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www.curesma.org
Sharing

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.

Submissions
To submit articles or make other contributions to our newsletter, please contact us at: newsletter@fsma.org. Digital images are encouraged! Send your digital pictures to: newsletter@fsma.org.

Change of address
Send changes, including ZIP code to: info@fsma.org or call 1-800-886-1762 or mail to: FSMA Membership, 925 Busse Road, Elk Grove Village, IL 60007 ©2013 Families of SMA. All rights reserved. No portions of this publication can be reprinted without written consent from FSMA. 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.

Registration for the 2014 Annual SMA Conference, held in Washington, DC June 12th–15th, at the Gaylord National in National Harbor, MD, is now available

For more information and to register, please visit www.CureSMA.org

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.

On the Cover: Our “It’s a Wonderful Life” panel from the 2013 Conference (Left to right): Ben Mattlin, Melissa Milinovich, Vicky Jurney-Taylor, Gina Semenza and Rocco Arizzi.

Families of SMA

DIRECTIONS

2 | Directions Fall 2013
Every year, Families of SMA sponsors a conference to bring together the leading SMA researchers, clinicians, and families living with SMA. Families of SMA has been hosting the Annual SMA Conference since 1989. The weekend is filled with a wide variety of workshops, a memorable children’s program, a family fun fest and carnival, many opportunities to connect and interact with families and receive first hand updates from the researchers.

Every year we look forward to reuniting as a community at this conference and showing our support for each other. 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 it as we have the Family and the Research Conferences run alongside each other. The interactions between the researchers and families at this one conference are extremely special. The annual conference also provides the children an opportunity to make new friends and have a great time. We are looking forward to a great attendance, in our nation’s capital, as a community.

The Gaylord National Resort & Convention Center has been carefully selected to meet the many needs of Families of SMA as the 2014 destination for the Annual SMA Conference. The hotel anchors the 300 acre National Harbor waterfront entertainment district, located eight miles south of Washington DC, and is situated along the shores of the Potomac River, across the river from Alexandria, Virginia. The hotel contains 2,000 guest rooms, seven restaurants, and features a 19-story glass atrium with views of the Potomac River. For your convenience the rooms are equipped with a mini-fridge, coffee maker and a 32 inch flat screen TV along with other modern amenities. Take care of business with in-room high-speed Internet access. This hotel also has a smoke free policy.

After a day of meetings, indulge in outstanding restaurants and lounges onsite (like the Old Hickory Steak House, Pienza Restaurant, National Pastime Sports Bar & Grill and more). Relax in the Junior Olympic-sized, 24-meter lap pool, perfect for families and fitness. Enjoy the Spectacular nightly Fountain Shows which project a majestic display of lights, sound and dancing water that shoots 60-feet high. Or, take a stroll through National Harbor which is an expansive waterfront that offers restaurants, shopping, entertainment, marinas and piers.

You must first register for the conference before you can reserve your discounted hotel room rate of $149 at The Gaylord National Resort & Convention Center.
The 2013 Annual Spinal Muscular Atrophy Conference
A Fantastic Success

The 2013 Annual Spinal Muscular Atrophy Conference was a fantastic success with over 1,300 families, researchers and professionals in attendance! 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 it for SMA families. The interactions between the researchers and families at one conference are so special. The annual conference also provided our children an opportunity to make new friends and have a great time. There were so many wonderful events that made this conference incredibly special and successful for everyone.

The Family Conference began Thursday, June 13th with the Newly Diagnosed Program where families, who were diagnosed from 2012 to present, were able to attend this special session prior to all of the other families arrival. It was a wonderful way to slowly introduce new families to the conference as well as other families and professionals on a much smaller scale. This program included:

- An introduction to the Conference and SMA Community
- Understanding Genetics and the Disease
- Evidence, Hope and Hype: Finding the Balance
- Life After Diagnosis – Parents Share Their Journey
- A Meet and Mingle with families, doctors, chapter officers and board members
- Optimal Care for SMA Type I
- Optimal Care for SMA Type II and Type III
- A Grieving Parents Session

While the conference was underway for the families, the 17th Annual International SMA Research Group Meeting was being held at the same time and is the biggest SMA research conference in the world. Families of SMA organizes the conference and financially underwrites the meeting by covering hotel, travel and registration for all research presenters. There were 215 researchers that attended from around the world. These researchers represent institutions, biotech and pharmaceutical companies from all over the world. The personal connections made between families and researchers meant so much to everyone who attended.
FSMA Thanks our Exhibitors for Attending
The 2013 Annual SMA Conference in Anaheim, CA

Families of SMA was thrilled to have 30 Exhibitors attend The 2013 Annual SMA Conference. Families of Spinal Muscular Atrophy thanks all of the 30 exhibitors for their generous support of The 2013 Annual SMA Conference. Exhibitors who attended the Annual SMA Conference were able to promote their products to SMA families, Researchers and Medical Care Providers from all over the United States. These exhibitors are partners in our community who are critical to the success in the battle against SMA.

Families of SMA would like to thank the following Platinum Exhibitors:

HAYEK MEDICAL DEVICES

Families of SMA would like to thank the following Gold Exhibitors:

Permobil
Triumph Mobility Inc.
Thomashilfen
Sunrise Medical
Sundance Enterprises, Inc.
ProBed Medical Technologies, Inc.
Hill-Rom

Families of SMA would like to thank the following Silver Exhibitors:

Counsyl, Inc.
RespirTech
Maxim Healthcare Services
Philips Respironics
Neotech Products
Stanford Hospital & Clinics Neuromuscular Program
NeuroNEXT

Families of SMA would like to thank the following Bronze Exhibitors:

Tobii ATI
Laurie’s Legacy
International SMA Patient Registry
LC Technologies, Inc.
THERAsurf
Electromed, Inc.
DisAbility Sports Festival
Access Medical, Inc.
atHome Living Solutions
Ability Center
Newborn Screening Translational Research Network
Superior Mobility
Advanced Pharmacy & Respiratory Care Solutions

If you are interested in being added to our mailing list to receive exhibitor opportunities for the 2014 Annual Conference, please email exhibitor@fsma.org.
Highlights from The 2013 Annual SMA Conference Meet and Greet

The 2013 Annual SMA Conference, held in Anaheim, California at the beautiful Disneyland Hotel, began for all attendees on Thursday evening at the Meet & Greet and Family Fun Fest. We welcomed new families, returning families, researchers, medical professionals and friends from all over the world.

This Thursday evening event is a tradition at the Annual SMA Conference where everyone comes together to begin a weekend full of making new friendships and connections, learning the most up to date information and being a part of a wonderful and caring community.

The ever-so popular Researcher Relay Race is always a highlight at this event as both researchers and kids line-up to see who can cross the finish line first, with one rule: that the researchers have to race in a manual wheelchair! All of the attendees stand around the outside of the course cheering on the racers to see who will be announced as the winner, and it is always the kids who cross the finish line first, leaving the researchers behind in the dust!

Another major component that makes the Meet & Greet such an enjoyable event is the Family Fun Fest with Carnival Games that are lined-up around the edges of the room. Kids and families rotate around to the endless amounts of games ranging from Bozo Buckets, to a Treasure Chest, to the LollyPop Tree and so many more. The games are adapted so that everyone who plays has a chance to win some great prizes. There was even a candy table station and tattoo station as well, filled with goodies and fun for all!

A wonderful family buffet dinner was served to all of the attendees as they bounced around to all of the different activities and experiencing the energetic conference atmosphere. There was even an opportunity to get your picture taken with everyone’s favorite Disney Characters, Mickey and Minnie Mouse!

The Meet & Greet is a special time for all attendees to come together in a fun and relaxed setting, prior to the start of the workshops.

To see photos from the 2013 Annual SMA Conference, please visit the Families of SMA FaceBook page.
Meet and Greet
Researcher Relay Race
On Saturday evening, was the ever exciting yet relaxing, Movie and PJ Party for families. Each person was greeted with a table full of treats and popcorn. Families were provided sheets and pillows to make themselves comfortable and encouraged to sport their comfiest PJs. The movie that captivated the audience was the original “Monster’s Inc.”, which was a huge hit for the children while parents caught up with old friends and met new ones!

Amy Marquez Scholarship Announced at The 2013 Annual Spinal Muscular Atrophy Conference

During the It’s a Wonderful Life workshop at The 2013 Annual SMA Conference, Families of Spinal Muscular Atrophy announced a new scholarship to celebrate the life and amazing strength and spirit of Amy Marquez.

The Amy Marquez Scholarship was started in memory of Amy Marquez, who passed away from SMA Type I at the age of 41. Amy was an active member of the SMA community, FSMA board member, and was a wonderful source of support and motivation for many SMA families.

This scholarship will be awarded each year to an adult with SMA to attend the Annual SMA Conference, in recognition of Amy’s involvement within the SMA adult community. The scholarship will cover both registration fees and hotel costs.

Amy never let SMA take away from her desire to be an amazing wife to Steve and caring mother to Harley and Danielle.
The First Ever Family Friendly SMA Research Poster Session at The Annual Spinal Muscular Atrophy Conference

At The 2013 SMA Conference a brand new event was held called the Family Friendly Research Poster Session. During this new event 23 research groups, including those representing 10 SMA drug programs, presented their scientific findings. This unique event provided an interactive venue for the 1300 families and researchers in attendance to share information on a one-on-one basis.

Please visit the www.CureSMA.org website to view the posters.

The titles of all posters can be seen below:

Family Friendly Research Poster Session

Please use the Family Friendly Research Poster Session sheet, found in your registration folder, to collect stickers at each of the research posters. Rotate around to each poster and once you have collected 8 stickers, you may enter your sheet into the bins located on either sides of the Center Ballroom stage. You will be entered into a drawing to receive a prize, which will be announced at the end of the poster session, at 8:30pm.

