjusted genetic risk assessments to the African American population.

The implications of this work are most significant for Hispanic patients who carry lower risks of SMN1 mutations and for African American patients who bear increased frequencies of two-copy alleles. These data will facilitate the accurate interpretation of clinical testing results and provide additional information for genetic counseling.

Differences in the frequency of SMA carriers were significant among several ethnic groups. This study provides an accurate assessment of allele frequencies and estimates of adjusted genetic risk that were previously unavailable to clinicians and patients considering testing.



## New Guide to Spinal Muscular Atrophy Research Published by Families of SMA

Families of SMA has released a new booklet covering the basics of Spinal Muscular Atrophy research.

This guide helps to answer questions such as what are the key areas of SMA research and what are the costs for conducting SMA drug development and clinical trials. Clear definitions and graphics are included to help explain and illustrate how SMA drugs begin and the key steps involved in developing new therapies for Spinal Muscular Atrophy.

The topics in this new booklet cover the following important areas of SMA research:

- SMA Researchers.
- Drug Research
- Gene Therapy
- Stem Cells
- Clinical Trials
- Government Research
- The FDA

If you would like to download a copy please visit our website.

If you would like a hard copy mailed to you please email us at [info@fsma.org](mailto:info@fsma.org) or call (800) 886-1762.





[www.curesma.org](http://www.curesma.org)

## The Spinal Muscular Atrophy Community Mourns the Loss of Tim Utzat - Contributor to the Type I Care Packages.

Families of Spinal Muscular Atrophy and the entire SMA Community mourn the loss of 38 year old Tim Utzat, of Toms River, NJ. Tim, along with his wife Mary Jane, assembled the Type I care package that Families of SMA currently sends out to Type I families.

These care packages include SMA type I related toys. Every new type I family, whose child has recently been diagnosed with Type I, receives one of these care packages. Tim was in a serious work related accident on August 10th where he sustained burns to approximately 80% of his body. He passed away on Sunday, August 16th.



The Utzat's lost their daughter, Samantha Jane, to SMA Type I on September 14, 2007, two days before her first birthday. They designed these care packages, in memory of Samantha, as a way to support newly diagnosed families with ideas that worked for their daughter. They were recently featured as one of the Families of SMA "25 Voices of SMA" (see page 69 for the 25 voices profile)

Mary Jane delivered a new baby girl named Sydney Ann on Thursday August 20th, just four days after Tim died.

Families of SMA would like to dedicate all of the Type I care packages sent to new SMA families in Tim's honor. He has helped to make such a difference in the lives of so many SMA families everywhere.



Tim Utzat with his daughter Samantha