Posters Include:

- Sara Custer, PhD, from the Androphy Lab at Indiana University presenting on “Basic biology of the SMN protein in the growth and maintenance of neuronal cells”.
- Wilfried Rossoll, PhD, from Emory University presenting on “The role of SMN in nerve fibers”.
- Lyndsay Murray, PhD, from the Kothary Lab at University of Ottawa presenting “Motor Neurons: What controls their diversity?”
- Armin Yazdani, PhD, from the Kothary Lab at the University of Ottawa presenting “Trichostatin A: Potential therapeutic implications for SMA”.
- Maureen Lynes, PhD, from the Rubin Lab at Harvard University presenting on “Modeling Spinal Muscular Atrophy with stem cells”.
- Shoutian Zhu, PhD & Arnab Chatterjee, PhD, from CALIBR presenting on “Finding drugs that keep motor neurons alive”.
- Zhihua Feng, PhD & Karen Ling, PhD, from the Ko Lab at University of Southern California presenting on “Motor circuit defects and restorations in SMA mice”.
- Tanya, J. Wyatt, PhD, from California Stem Cell, Inc presenting on MotorGraftTM for the treatment of SMA.
- Yong-Cha Ma, PhD, from Northwestern University presenting on “Understanding the molecular mechanisms of motor neuron degeneration in SMA”.
- Genevieve Paris, from the Cote Lab at University of Ottawa presenting on “Investigating the contribution of SMN’s ‘favorite protein flavour’ (methylated arginines) in SMA”.
- Kristina Lemonidis, from Isis Pharmaceuticals presenting on “Developing an antisense drug for the potential treatment of SMA”.
- Michael Tones, PhD, from Pfizer presenting on “Families of SMA, Biotech and Pharma: working together to develop new medicine for SMA patients”.
- Brian Kaspar, PhD, from Nationwide Children’s Hospital presenting on “Update on gene delivery for SMA”.
- Christine DiDonato, PhD, from Northwestern University presenting on “Animal models, cell requirements and therapeutics for milder forms of SMA”.
- Panayiota Trifillis, PhD, from PTC Therapeutics & Irene Gerlach, PhD, from F. Hoffman-La Roche, & Sergey Paushkin, PhD, from the SMA Foundation presenting on “A partnership between Roche, PTC Therapeutics and the SMA Foundation to help children with SMA”.
- Stephen J. Kolb, MD, from The Ohio State University presenting on “SMA Infant Biomarker Study - What, why and how?”
- Eric Dessaud, PhD & Rebecca Pruss, PhD, from Trophos presenting on “The path to the discovery and development of olesoxime (TRO19622) for SMA”.
- Marco Passini, PhD, from Genzyme Corporation presenting on ”Progress towards AAV-SMN gene therapy for Spinal Muscular Atrophy”.
- Jeff Jasper, PhD, from Cytokinetics presenting on “Fast skeletal troponin activators for the potential treatment of neuromuscular diseases”.
- Umrao Monani, PhD, from Columbia University presenting on “Defining an optimum window for the treatment of SMA”.
- Christian Lorson, PhD, from University of Missouri presenting ”Inhibiting the Inhibitors: Morpholino ASOs rescue SMA mice”.
- Rhannon Desideri & David Valdivia, from the Sumner Lab at Johns Hopkins University presenting on “Comparative pathology of human SMA and conditional mouse models”.
- Arthur Burghes, PhD, from The Ohio State University presenting on “New morpholino antisense oligonucleotides for the treatment of SMA”.
Family Friendly SMA Research Poster Session
This year’s Children’s Program was such a fun and entertaining time, had by all! Thanks to so many of our wonderful volunteers, all of the children had an exceptional time. Children stayed busy with countless arts & crafts projects, loads of toys, movies, fun activities, Wii stations and so much more.

There was exciting entertainment for the children to enjoy such as a magic show, ventriloquist show, adorable tea party, puppet show as well as a bubble show!
Adaptive Arts Project for The 2013 Annual SMA Conference a Huge Success!

The Jacob Isaac Rappoport Foundation awarded Families of SMA with funding for an Adaptive Arts Project for the Annual SMA Conference which was a huge hit with all the children who attended!

A new and exciting Adaptive Arts Project was added to the conference agenda this year and was sponsored by the Jacob Isaac Rappoport Foundation. All of the children at The 2013 Annual SMA Conference had a great time participating in such a fun and inspiring event. Children of all ages were able to take home wonderful art pieces that they painted with their wheelchairs. Families of SMA was excited to be able to offer kids this fun opportunity to express their artistic personalities!

Zot Artz designs and makes adaptive art tools so that children can create amazing art pieces with adaptive tools and assistive technology which transforms wheelchairs into giant paintbrushes. Children of all abilities and ages can stamp, draw and paint to make their mark.

Families of SMA would like to thank the Jacob Isaac Rappoport Foundation for their generous support!
To learn more about the Jacob Isaac Rappoport Foundation, please visit www.ourshootingstar.com.
To learn more about Zot Artz, please visit http://www.zotartz.com/.
During the opening general session of The 2013 Annual SMA Conference The Jacob Isaac Rappoport Foundation was honored for exceeding the milestone of raising over $1 Million. In an amazing demonstration of their generosity and dedication the family presented an incredible donation check for $100,000!

Here is the speech given by Shaina at the conference:

“I have imagined this moment many times. Eleven years ago, Adi and I attended our first FSMA conference, with a five month old Jacob in tow. He had been diagnosed only 6 weeks before. We were greeted with open arms and hearts, and while we asked ourselves how it was possible that we had become a part of this family, we knew we had come to the right place.

During that first conference, a $100,000 donation was announced, and it was in that moment that I knew when I could no longer fight for Jacob’s life, I’d fight alongside this organization until a cure is found.

The first day Jacob was diagnosed, we reached out to FSMA and heard Colleen’s voice on the other end of the phone. Back then, there was no South Florida chapter nor clinical trials. We have come so far! Our FSMA family welcomed us, educated us, connected us, supported us, loved us, loved Jacob. We would have never been able to do all that we have done without the love and support we have felt from FSMA.

Fast forward one decade, and I find us here, celebrating having raised $1 million. Over the years, people have asked us how we do it. Or have told us that there is something extraordinary about our efforts. True, most people do not raise a million dollars in ten years, and true, I’d be lying if I said it was easy. And yes, a million dollars is an unbelievable, significant milestone.

But really, I’m just a mom with a son. And because I am not able to have the relationship I would choose to have with my son, I had to re-invent a mother-son relationship for Jacob and me.

So for the past eleven years, instead of packing lunches, I make corporate packages soliciting donations and draw event site plans; instead of choosing schools, extracurricular activities and halloween costumes, I choose event t-shirt colors and logo designs; instead of driving to playdates and baseball games, I drive to pick up donations of food and posters with pictures of other SMA heroes.

Different tasks, yes. But the commitment and love? That is not any different from what any mom would do for her son.

To Adi, my true hero in every way—although it is still hard to comprehend that this is our path—I can’t imagine walking it with anyone else. I love you more each day.

To Jordan, Max and Sam: this is your moment too. As much as SMA has taken from my own life and from Jacob’s life, it takes from our living children’s lives too. But it has given us the opportunity to see compassion, generosity and love we would have never been able to see. Thank you three, for giving me purpose each day— and I know that Jacob lives inside each of you.

To my parents, who traveled across the country to be with us today— thank you from the bottom of my heart. Your love and support for our family enabled us to be the best parents possible for Jacob while he was with us on earth— and your unending support and love for him has been instrumental to our fundraising success. Thank you for supporting our endeavors in every way.

We have never made a public presentation of the foundations contribution. However, in the spirit of the $100,000 donation we heard announced eleven years ago; to mark the first chapter of our fundraising journey; and perhaps to inspire someone to be up here celebrating one million fundraising dollars—1 decade down the road—the Jacob Isaac Rappoport foundation would like to present FSMA with $100,000 donation today.

Chalk it up to the commitment and love from another mom- for her son. Thank you.”
Many workshop presentations from The 2013 Annual SMA Conference that was held at Disneyland in California are now available for you to view and download on-line, on the FSMA website.

Friday and Saturday offered families informative workshops and sessions. There were over 40 workshops offered during the conference covering the latest information on critical topics in SMA Care including:

- Sharing your SMA Type experiences, as well as a session for Adults with SMA, Grandparents of SMA children and a session for grieving parents
- Life Care Planning
- Genetics and Reproductive Options for SMA Families
- Hands on Physical Therapy
- Dad’s Time: A Workshop for Fathers Only
- Care for the Caregiver
- It’s a Wonderful Life
- SMA Kids Talk it Out, Sessions I and II
- Siblings Talk it Out Workshop Sessions I and II
- Orthopedic Management
- Healing the Grieving Heart Parts I & II
- What Parents & Guardians Need to Know About Healthy Sexual Development in Adolescents
- Writing Our Stories: A Parent’s Workshop in Healing Through Creativity
- Breathing Basics and Care Choices for SMA Type I
- Breathing Basics for Type II and Type III
- What it Means to be a Teen on Wheels
- Events Success & Fundraising Resources
- Nutrition for G-Tube Feeders
- Transition to Adulthood
- Writing Our Stories: A Parent’s Workshop in Healing Through Creativity
- Keepsake Creation: Grieving Through Art
- Anticipatory Grief: Preparing to Expect the Unexpected
- Yoga for SMA
- Aquatic Physical Therapy for Fun & Function
- Recognizing and Managing Pain in SMA

To view the conference workshop presentations, please visit www.CureSMA.org
Presentations will continue to be added as we receive them from conference speakers.
Photos from The 2013 Annual Spinal Muscular Atrophy Conference Are Now Available

The photos taken at The 2013 Annual SMA Conference in Anaheim, CA at Disneyland are now available on the Families of SMA Facebook page.

The photos were taken during many of the events at the conference such as the Meet and Greet, the Disney Character Event, the Researcher Relay Race, the Family Friendly Research Poster Session, the Disneyland Fireworks Event and the PJ Party and Movie Night. Other photos were taken during the Research meeting, as well as many workshops throughout the conference. There are also photos from the incredibly fun Children’s Program, where hundreds of children kept busy with activities, entertainment and games.

To view photos, please visit the Families of SMA Facebook page. The photos are in the albums section.

If you have any photos that you would like to submit from The 2013 Annual SMA Conference, please email them to FamilySupport@fsma.org.
For the past 4 years we have been able to provide the conference with a more professional and organized feel as the Expo Convention Contractors, and Jennifer Miller Smith & Aaron Smith of Miami, FL have donated all of the signs, banners and registration tables throughout the conference. All of these items are worth over $35,000 and we cannot thank each and everyone one of these individuals enough, for their incredible generosity. You truly have an important impact on making the conference such a great success year after year and helping to create an atmosphere unlike anything else for families, researchers and medical professionals, alike.

FSMA ANNOUNCES TWO WINNERS OF THE 2013 ANNUAL SMA CONFERENCE IPAD SURVEY GIVEAWAY

At The 2013 Annual SMA Conference, Families of SMA was able to offer a great incentive to all attendees who completed a conference survey this year. Any conference attendee who submitted a conference evaluation form before the Researcher Q&A on Sunday morning had their name entered into a raffle to win a new Apple iPad. Congratulations to both of the winners at this year’s conference. Everyone at Families of SMA would like to thank Isis Pharmaceuticals, as well as Kristina Lemonidis from Isis, who donated and helped award the iPads to the two winners. Their generosity has made these two SMA families extremely happy! Thank you to all conference attendees who completed a conference survey, this has helped provide us with great feedback to consider for improving next year’s conference.
Families of SMA held a Continuing Medical Education Conference for Medical Professionals on Wednesday, June 12th, prior to the start of The 2013 Annual SMA Conference in Anaheim, CA. Over 100 Medical Professionals from around the United States attended the Continuing Medical Education Conference titled “Interdisciplinary Perspectives on Spinal Muscular Atrophy: Defining Your Role”.

This one day conference, held exclusively for medical professionals, focused on the diagnosis of infants and children with neuromuscular weakness, clinical application of care standards to individuals with SMA, and discussion of pathophysiology and therapeutic strategies for intervention. Members of the Families of SMA Medical Advisory Council, as well as expert speakers from across the United States, led the CME Conference, which included lectures, panels and discussions.

This CME Conference for medical professionals is a great step Families of SMA is taking toward broadening our support programs by educating medical providers. The Annual Conferences previously covered two components of SMA, Research and Support. The Research aspect covered scientific, research and clinical updates, while the Support aspect included the family support workshops and family networking.

The new addition provides a third component of Care to the Families of SMA Conferences by educating medical providers on SMA. Families of SMA was thrilled to be partnering up with University of Wisconsin School of Medicine and Public Health, who are accredited to provide continuing medical education credit for medical professionals.

The 2014 Continuing Medical Education Conference will be held on Friday, June 13th in Washington, DC at the Gaylord National Resort and Convention Center. More information will be available soon on the FSMA website.
Families of SMA Releases Keynote Speeches from the Research Question and Answer Session at The 2013 Annual SMA Conference

The 2013 SMA Conference brought together over 1,300 families and researchers from around the world. At the conclusion of the conference, families gathered for a special opportunity to hear about the latest SMA research.

The Keynotes Presentations given during the closing session at The 2013 SMA Conference addressed new advances, strategies, and challenges in SMA drug development.

- Update on Families of SMA Research Activities, Jill Jarecki, Ph.D., Families of SMA Research Director.
- Major Findings from the 17th SMA Research Meeting and Implications for SMA Therapy Development, Douglas Kerr M.D., Ph.D., Medical Director, Experimental Neurology, Biogen Idec.
- Considerations and Importance of Clinical Trial Participation, Thomas Crawford, M.D., Professor Of Neurology & Pediatrics, Johns Hopkins University.

Please go to www.CureSMA.org to download the presentations.

Families and Researchers Interact at The 2013 Annual Spinal Muscular Atrophy Conference

215 researchers from 15 different countries, 70 distinct organizations, and 17 biotech and pharmaceutical companies attended the 2013 meeting.

The SMA Research Group Meeting is the largest research conference in the world for SMA. It was held June 13, 14, 15 at the Disneyland Hotel in Anaheim, California. 110 updates were given on the latest breakthroughs in SMA research.

The research meeting is held together with The Annual SMA Family Conference. Running the two conferences simultaneously gives the unique opportunity for SMA families, researchers, and clinicians to interact and meet each other.

At the Annual SMA Conference multiple events were held for researchers and families to interact:

1) The annual researcher relay race
2) A brand new family friendly research poster sessions
3) A research question and answer panel

Twenty Three Labs, Including Those Representing 10 Drug Programs, Presented at the First Ever Family Friendly Research Poster Session.
The Research Question and Answer Session closed The 2013 SMA Conference, where 1,300 researchers and families attended to learn the latest information about Spinal Muscular Atrophy. This session addressed new advances, strategies, and challenges in SMA drug development. Leading experts on SMA drug development answer questions about their drug programs.

**The first video includes:**
- An update on Families of SMA Research Activities, Jill Jarecki, Ph.D., FSMA Research Director.
- Major Findings from the 17th Annual SMA Research Meeting and Implications for SMA Therapy Development, Douglas Kerr M.D., Ph.D., Medical Director, Experimental Neurology, Biogen Idec.
- Panel 1: Representatives of Drug Programs in Preclinical Development:
  - Brian Kaspar, Ph.D., Associate Professor, Department of Pediatrics, The Research Institute at Nationwide Children’s Hospital.
  - Katherine Klinger Ph.D., Sr. Vice President, Genetics and Genomics and Presidential Fellow, Genzyme Corporation.
  - Douglas Kerr, M.D., Ph.D., Director of Experimental Biology, Biogen Idec.
  - Irene Gerlach, Ph.D., Project Leader, F. Hoffmann-La Roche AG.
  - Chris N. Airriess, Ph.D., Chief Operating Officer, California Stem Cell, Inc.

**The second video includes:**
- Considerations and Importance of Clinical Trial Participation, Thomas Crawford, M.D., Professor Of Neurology & Pediatrics, Johns Hopkins University.
- Panel 2: Representatives of Drug Programs in Clinical Development or of Human Research Studies:
  - Jerry Mendell, M.D., Professor of Pediatrics And Neurology, Nationwide Children's Hospital.
  - Kathie M. Bishop, Ph.D., Director, Clinical Development, Isis Pharmaceuticals.
  - Gene Liau, Senior Director, Rare Disease & Hematology, External Research Unit (ERDI) Pfizer Worldwide R & D.
  - Rebecca Pruss, Ph.D., Chief Scientific Officer, Trophos.
  - Stephen J. Kolb, M.D., Ph.D., Assistant Professor, Neurology, The Ohio State University.

For the latest news visit the FSMA website: www.curesma.org
Families of SMA thanks all of the following organizations for their generous support of the 2013 Conference. These sponsors are partners in our community who are critical to success in the battle against SMA.

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The Jacob Isaac Rappoport Foundation

fighting spinal muscular atrophy in memory of our shooting star

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Families of SMA would like to thank Quest Diagnostics for being the Presenting Sponsor of the 2013 Annual SMA Conference.

Quest Diagnostics generous sponsorship helped contribute to scholarships and family assistance programs for the 2013 Annual SMA Conference and for travel and lodging expenses for the International SMA Research Meeting. Without their support many patients, families and researchers would never be able to attend and join together in this fantastic meeting.
FSM A  G oals for the SM A  R esearch G roup M eeting
An important way Families of SMA advances research is The Annual International SMA Research Group Meeting. This meeting is the largest SMA research conference in the world. For 17 years, FSMA has organized the conference, and has financially supported the meeting by covering hotel, travel, and registration for all research presenters for a total cost of about $200,000 each year. The meeting this year was held at the Disneyland Hotel in Anaheim, California. 225 researchers attended the conference to learn about the latest developments in SMA research. The researchers attending represented over 60 academic organizations and 17 biotech and pharmaceutical companies, from 15 countries around the world. 110 research presentations were given at the conference this year, during 10 different scientific sessions. Families of SMA has specific goals for the meeting to help benefit the entire SMA community:

• Enables open communication of early, unpublished scientific data among researchers to accelerate the pace of research.
• Creates a vital sense of community among SMA researchers to promote a collaborative spirit and produce research partnerships.
• Allows cross-disciplinary dialog among basic researchers, clinicians, and industry representatives, vital in creating effective therapies.
• Motivates SMA researchers by allowing for direct interaction with families and patients living with SMA.
• Provides a venue to integrate new researchers and drug companies into our community as efficiently as possible.
• Allows young researchers to interact with experienced leaders in the field to help build the future of the SMA research community.

Please see www.curesma.org for a detailed summary of the scientific findings from the 2013 SMA Research Group Meeting.

What Do Researchers Say About The SMA Research Group Meeting?

“This meeting is immensely important. It grounds and motivates all the researchers to see the families and patients their work is meant to help. This is the single largest concentration of global SMA researchers and it provides great opportunities for networking, catching up and sharing new ideas. Seeing new data and ideas at this meeting really allows the research community to grow and collaborate.”

“Definitely is the yearly opportunity to network and forces us to reach out and talk about each others unpublished results, in the end fostering true, useful collaborations, resulting in more thorough and solid research and discoveries.”
The 2013 Annual SMA Conference brought together over 1,300 families and researchers from around the world. At the conclusion of the conference the families gathered for a special and unique opportunity to hear the very latest research announcements along with a Question & Answer session focused on the leading drug programs and human clinical research studies in SMA. Summaries of each of these programs are below.

Roche, in collaboration with PTC Therapeutics and the SMA Foundation, is developing small molecules that increase the production of SMN protein, which is deficient in patients with SMA. Pre-clinically, the compounds reach the central nervous system with oral administration. In various mouse models of SMA, these molecules have been observed to increase SMN levels, normalize the phenotype of neuromuscular junctions, improve motor function, increase body weight, and prolong survival. Current development efforts are focused on profiling the safety of such drugs in animals while exploring clinical biomarkers and defining the best measures for safe, rigorous and efficient clinical studies in SMA patients.

California Stem Cell, Inc is actively pursuing Phase I clinical trials for the lead therapeutic candidate MotorGraft in SMA Type I. MotorGraft is a high purity population of motor neuron progenitor cells from human embryonic stem cells that has been shown in vitro to be functionally active, secrete beneficial growth factors, and have the capability of innervating muscle. Animal studies have demonstrated that MotorGraft survives and integrates following transplantation. The functional benefits of MotorGraft observed in vivo include increased body weight, improved muscle EMG recordings, preservation of muscle function, and improved cardiorespiratory function. These benefits are supported by histological findings that reveal enhanced neurite branching, sparing of endogenous motor neurons, preservation of proprioceptive input to the motor neurons, increased muscle fiber diameter, and increased neuromuscular junction innervation and maturation with transplantation of MotorGraft. Extensive preclinical safety testing demonstrates that MotorGraft does not form tumors, does not induce allodynia, does not biodistribute, and does not have toxic effects. California Stem Cell, Inc has submitted an application to the FDA to start a clinical trial, and is currently on clinical hold in the US. While the clinical hold issues are being addressed for the US clinical trial, California Stem Cell is also pursuing a Phase I clinical trial in the UK. California Stem Cell’s program was presented at a recent Scientific Advice Meeting with the UK regulatory agency, the MHRA. A full clinical trial application will be submitted to the MHRA by the end of 2013.

See our website for details about FSMA funding of the CSC project.

Genzyme (a Sanofi company) has an active program in SMA gene therapy, focused on AA49-SMN gene delivery into the cerebrospinal fluid (CSF). In studies in mouse models of SMA, we have shown that delivery of AA49-SMN1 into the CSF results in transfer of the gene to the spinal motor neurons, and expression of SMN protein. The treated mice show significant improvement in strength, motor function, and survival. We have determined the lowest percentage of motor neurons that must be modified by AA49-SMN-1 in order to make significant improvement in SMA mice. We have also shown that this level of gene transfer can be achieved using CSF delivered gene therapy in large animals, such as juvenile pigs and non-human primates. These findings provide the foundation for continued development of this therapeutic concept for SMA.

Gene delivery using novel Adeno Associated Virus, such as AA49 has demonstrated great promise for delivering genes to the brain and spinal cord. Two delivery routes have emerged as being effective for targeting motor neurons. The first route is systemic delivered AA49 through the bloodstream. Recently, we and others have demonstrated another route of delivery, injection into the cerebrospinal fluid (CSF) surrounding the brain and spinal cord that also effectively targets motor neurons. For the systemic route, we anticipate submitting an Investigation New Drug Application to the FDA over the coming months for approval of a Phase I trial entitled “Phase I Gene Transfer Clinical Trial for Spinal Muscular Atrophy Type I Delivering the Survival Motor Neuron Gene by Self-Complementary AA49”. The trial is anticipated to enroll 9 infants with SMA Type I at Nationwide Children’s Hospital in Columbus, Ohio. The study will enroll infants of nine months of age and younger with type 1 SMA as defined by the following features: a) proven mutations of the SMN1 gene with two copies of SMN2, b) onset of disease at birth to 6 months of age, and c) hypotonia and muscle weakness demonstrated at time of enrollment. Additional exclusion criteria will also apply. The primary trial outcome will be safety, and the secondary outcomes are the CHOP INTEND functional motor scale and electrophysiological measures. The trial design will be an open-label, dose-escalation clinical trial of scAA49.CB.SMN injected intravenously through a peripheral limb vein. The team will evaluate short-term safety over a two-year period. Patients will be tested at baseline, on days 7, 14, 30, and once every month for up to 2 years, with long term follow-up for at least 15 years. Trial launch is subject to FDA approval. The team also has excellent pre-clinical data on the finding that AA49 can be delivered to motor neurons efficiently by placing the virus into the CSF and are advancing on this program too. CSF delivery permits the use of less virus, thereby potentially allowing treatment of older and bigger patients. Dr. Kaspar recently was awarded a $3.8M cooperative award from NINDS to advance a CSF directed gene therapy for SMA.

See our website for details about FSMA funding of the Dr. Kaspar’s gene therapy project.
Kolb Laboratory from the Wexner Medical Center at The Ohio State University is conducting the SMA Biomarker in Infancy Study. This study will establish the natural history of putative SMA biomarkers during the first two years of life in SMA and healthy infants. Our results will aid in the rational design of future SMA interventional studies in infants and will serve as an important data set in the design of future clinical trials in additional diseases of infancy. The Biomarker Study is currently enrolling subjects and includes infants with genetically confirmed SMA and healthy infants without SMA. The Biomarker Study is being performed at 15 sites across the nation and is sponsored by the National Institutes of Health and FSMA. It is also the premier study for the NINDS Network of Excellence in Clinical Trials known as NeuroNEXT. For more information about the study, please type “SMA Biomarker Video” into your search engine and view our Informational YouTube™ Video and/or contact Amy Bartlett, CCRC at (614) 366-9050.

See our website for details about FSMA funding of the NeuroNEXT SMA project.

Trophi is conducting a Phase II study of Olesoxime (TRO19622) in 3-25 year old Spinal Muscular Atrophy (SMA) patients. Olesoxime prevents neuronal death and promotes neuroregeneration in multiple preclinical models of neurodegeneration. Olesoxime has been studied in several clinical trials showing that the compound is safe and well tolerated. Pharmacokinetics have been in healthy volunteers and both adults and children with spinal muscular atrophy. A Phase II, multicentric, randomized, double-blind, placebo controlled study is ongoing to study the effects of olesoxine in the broadest range of SMA patients. Between November 2010 and September 2011, 165 non-ambulatory type 2 and 3 SMA patients ranging in age from 3-25 years, old were enrolled. Subjects were randomized 2:1 on olesoxine or placebo. The trial is conducted by experienced clinical child neurologists in 22 clinical centers in seven European countries. Patients receive olesoxime or matching placebo in an oral liquid formulation once a day with a meal and are followed every three months for the 24 month study. Motor function assessed using MFM is the primary endpoint in the study. HMFS, respiratory function and EMG parameters (CMAP and MUNE) assessed at 6 month intervals are secondary endpoints, together with Pediatrics questionnaires (PedSQL). Interim efficacy and futility analyses were performed after all patients had been treated for one year. A data monitoring committee evaluated these interim data for safety and possible efficacy and recommended to continue the trial as planned. Final results of the trial should be available by the end of 2013. In parallel, blood samples are being collected at baseline during the screening visit, at one year and after completion of the trial for biomarkers analysis in collaboration with SMA Foundation. This clinical trial is funded by the AFM Telethon (French association against myopathies), which also supported the discovery and development of olesoxime specifically for SMA.

In January 2013, Repligen announced a licensing agreement with Pfizer to advance its spinal muscular atrophy (SMA) program, originally in-licensed from Families of SMA (FSMA). The program included RG3039, a small molecule therapy candidate in Phase 1 development, as well as backup compounds and enabling technologies. Pfizer is committed to advancing potential new treatment options for SMA; a rare disease with significant unmet need. At this time, RG3039 has completed two of four planned cohorts in the Phase 1 study initiated by Repligen. This initial study, in 16 healthy volunteers, provided a rich source of information about the profile of RG3039. Pfizer believes the Phase 1 data gathered to date is sufficient to achieve the research goals of this study at this stage. We have closed the Phase 1 trial to additional participants, and are working to generate a robust biomarker plan for RG3039 and to understand optimal dosing for this experimental molecule. We believe these steps will enable a better understanding of how to design future clinical studies. In parallel, we are exploring earlier-stage experimental programs of research that may provide potential treatment options for SMA patients in the future.

See our website for details about FSMA funding of the RG3039 (quinazoline) project.

ISIS-SMNRx is an antisense drug Isis is developing to treat SMA. SMA is caused by a loss of, or defect in, the SMN1 gene leading to a decrease in the amount of SMN protein. SMN protein is critical for the health and survival of nerve cells in the spinal cord that are responsible for neuromuscular growth and function. The severity of SMA correlates with the amount of SMN protein. Isis has designed ISIS-SMNRx to potentially treat all types of SMA by altering the splicing of a closely related gene, SMN2, which leads to the increased production of SMN protein. A Phase 1 clinical study evaluating the safety of single doses of ISIS-SMNRx in children with SMA has been completed. In this study, ISIS-SMNRx was well tolerated at all dose levels tested with no safety or tolerability concerns. The compound is delivered by an injection into the lower back (an ‘intrathecal injection’) into the space containing cerebral spinal fluid below the spinal cord in order to best distribute the drug to spinal cord motor neurons. The intrathecal injection procedure was also well tolerated in the children. ISIS-SMNRx is now being studied in Phase 2 clinical studies that are designed to examine the safety and tolerability of multiple doses of the drug given over a longer time period. These studies are being conducted in children with Spinal Muscular Atrophy aged 2 to 15 and in infants with SMA who are <7 months old and are expected to complete in 2013. Following on, larger controlled Phase 3 studies are planned to begin in early 2014. In January 2012, Isis and Biogen Idec entered into a preferred partner alliance that provides Biogen Idec an option to develop and commercialize ISIS-SMNRx. Isis also acknowledges support from the following organizations for this program: Muscular Dystrophy Association, SMA Foundation, and Families of Spinal Muscular Atrophy.

See our website for details about FSMA funding of the Isis project.
For 17 years, the SMA Research Group Meeting has brought together the leading researchers from around the world to share ideas and the latest scientific breakthroughs about SMA. Doing so has helped new researchers enter into the SMA field, promote collaborations among established research groups, and allowed pharmaceutical companies interested in SMA to quickly integrate into our community. Please read several real world examples from the SMA research community about how the SMA Research Group Meeting has impacted their organization.

Arthur Burghes, Ph.D., Professor, The Ohio State University

“I have attended the Annual International SMA Research Group Meeting hosted by FSMA from the very start 17 years ago. The meeting was first started after the identification of the SMN gene to give a forum for all researchers from many different scientific areas to attend, but with all focused on the common theme of SMA. Prior to that point there had been some research meetings, but these were specifically concerned with exchange of genetic data to find the SMA gene. The subsequent meetings, which developed into the Annual SMA Research Group Meeting, became much broader in topic areas and have expanded from a few people around a table to many people in a conference setting. The conference serves many important roles, and one of these is mentorship. At the SMA Research Group Meeting, there is the opportunity for one-on-one interactions with those at all levels of scientific training from junior to senior investigators. Here researchers are free to ask multiple questions, get advice and help with proposals, and obtain reagents or suggestions on how to perform a technique. Second the junior investigators have the chance in either poster sessions or talks to present their work and obtain feedback from senior investigators. Lastly there is exposure of the young investigator to the leaders in the SMA field. In summary at the FSMA meeting, there has been exchange of reagents, ideas, and the formation of collaborations between many groups. The meeting has also encouraged spirited debate, and the presentation of new ideas. This is helpful for junior researchers to see, as it defines the scientific problems more clearly. This fosters the design of experiments, typically completed by the junior investigators, to be more directed and thus resolve the scientific issues surrounding SMA more clearly.”

Christine DiDonato, Ph.D., Associate Professor, Northwestern University

“The FSMA International meeting has been instrumental in moving basic science forward toward our understanding of why low SMN levels affect the nerves that control our muscles and the development of a number of different therapeutic strategies aimed at increasing SMN levels or treating other symptoms of disease. An important aspect of moving basic findings towards therapies is the collaborative efforts between academics and biotechnology/pharmaceutical companies. FSMA and the yearly international research meeting has been critical in fostering these interactions, and my laboratory has developed collaborative efforts with Repligen, Inc through the conference. Our collaborative work with Repligen and now Pfizer has focused on performing pre-clinical efficacy studies in mice using RG3039, the novel quinazoline compound, which has recently completed a Phase I clinical trial. In fact, Pfizer hosted one of their first in-face meetings with Key Opinion Leaders at the 2013 conference. Our pre-clinical work helped lay the foundation for filing an Investigational New Drug (IND) application with the FDA. Our current research correlates survival and functional benefits and drug exposure in mice and has helped in guiding dose levels in the first in human studies. This work has been a long-term labor intensive endeavor to reach this point, but personally very rewarding for my academic group to know that we are helping to bring a drug forward to the clinic for our patients.”

Kathie Bishop, Ph.D., Director of Clinical Development, Isis Pharmaceuticals

“I first attended the Families of SMA Annual Research/Families Meeting in 2010, shortly after joining Isis Pharmaceutical and starting to work on their ISIS SMN-Rx drug development program. That meeting was invaluable for myself and the other Isis attendees, as it was where we held our first advisory meeting for the program and made initial connections with advisors, collaborators, foundation members, and potential clinical investigators, which has set the stage for many of our activities since then. I also attended in 2011, 2012, and 2013, again finding the experience invaluable for learning about the latest research and clinical advancements in SMA, but also for the opportunity to network and meet with existing and new collaborators and advisors to our program. In 2011, we also held an advisory meeting, this one to seek advice and input on the initial Phase 1 clinical study of ISIS SMN-Rx. In 2013, Isis held at training meeting at the conference for our clinical trial evaluators. In 2011 2012, and 2013, I was also honored to participate in the Families Meeting Drug Development panel, an experience that I think is very important for connecting with SMA families and learning about what their questions and concerns about the drug development process are, and hopefully answering those questions. In addition, for all of us here at Isis who have attended the meeting, the unique experience of the side-by-side meetings with researchers and the SMA families is extremely motivating, as is meeting and talking with the families and children with SMA.”
Families of SMA is so fortunate to have received three new Tumble Forms® Starfish Bath Chairs donated from the Joseph Lillo Spinal Muscular Atrophy Foundation for Children, for the FSMA Equipment Pool! These bath chairs will be sent to SMA families in memory of Joseph Dominic Lillo. Thank you to the Joseph Lillo Spinal Muscular Atrophy Foundation for these wonderful additions to the Equipment Pool!

Families of SMA is so grateful to have received a special delivery of hundreds of Scentsy Antibacterial Fragrance Foam bottles from Patti Slojkowski for our Newly Diagnosed Care Package Program! These will be included in the care packages that will be sent to newly diagnosed families when they first contact Families of SMA. Thank you Patti for this amazing donation for our care packages and for your incredible support!

Thanks to some volunteer football players from Maine South High School in Park Ridge, IL, the truck was packed full with over 1,000 boxes and shipped to the Disneyland Hotel for the 2013 Annual SMA Conference!
Hello FSMA,
I received the package today, and I can’t thank you enough for all the little and big things that were in there. From one of those small toys, I gave my son the ball shaped rattle, he held it and when he was moving his hand he seemed surprised and happy as well. I’m sure those toys will definitely be enjoyed by him. Thank you once again to the entire FSMA team who helped putting a package like this together.
Thank you,
Bushra Ali of Ohio

Families of SMA,
Words can not express our family’s appreciation for our care package from FSMA that arrived this evening! We are overjoyed and our daughter, Faith, kept saying, “Woah!” Thank you so very much! We’ve only known about this diagnosis for a week now, so we’ve really been through a roller coaster of emotions and this helped our spirits immensely! We are so excited for all of the support we’ve received from FSMA! Beginning this diagnosis of SMA has been a little easier with the support & gifts we’ve received through your organization! I’m excited to see there is such a great support system here & look forward to being active in this with your team!
Sincerely,
Don & Leann Fortenberry of Texas
Parents of Faith Opal - SMA Type II - 17 months old

FSMA,
We have received the care package. Thanks so much! That was so wonder-fully generous of you guys! We have checked out the site and plan on becoming very involved with as many resources as possible. My daughter Gabriella will be 11 months on 3-22. We were very shocked when we received her diagnosis. But we are very hopeful and amazed at all of the support and prayers from friends and family. The Families of SMA will now hold a special place in our hearts. We hope to attend the walk and roll in April. We are very excited to meet others who have also had to take on the disease. Thanks again for the info and care package, my daughter loved all of the fun toys. It was like Christmas all over again when she opened it.
Margaret, Jared, and Gabriella :) of Florida

Families of SMA,
Thank you so much for David’s care package. It made his whole week. He had to sleep with everything in his bed last night and wanted to take it to his teachers to show them. We are truly appreciative.
Thank you,
Erica Perez of Michigan

FSMA!
Thank you so much for David’s care package. It made his whole week. He had to sleep with everything in his bed last night and wanted to take it to his teachers to show them. We are truly appreciative.
Thank you,
Erica Perez of Michigan

Families of SMA,
Here is a picture of Waylon in the wagon that FSMA sent us. He was so excited and wanted to go for a ride but we didn’t have it together yet.
We want to thank FSMA for sending us the care packages. Waylon wanted to open and play with everything before I could get a picture. Ellie is a little young yet to appreciate and play with everything but she loves the O-Ball and the beany baby. We also want to thank you for the wagon. We have gotten a lot of use out of it already. Both Waylon and Ellie in her car seat fit in it so it is easy for us to go for walks to the park. Being able to get out with both kids with ease is a blessing.
(Waylon is 2 years old and was diagnosed in February. Ellie is 4 months and was diagnosed in April. They have SMA Type II/III)
Thank you for everything,
Ben, Erika, Waylon, and Ellie Budtke of Wisconsin

Thank you!
A big thank you to the Families of SMA for the amazing care package you sent Kamdyn Hartung. That was such a ray of sunshine during such a difficult time for our family.
Sincerely,
Travis & Amber Hartung of Pennsylvania

Families of Spinal Muscular Atrophy,
Thank you so much for this wonderful gift. She loved the wagon. We go out EVERYDAY to swing. You all made this possible. Also, thank you for the new booklets. My family and friends are always reading them.
Thank you again,
Mary Oaks of Arkansas
I Know What You’re Thinking

Article by SMA Parent, Stephanie Geraghty, 2012 Marine Corps Spouse of the Year

Article originally featured in Military Spouse Magazine

Have you ever wondered why the little boy in the grocery store is zipping around in a wheelchair? Why the girl sitting next to you in the movies is rocking back and forth? Or why the boy in your son’s karate class does not speak?

I do. Curiosity is human nature. But along with curiosity comes questions: Do you ask? Do you not ask? What do you tell your children when they ask you?

According to the U.S. Census Bureau, approximately 56.7 million people living in the United States had some kind of disability in 2010, accounting for 18.7 percent of the entire population. The National Military Family Association currently estimates that more than 100,000 military families include members with special needs.

My point: there’s A LOT of us. My family included.

My son Cole has a genetic motor neuron disease called Spinal Muscular Atrophy (SMA). Missing genes in his DNA ultimately result in muscle weakness and atrophy, especially in his legs. Cole possesses the ability to move through crawling and cruising. He uses leg braces, a gait trainer and a power wheelchair to travel greater distances. In addition to using these resources, our family does whatever we can to meet Cole’s needs.

REACHING OUT

One day last summer, I took Cole to a local pool for some physical therapy. I briefly explained to the staff that Cole had SMA and I was interested in doing aquatic therapy with him. A young lifeguard eagerly jumped to help accommodate us. As we set up the equipment, he voiced interest in Cole’s disorder by simply asking, “What are his special needs?”

He admitted he had never heard of that disease and asked thoughtful questions without hesitation or embarrassment. I was pleasantly surprised by his honesty and was happy to respond. I described the disease and explained that physical therapy is the best and only therapy, hence our interest in aquatic exercise.

For me, that straightforward question of “What are his needs?” was an easy way to open the discussion. He made eye contact with me and asked it with genuine kindness. Another thing I loved about the dialogue: How he addressed Cole personally. He wasn’t scared to talk to Cole.

MAKING A CONNECTION

The lifeguard’s questions also gave me a chance to brag about my sweet boy. It opened the door for me to talk about how he loves to wrestle with his dad and his brother, builds creative projects with his Legos, and gets into mischief just like any other 3-year-old.

I talked about his profound ability to problem-solve and how he does not allow his disability to define him. I talked about how proud we are. The lifeguard was so inspired and impressed with Cole, he asked if he could run his upcoming half marathon in Cole’s honor. My heart soared.

What a wonderful gesture by a stranger. This honest open exchange was so easy yet so rare. Why does our society shy away from embracing and understanding differences? Every person wants to be loved and included. Awareness proves vital to the acceptance of all disabilities, and for some, it is the only way to find a cure.

That day I vowed to use my experiences to help raise awareness and foster open and honest communication about disability. I’m still learning myself, so I spent time reading and gathering responses from friends and family to explore all angles. My efforts resulted in two main conclusions:

PEOPLE ARE AFRAID TO ASK QUESTIONS

People don’t ask questions about the differences or disabilities because they do not want to say the wrong thing or cause feelings of discomfort, hurt, or awkwardness. They don’t know what to say, so they refrain from saying anything at all.

MOST FAMILIES ARE HAPPY AND WILLING TO RESPOND TO OPEN AND HONEST QUESTIONS. They welcome dialogue as a means to increase acceptance and awareness.

So, how do we bridge the gap? If you’ve just arrived at a new duty station, or you’re making plans for your next move, what can you do to make sure you’ll connect with other parents who happen to have a child with special needs?

We can aim to build confidence in discussing disabilities and fostering new relationships. My hope is that the following ideas will help you feel more confident to initiate discussion and engage in positive interactions.

FIRST, WE CAN TEACH TOLERANCE AND INCLUSION AT A YOUNG AGE TALK TO YOUR CHILDREN

Talk to them about disabilities, their own or those of others, and alleviate any fears they may have about what they may not understand. We encourage our 5-year-old son Caden to be his brother’s best friend, helper, and advocate.

LET THEM ASK QUESTIONS

I recently realized we had not discussed his interactions with other children who possess a disability. So, we discussed how God makes all of us unique and that those differences are what makes us special. We talked about despite how
someone looks or acts on the outside, we’re all the same on the inside. We
encouraged him to treat all children with
equality, love, and respect no matter their
appearance or behavior.

HELP THEM UNDERSTAND WHAT THEY CAN DO
We told Caden to include all his class-
mates in activities, to offer help whenever
a need arises, and to never poke fun at
someone for their differences. As parents,
we can all make a positive impact in our
community by taking the time to educate
our kids about appropriate conduct and
especially about the harmful effects of
bullying.

SECOND, WE CAN REACH OUT
FIND OPPORTUNITIES TO EngAGE PEOPLe
Opportunities will appear, whether at
your current duty station or at one you’re
PCS’ing to this year. Acts of kindness
toward any person, especially to a parent
or a child with special needs, can mean
the world.

LEARN BEFORE YOU DECIDE
Snap judgments, uninformed suggestions
and assumptions can be hurtful.
Following Cole’s diagnosis, we spent
countless hours researching the
disease, drug trials, and therapies.
Suggestions such as, “Did you try this”
imply the family has overlooked an
obvious solution. Instead, spend time
 gaining more insight about their
specific situation and then go a step
further and perform your own
research later.

People sometimes assume that
inadequate life choices and environ-
mental factors caused the disability.
There are countless reasons ranging
from inherited disorders to unfortu-
nate accidents. Without knowing any
of that family’s specific history, an
assumption related to the cause is
unsupported.

Whenever someone says, “I’m so
sorry,” “Bless your heart,” or “I don’t
know how you do it,” I know in my
heart that their comments come from
a good place. People have even stopped in
the middle of the street to stare. They are
recognizing and appreciating that there
are extra challenges. However, when it
comes to our children, we see perfection
and we want you to see that too. Extend
understanding instead of pity.

WISER WORDS
As Cole grows, I’ve become aware of the
language I use and how that will impact
his confidence. Through my research, I
learned that other people are guarded for
the same reason; they don’t want to say
the wrong thing. Here are a few recom-
endations for appropriate language
choices to encourage dialogue. “Handi-
capped” and “disabled” tend to hold
negative connotation.

We should all aim to omit the derogatory
use of the word “retarded” from daily
speech. A person is not a “victim” of a
disorder nor do they “suffer” from a
disability. Cole is not disabled; rather,
he has a disability. We can also be
conscious of the need versus recognizing
it as a problem.

There is nothing “wrong” with Cole’s legs
and he does not have a “problem”
walking. Rather, Cole’s legs work differ-
ently and he has a need for a power
wheelchair. These same concepts apply to
any situation. Always remember, we’re all
people first, with unique personalities,
talents, and strengths. Keep that at the
forefront.

ENGAGE DIRECTLY
If you’re curious, you can ask questions
related to the child’s assistive devices,
such as, “Are those cochlear implants?” or
“Do you mind if I ask why he needs a
wheelchair?” Out of respect, consider
asking questions when the individual is
out of earshot or ask the question to
them directly. By asking questions with
kindness, you display recognition that it
could be a sensitive topic and you
understand if they prefer to refrain from
discussion.

If your questions are not received well,
extend compassion and the benefit of the
doubt. Be patient. Chances are, maybe the
family has had a difficult day and just
wasn’t up for talking about it at that time.

I’ve learned that as Cole’s parent and
advocate I can help encourage communi-
cation too. When I initiate the conver-
sation, people seem more at ease and
willing to talk. I can set a good example
by using positive language and focusing
on what Cole can do instead of what he
cannot. I also plan to help my children
learn how to receive questions. There will
be times when words and actions will be
hurtful, but I hope to help them accept
and seize this as an opportunity to
educate and build new relationships.

Reflecting back to the day at the pool, the
lifeguard didn’t do anything extraordi-
nary. He treated us with equality and
kindness and strove to make a connec-
tion, the same things every person wants
and needs. So if you ever wonder, it’s ok
to ask! Chances are we’ll welcome the
opportunity to share. And who knows,
you might make a new friend—and you
might make someone’s day, just like that
lifeguard made mine.
Overcome Fear for a Fun New Hobby

By Jack Murphy, featured in the Genetic Alliance Magazine, June 2013

It’s not always easy coming up with hobbies you can do with your children. Our son, Connor, gives us unique challenges because he has a genetics disease called Spinal Muscular Atrophy (SMA) Type I. SMA limits his ability to move, including the use of his arms and hands.

Connor tried various hobbies, such as photography. But it was hard to set up, change lenses and film, etc. For a long time we had heard about beekeeping. Three years ago we finally decided to attend a local seminar and field course. The whole thing was far more involved than we could ever have imagined. The beekeepers were a diverse group, ranging from farmers to nuclear engineers. They all shared a common interest and language, that art of beekeeping.

We purchased a book and picked up the vocabulary. Our mentor, Larry, told us there were a lot of good books on beekeeping. The only problem is the bees haven’t read them yet! Ask four people what to do and you will get four different answers, sometimes more. But for us, this is part of what made it such a great hobby.

We started by building our hive bodies and frames. This was an enjoyable time. Connor was my glue man. I was the nail man. All the while we listened to vintage Bob Marley and discussed the universe, God, life in general, and the big “Why?” It took a long time just to build one frame. If we were getting paid by the hour or by the frame we could have not made a living! But we got a lot of father-son time, which was fun. Our goal at that stage was to construct the best hives possible, to create durable homes for the bees that would withstand the test of time, as well as any mistakes by the novice beekeepers. We had our vision: to collect honey.

Not everyone in our house was excited about our plan. My daughter, Caitlin, is scared to death of bees. She was six when we got them. “Bees?” she asked. “Aren’t those the things we run away from?”

In a way she was right. When we got the bees, the scenario changed. Our vision turned into reality, but it was not simple, or painless. Bees’ sting. “Can they sense my fear?” we wondered. We had envisioned our bees loving us and understanding that we had their best interest at heart, even if we were stealing their honey. We grew to realize that the bees would probably never love us and that a good bee suit would be an essential tool. Not only does the suit protect from bees, it gives a big confidence boost for reaching into the hive to handle the honey.

Eventually we stopped focusing on ourselves and overcame our fear. Connor is tough. I once made a mistake I will never repeat. We were just going to check on the hive, not do much. Connor’s bonnet was on, but it was not fastened or zipped. When I opened the box, I could hear the hive buzzing. The bees were getting agitated. Connor informed me that he had a bee in his bonnet. It stung him right in the temple, but he did not cry or complain about it. Shortly after, I got stung in the temple, too! And it hurt a lot. We shared that pain and discomfort like we shared the pleasure of building the frames together.

Now we are part of a beekeeping association. We go to meetings, talk with other beekeepers, and learn from their experience.
experiences. The veteran beekeepers who have been at it for decades educate us and share their war stories about dozens of stings. After our mishap with the bee in the bonnet, we know to wear full protection. The full bee suit is hard for Connor to get into and out of, so we are going to get a new suit. He has no more fear, though. He puts on the bonnet and gets right into the action. When robbing bees of their honey, all you can do is repeat to yourself, “stay calm, stay calm, stay calm.” It becomes your mantra amidst the inevitable cloud of bees.

Even Caitlin came around. Our hives were productive last year, we got three gallons of honey. We’ve been eating a lot of honey and giving a lot of it away, and we started making lip balm from the beeswax. Caitlin is involved on the production line. We melt the beeswax, clean it up, add oils, and fill up empty tubes. It makes a nice gift! When we get more wax, we hope to make candles too.

The past few years have been a learning process. We thought that understanding between us and the bees could be achieved as we rallied against common enemies. The honey could be divided later; together we would fight the dreaded hive beetle, mites and even ants. However, it seems that we will not reach any understanding with the bees, at least as long as we are taking their honey.

Even though they do not always love us back, we love our bees. They have taught us a lot about the complexity of life. We learned to stay calm in the face of a swarm of angry bees, a skill that can certainly be translated into patience and courage in other parts of life. We feed and medicate the bees, taking care of them to the best of our ability. But it doesn’t always work. We’ve had as many as four hives, but now we are down to two. The bees disappeared, abandoning honey in their hives. WE don’t know why. This year we hope to split hives so we can expand our apiary.

Above all else, the most rewarding part of it all is that Connor and I engage together in a hobby that we really enjoy. Of course, it doesn’t hurt that there’s a bit of glory in it – in 2011, our honey came in third place at the Anderson County (Tennessee) Fair!

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Helpful Hints

Families of SMA is gathering helpful hints and tips when it comes to managing activities of daily living with SMA. We hope to publish these in each issue of the Directions Newsletter and also in a possible Care Series Booklet.

• Leg sling for PVC jungle gym

In the care kit you sent us was instructions on building a PVC pipe play gym. Well, we put it together and added these legs slings. We started off with 2 wash clothes tied at the corners, then used a sheepskin, and hung them from the rings. Riley can move his legs and loves it. Just wanted to share this idea, so maybe it will work for other kids too.

Materials
2 - 6 inch x 12 inch piece of sheepskin
8 – 3/8 inch eyelets
2 – Stiff metal coat hangers
8 – Rubber caps

Instructions
Cut sheepskin into 2 pieces
Install the eyelets at each corner of sheepskin
Cut coat hangers into about 16 inches long
Bend coat hangers (see picture)
Install rubber boots on end of hanger for safety
Use string or shoe laces to hang sheepskin under jungle gym

–Elena Johnson, Alpharetta, GA
As the Byrd family piled into the reception lobby at Chase Field before a Diamondbacks game in April, the stares of strangers fixed on the two children in wheelchairs. No one said anything; that would have been rude. But the sight of 5-year-old twins Lauren and Kyle – smiling, chatting up the adults around them, showing off their fan gear – likely stirred various emotions in the onlookers: curiosity, sympathy, pity, admiration and maybe even wonder at the double misfortune, gratitude that it was not their children. But what caught my eye, what caused me to wince a little, was the machine that the twins’ grandmother was carrying. I knew right away what it was. It’s called a cough assist. The Byrd’s model was a smaller, sleeker and digital version of the bulky device we had for our son, Wyatt, but the purpose was the same: to help a child breathe because he’s too weak to clear his own throat. Staring at it, I flashed back to the last time we used ours, frantically pushing the mask against Wyatt’s face, watching his oxygen level dip to zero, feeling him go slack in our arms.

When I saw the Byrds, especially Kyle with his Diamondbacks jersey, miniature bat and kid-sized glove that his hand is too weak to hold – I felt anger. The unfairness of this disease, known as spinal muscular atrophy – to be on the wrong side of 6,400-to-1 odds – is bad enough. But to have it happen twice to the same family. How can the odds be so cruel?

‘That was like magic’

Kyle and Lauren understand they can’t do things right now that other kids can, but there’s the sweet – and even sad – naiveté of a 5-year-old that gives them hope that any day now they’ll be able to run the bases or ride a bike.

And on this day, Kyle believed he was going to play baseball. Real baseball. That was the wish he was told he’d be granted with the Arizona Diamondbacks – to be made a real member of the team for a day.

Second baseman Aaron Hill and first baseman Paul Goldschmidt, whose impressive slugging this season has him in contention for the National League Most Valuable Player award, were waiting for him at a kid-sized batting cage and baseball diamond on the concourse of Phoenix’s Chase Field. Wielding his pint-sized bat, Kyle swung toward the whiffle ball, hitting more often than missing Hill’s pitches. Then Kyle got a solid hit. “All right, there it is, go, go, go!” Hill said, as Kyle started to cruise the bases in his motorized wheelchair.

“You hear the home run call? You hear it.” said Hill, as the team’s signature home run siren blared over the loudspeaker.

“How did that happen?” said Kyle, with a look of awe on his face. “How did that happen? That was like magic!”

Later, after a tour of the clubhouse and a series of pictures with the team on the field before batting practice, came that difficult moment when Kyle’s mom and dad told him they had to leave the field.

“No, I want to play,” he said, and he frowned in anger, wheeling his power chair around as he lowered his head and tucked in his chin like a turtle receding into its shell.

Bound by a disease

I wanted this assignment for ESPN’s My Wish series, a partnership with Make-A-Wish, because the Byrds and I are bound by this disease, which I knew nothing about in July 2010 when our son, Wyatt, was born. I had never even heard of spinal muscular atrophy. When you lose a child, you try to find some meaning, some answer to the “Why us?” questions that persist. And I had vowed to do what I could to make more people aware of SMA, a devastating genetic disease for
which there is no treatment and no cure. If more women of child-bearing age and their partners knew about SMA carrier screening, there are steps they could take to avoid having a child with the disease. 

More attention could also bring more money to SMA organizations that help fund research to cure SMA and support the families already struggling with the disease.

Wyatt was just 5 months old when he died. We held him, read his favorite books, snuggled him with his teddy bear as he took his last breaths on Dec. 12, 2010. The day Wyatt died was devastating. It was just a couple weeks before Christmas; we passed homes decorated with lights on our ride to the funeral home, while I held Wyatt in my arms. I was envious of those families and the holiday celebrations they would have while we were planning our son’s funeral.

Wyatt had SMA Type I, a death sentence for most babies. SMA is a degenerative neuromuscular disease, which means muscles, including those used to eat and breathe, weaken until they can’t function anymore. It’s caused by a defect – actually a missing part – in the Survival Motor Neuron 1 gene. The defect reduces levels of a protein needed to sustain cells in a certain part of the spinal cord. It’s an autosomal recessive disease, which means that both parents must be carriers of the defect in order to pass it on to their child. With each pregnancy, there is a 1-in-4 chance the child will have SMA.

Children with Type I SMA are usually diagnosed before they’re 6 months old. Wyatt’s progression was typical. He could never hold his head up on his own, much less sit up, crawl or roll over. He could move his arms, mostly his forearms and hands, and that was about it. He quickly lost his ability to swallow, needing a feeding tube in his stomach shortly after he was finally diagnosed at 3½ months after testing for a myriad of other diseases. When he was a month old, doctors thought his irregular breathing was a respiratory virus. A few weeks later, when we noticed that one of his hands was limp and he couldn’t hold his head up, he was tested for cerebral palsy. Weeks passed, more tests, and eventually, after our pediatrician consulted with a geneticist, an SMA test was ordered.

We were in our doctor’s office, where we had brought Wyatt to get checked out for a possible cold. When I made the appointment earlier that morning, the office had called my husband to ask him to join us. As foreboding as that request was, we didn’t expect SMA, because we had been told it would take a few weeks to get the test results. Only 10 days had passed, so we thought it was something else. I was trying to get Wyatt’s clothes back on as our pediatrician said he had some bad news, that Wyatt had tested positive for SMA, and we all started crying. I was shaking so much I couldn’t even button Wyatt’s onesie, and our nanny stepped in to take over. That’s the moment it all came crashing down. All the dreams, plans, hopes, everything.

When I was pregnant, I had a funny litmus test for choosing a name. It couldn’t be too stuffy for a baby. It couldn’t be something kids could distort into a tease on the playground. It had to sound professional on a business card, proud being announced in an auditorium during graduation, and powerful when mentioned on “SportsCenter.” Those were the big dreams I had for our son, but what was most important for me was to have a child who would enjoy baking cookies, hiking in the woods and making crayon drawings for our fridge.

Big dreams

The Byrds had dreams of their own for Kyle and Lauren. Chris and Cassandra met in an AOL chat room in the late 1990s. They married in 2002 and had a little girl, Jenna, who was 3 when the twins were born in the summer of 2007. When Cassandra was pregnant, and the ultrasound tech casually asked if twins ran in their families, Chris rocketed back in the rolling office chair and Cassandra says she recalls uttering something probably unfit to print. They hadn’t known there were two, but they were happy with the news.

The day Chris Byrd found out he was having a son, his mind took off with infinite possibilities. Sports were always big in the Byrd household. Chris asked Cassandra to marry him via a message broadcast on the video board at what is now Chase Field. He had played football and volleyball at Mississippi State and avidly follows professional soccer.

“I figured, he’s my son. He’s going to sign a football contract with the Chelsea football club by the time he’s 16,” Chris says. He even humorously envisioned being the boy’s agent and one day moving to London, all the while vowing not to really push Kyle into any sport he didn’t choose on his own.

But those scenarios all changed with the diagnosis.

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“That dream, it went out the door and it went out the door pretty fast. He’s never going to ride a bike. He’s never going to be able to sit on the toilet on his own.”

Chris and Cassandra Byrd, like many other SMA parents, also went through several months of mystery in trying to figure out what was wrong with Kyle and Lauren. Though there were vague hints of trouble early on (in retrospect, Chris says the children were never fond of “tummy time,” the practice of putting weeks-old babies on their stomachs to encourage them to develop neck muscles; he remembers them lying with their heads down in snot and tears), the first real warning sign came the day after Christmas in 2007 when the twins were 6 months old and not even close to being able to sit up on their own, something that most children can do at that age. Chris and Cassandra were encouraged to be patient. They would be told the same in regard to pulling up to stand, with doctors and other parents constantly reminding them that all children develop at their own pace. Cassandra says her concern escalated when the children’s grandfather noted that Kyle didn’t seem to be able to support any weight on his feet when he held the boy on his lap and tried to help Kyle stand. But at the twins’ 9-month checkup, their pediatrician told Chris and Cassandra to hang tight and see what happened when the twins turned 1.

When that deadline rolled around, and they still weren’t sitting up – much less crawling – everyone knew something was wrong, and so began a litany of possible diagnoses that led to tests, that led to waiting on results, that led to more dead ends. Doctors ruled out all sorts of maladies, including leukemia, before the Byrds finally were referred to a pediatric neurologist when the twins were 16 months old.

“The neurologist took two steps inside that room, and before he even shook our hands, he knew it was SMA,” Cassandra says. Medical staff drew blood from both children, advised their parents of the wait for results, and a nurse sent them home with one directive: Stay off of Google.

As it turned out, Kyle and Lauren had SMA Type II, which is better – in a relative sense – than having the more common Type I. The nurse didn’t want the Byrds to be scared by all the information they would see if they read about the worst-case scenario – the Type I babies, like our Wyatt. Type II children have a better prognosis. Their lifespans vary widely, with some living only a few years and others making it as far as 40 or 50. The Byrds said they’re hopeful that the twins will at least reach young adulthood.

SM A doesn’t affect the brain, so an SMA child’s mental capacity is the same as a healthy child’s, and scientific studies have shown that older children with SMA tend to be more intelligent than children without SMA.

As Type IIs, Kyle and Lauren can hold up their heads while sitting upright, can move with a range of upper-body motions and can breathe on their own. They do need special machines to help them clear their throats, and they wear a breathing mask hooked up to a type of ventilator overnight while they sleep. They can still eat regularly by mouth, but they get extra nutrition via a feeding tube in their stomachs. Orthotic braces keep their legs and feet aligned correctly.

These things are commonplace to the Byrds now. Four years ago, they had no idea what to expect when they first got the SMA diagnosis.

Although many mothers, me included, would hear that directive to stay off the Internet and then head straight to their computers, Cassandra said she didn’t. She willed herself to avoid the Internet as she had during most of their struggle to get a diagnosis, and kept her mind busy with the day-to-day tasks of caring for two babies and a young daughter.

It was only at night, when Cassandra would go into each baby’s room, gather each one in her lap to nurse and rock quietly, that she would let go, and cry quietly.

“I was just looking at this child and saying, ‘I don’t know what’s in your future. I just know I have to help you get through this,’” she says.

The best life possible

The vast unknown is something all SMA parents struggle with, especially with a disease for which there really isn’t any treatment and no hope their children will get better, save for some medical miracle. Pretty much all that can be done is to manage symptoms and wait for the next emergency. At first that was a huge roadblock for the Byrds; Chris says he wasn’t sure whether they should be making plans for a funeral, or saving for college.

Because the more severe SMA Type I is more common, that’s much of what the Byrds came across in those first few weeks of diagnosis – and almost all they read and heard was scary. It was tough just to look at the pictures, images of babies with
tracheostomy tubes coming out of their necks and toddlers lying supine on special reclining wheelchairs with frozen looks on their faces because they had lost the ability to smile.

But as Chris and Cassandra met other Type II families, they almost started seeing themselves as somewhat fortunate. Yes, their children were in wheelchairs, but at least they could use a wheelchair. At least they could blow bubbles and smile and laugh with their friends. Since SMA doesn’t affect the brain, Kyle and Lauren can do all the things other kids do intellectually – they read lots of books, play on their iPad and go to school. And they’ve developed the twin bond. The Diamondbacks experience was Kyle’s wish, but it was almost impossible to separate him from his sister. Even in the middle of his favorite moment of the weekend – playing in the kid-sized baseball diamond with two of his favorite players – he asked if Lauren could have a turn hitting.

As the Byrds watched their twins grow into toddlers and then into kindergartners, with their inability to walk, their power chairs, the funny way their hands drooped and the other physical manifestations of their disease becoming apparent, Chris and Cassandra adapted to something familiar to any parent of a special-needs child – something called “the new normal.” The disease transformed from an earth-shattering, life-changing event to part of the Byrds’ day-to-day routine: Tasks like loading the twins’ power chairs into their adapted minivan, waking up several times a night to turn Kyle or Lauren in their beds or adjust their breathing masks, and changing their diapers became normal for Chris and Cassandra.

“You accept the cards you’ve been dealt. As their dad, I’m going to give them the best life possible and make them as comfortable as possible and get them to live as close to what society would deem a normal life as possible,” says Chris, who coincidentally works in the field of medical research. “Any piece of equipment. Any surgical procedure. That’s what my job is now. It’s not to get them into the best soccer clinic.”

When the disease is out of your hands, you control what you can. Although Wyatt’s case was more dire and our time with him shorter, our mantra was the same as Chris’: to give him the best life possible. Even though Wyatt was only an infant, we did what we could. He played with a penguin on a trip to the zoo. He “rode” a horse on his grandparents’ ranch. He enjoyed a special visit with Santa a few days before he died. We made a conscious, and – to some – controversial decision not to use any invasive means to prolong Wyatt’s life. We did not go to the hospital. We used few machines. When it became harder and harder to revive Wyatt and to keep him breathing, we knew it was time to let go.

The Byrds told me about the Type I children they’ve met – including families with more than one Type I child - and how seeing others’ struggle has made them feel almost fortunate. I’m not sure how to take that, not knowing whether their situation is any better than mine. How can you judge better or worse in a disease like this? There are no guarantees.

The Byrds had a scare earlier this year when Lauren passed out in her power chair, stopped breathing and was unresponsive for two minutes because she couldn’t clear the mucus caught in her throat. They hate to think what might have happened had they not been in the parking lot of an urgent care facility, where they had gone to get an infection on Lauren’s lip checked out. In February 2009, when the twins were 20 months old, they were in the hospital for two weeks with a respiratory virus that lots of toddlers get, but one that is potentially fatal for an SMA child.

“They’re pretty strong-appearing SMA kids, but that doesn’t mean that one cold won’t turn into pneumonia, won’t put them in the pediatric intensive care unit and they’ll die,” Chris says. “I don’t want to sound morbid, but that’s what goes through your head as the parent of a kid with SMA.”

Testing is vital

That uncertainty is part of why Kyle and Lauren got their Make-A-Wish requests fulfilled at a relatively young age. Lauren asked for and got a trip to Disney World. Kyle’s choice was totally expected. His room is a Diamondbacks shrine. Baseball bobbleheads jockey for space on his headboard alongside autographed baseballs. He has a wardrobe of Diamondbacks T-shirts. He learned the rules by watching games on television, listening to them on the radio and playing baseball on his Nintendo Wii. When the family went to the games, and Lauren and Jenna were begging for cotton candy or snow cones, Kyle’s gaze

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would be fixed on the field.
There was irony in his father’s realization: “It can easily be taken the wrong way when I say, ‘He’s the son I’ve always wanted.’ But just from the sports, the fact that he gravitated toward sports, is really what I mean.”

The tough part is knowing Kyle will never be able to participate fully in the sports he loves. It was evident in Kyle’s frown and in his refusal to eat lunch with his family after he had to leave the stadium. The clubhouse locker with a Byrd No. 5 placard, his name on the roster, a special Byrd Diamondbacks jersey and even his own player bobblehead had made him smile, but Kyle’s focus remained on the game itself.

“He literally thought he was going to play today,” says his dad. “He probably doesn’t understand he’s never going to play baseball. He’s literally never going to be able to run the bases.”

The questions are going to start any day now, and the sorrowful realization will soon follow.

“He’s about to get to that age where he is going to start asking ‘Why?’ himself. And I don’t have an answer for that,” Chris says. “This is the one where I don’t know if I’ll really have the answers for that. I just don’t know. I don’t know why.”

“He knows he can’t hit a home run because his arms are too weak – but he thinks maybe if he practices,” Cassandra says. “So I think there’s some hopefulness there. It’s a delicate balance to keep him realistic but still nurture the what-ifs, the fantasy.”

Even though the Byrds are optimistic for a cure – and there is promising research – they know it will likely come too late for Kyle and Lauren.

“They’ve declined so much physically that if a magic cure came tomorrow, it wouldn’t fix all the physical problems. You can’t fix scoliosis, the muscle contractions, the weak lungs,” Cassandra says. “It

He sat there, surrounded by Eaton and Diamondbacks infielder Willie Bloomquist and two camera crews, when the announcer broke in: “And now, throwing out the ceremonial first pitch, Arizona Diamondback No. 5, Kyle Byrd.” Kyle gingerly balanced the ball in his left hand, raised his forearm and threw with all his might. The ball landed just a few feet in front of his wheelchair.

As a loud cheer rose from the stands, I looked up at the crowd. To the fans this was probably just another nice thing: How sweet that that little boy got to throw out the first pitch. I wanted to shake them, to make them realize that probably one in every 40 of them sitting out there carried this defective gene that could put their future children at risk.

I want them to get tested. I want them to know what SMA is. I want them to donate money to find a cure.

I want them to know what just one day of life is like for Chris and Cassandra. I want them to see the Byrds changing diapers for twins who are now 6 years old, waking up several times a night to turn Kyle or adjust Lauren’s breathing mask, or loading up the van with two wheelchairs and an assortment of machines for every outing. I want them to know Wyatt, the sweet little boy with blue eyes and blond hair and a sly smile, who loved sweet potatoes and books and glittery toys, but who never had a chance.

I want them to realize how fortunate they are.

I do now. Nine months after Wyatt died, I got my wish. I was pregnant again. Healthy twins. A boy and a girl, who are just starting to walk.

Please visit the following link to view the article on ESPN.com:
Wait? Where am I? How did I get here? Looking to my left and then to my right I see nobody. I am truly alone. No people, no cars, no buildings; just me, the beach, and the power of the ocean in front of me. Not even the sound of squawking seagulls overhead can be heard. As I sit in my wheelchair, face to face with the big blue ocean I begin to feel a sense of power building up inside of me. Its as if the ocean, with every crash of a wave, feeds another surge of energy straight to my soul. I look up to the heavens to receive the blessings and to thank God but suddenly I feel a cool breeze brush over my skin along with the sound of a low rumbling. I slowly lower my head to look onto the horizon. A swell in the ocean began to form in the far distance. It was as wide as my eyes could see. The whole shoreline grew in size in front of me. Fish began to flip and flop helplessly on the now dry sea bed. It was as if God himself lifted up the ocean water and pushed it backwards. But, the swell became larger and larger over time and I could tell it was coming straight towards me. In seconds the swell towered over the horizon and began to gain so much speed that I was able to feel the mist falling off the crest of this massive wall of water. As the wave rumbled and sprayed its mist down onto my head it suddenly stopped. There I was, the size of an ant, face to face with a wave so tall it reached straight into the heavens. Then I awoke from my dream.
My name is Aaron Schindler, I live in California, I have SMA Type III, and I am the Director of “Why The Sky Is Blue”; the first full-length 3D animated movie created in Central California.

My parents have told me that I was diagnosed at first with Werndig-Hoffman at infancy and later re-diagnosed with Kugelberg-Welander. If you ask me I think the doctor didn’t know what he was talking about in the first place. The doctor (who isn’t a doctor any longer) told my parents that they might as well put me in a special home cause I wouldn’t live long or amount to anything. Boy would I love to slap him now!

Growing up with SMA was never really a challenge for me. I had the normal upsets that any kid would have growing up with a disease. Not being able to do the same stuff as other kids my age did sucked at times along with having to deal with bullies (I handled it by running them over). However, I was fortunate to have parents that didn’t treat me like I was handicapped. If I wanted something they’d say “your not handicapped, go get it yourself.” And of course, they told me in a joking manner and with lots of love. Little did we know that the independent monster would rear up and bite me in the butt.

In high school I wasn’t the best behaved kid. I would not do my homework, skip classes to chill out with girls in bikinis...oops sorry gotta remember, this is a family newsletter! Kids ignore that last sentence. Anyways, I found my love for computer graphics in high school. And when I was told I could make money doing it! Wow, I thought life couldn’t get any better than that. Whenever I was home anyone could find me in my room, in front of my computer creating art. It first started with logos and print graphics. From there it went crazy. As I matured (yea right) and went off to college, I got into 3D animation and visual effects. Four movies later, and I’m directing my first full-length movie.

Why the Sky is Blue is a family focused, animated movie full of fun, adventure, and trails. Kalani is a 13 year old boy growing up in the 1950’s on an island in the Pacific. Together with his precocious younger sister, Malana, they face the typical and not so typical problems of growing up in a single parent home. Daily Kalani must also face the school bully and the tough trials of school that still resonate today. Through all the obstacles life is throwing him, just when he thinks he will never be more than just the poor island boy, he learns he has been chosen by the gods to surf the great wave and keep the sky blue.

The movie is based off of my dream I had way back in 2001. I played with the idea of writing it as a childrens story. Then I forgot about it all together at one point. But when I met the woman of my dreams she convinced me to make it into a movie. Nita Rose, now Nita Schindler (whom I married on June 22nd of this year! Woot!) had a college background in Secondary English Education, so she knew how to develop character profiles and add plenty of details to my story. Together we worked over the internet to develop the 78 page script for the movie. It still makes me laugh when I think about some of those script writing days. Nita lived in Illinois at the time and when she would come home from work, we would talk and I would explain how Aolani (one of our movie characters) did this or said that. It was like our family instantly grew and the characters we invented were our kids.

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After the script, our film family grew even more! We now have Paul McGill, from California, who is our Character Designer and Art Director, Crystal Pilker, from Canada, that's in charge of Marketing, Curt Douglas, from West Virginia, our Programmer (a.k.a. The guy that makes it go), and Alyssa Phillips, also from California, that does the Public Relations. Sorry had to do a shout out. What-sup family!!! I would have never dreamed of this incredible family.

Nobody knows how far their dreams will take them and sometimes we have dreams that are so confusing or shocking we don't know what exactly to do at that time. But as they saying goes: If life throws you lemons, you make lemonade. To the parents that have children with SMA?..MAKE LEMONADE! I won't lie to you, it will not be easy You have the biggest challenge in your life ahead of you. However, when did anything worth doing ever get to be easy? I guarantee if you surround your child and yourself with loved ones; as your child with SMA grows, even if for an unfortunate short time, you and everyone around that child will learn something. You'll learn that life isn't about how long you live or how much wisdom you have, or even how many possessions you own; its how you lived it. And if you can grasp that... then while your child rides the wave of life, eventually ending up in heaven, you can be sipping on that sweet glass of lemonade wherever your at.
SMA Awareness Month

Families of Spinal Muscular Atrophy has been coordinating a National Awareness Month for SMA since 1996. Raising awareness of SMA in the general public can help lead to increased resources for SMA research and better care for SMA patients. The majority of people, including doctors, nurses and community members, do not know about SMA until it directly affects them.

One of the highlights of SMA Awareness month is the SMA Candle Lighting. The Annual SMA Candle Lighting was held on Saturday, August 10th. Many families and SMA organizations around the country participated by lighting a candle at sunset to remember those SMA Angels who have lost their battle with SMA and to honor those SMA Warriors who are still here fighting everyday! Here are the great pictures that were posted to the Families of SMA Facebook page!
SMA AWARENESS – SMA CANDLE LIGHTING
These booklets are available on a variety of specific topics such as Genetics and Diagnosis, Respiratory Care, Nutrition and many more. SMA Care Series Booklets are also now available in Spanish, as well as on our website for download.

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

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

This section is designed so it can be removed from the center of the newsletter.

Photo of: Tonya Willingham and her daughter, Hanna
Reflection on Grief

by Liz Bahrenburg

Today is June 21, 2013. It’s been exactly 5 years since our sweet Skylar became an angel, but my grief is still alive. Losing a child to Spinal Muscular Atrophy (SMA) creates a very complex grieving process. These are smart, vibrant, and loving kids with huge personalities stuck in weak bodies. It is difficult to place your feelings while you grieve. You have the typical emotions of heartbreak, gut-wrenching sadness and profound loss but tied up in these emotions are guilt and a sense of relief. Guilt that you didn’t do enough and that somehow his passing away is your fault... Even more guilt stems from your sense of relief (not for yourself) but relief for your child... he won’t go through more pain on a daily basis, more life-threatening, invasive surgeries, and endless hours of respiratory therapy when he should be out playing with his friends. He won’t suffer disappointment and heartbreak when he can’t keep up with his friends because his useless body is an impediment even with all the latest technologies in mobility.

As I sit here and reflect, there is one memory forever etched on my heart that epitomizes these complicated feelings:

I see Skylar racing up the ramp with Marco. His ear-to-ear smile and joy radiate off the glare of his bright yellow powerchair. Suddenly Skylar stops on a dime. The ramp has ended. A narrow gangway, barely wide enough for a child to navigate much less a 250 pound powerchair, beckons the children to board the boat. Skylar is stuck. Marco, caught up in his glee, races ahead. Children are scrambling around Skylar as they spring onto the gangway. Marco is gone, lost in a sea of exuberant children.

My gut tightens, tears spring to my eyes. I squeeze my eyes shut trying to hold them at bay so Skylar won’t see my distress. Kids’ laughter permeates my thoughts...”Come back Marco”, Skylar whines.

“We’re partners! We’re supposed to stay together!” he cries out to no one but the wind rolling off the San Francisco Bay.

“Marco Come Back!” he tries to convey his message across the boat... but the sound is weak and is drowned out by the cacophony of children’s voices.

My salty tears run into my mouth, their bitterness alarms me. I had not recognized their descent, I was so fixated on Skylar’s fallen face. Head down, utterly defeated, Skylar rolls his chair back down the ramp toward me.

“We’re supposed to stay together...” he whispers as a single tear rolls down his cheek.

“Skylar”, I say gently. “Let Marco have this time. You two have been together all day. He’ll be back soon.” I try to distract him with a hug and offer him a snack.

The light is bright, highlighting Marco and the other children as they bound around the boat. They steer it to new adventures and bring up the nets bursting with bright red plastic lobsters. It is just another day at sea for them, while Skylar sits in the dejected shadows below the bow.

I want Skylar to be healthy! I want him to have strong legs so he can run with his friends! I try to use anger to stem my tears. Who developed this discovery museum? What were they thinking? Why can’t the damn boat be accessible? My anger at the unfairness of the situation can not erase the deep sadness I feel for Skylar. In his seven years, with all that he deals with on a daily basis, this is truly the first time I have seen him suffer.

I am at a loss. I feel helpless. I can’t protect him from hurt. Skylar’s white face is starkly contrasted by the red rivulets running down his cheeks...

“Come back Marco! We’re supposed to be buddies”, he continuously calls to the air.

My broken heart aches.

Suddenly Marco comes into view. He is searching for something... someone... He spies Skylar below him. His huge smile falls away, replaced by an expression of horror. He leaps off the boat and sprints down the ramp. He launches himself at Skylar and throws his arms around him....

“I’m so sorry Skylar. Please don’t cry. I’m sorry!” he ardently exclaims! “I promise I will never leave you again!” His own tears joining Skylar’s in a waterfall of emotion.

I finally give in to my tears and they fall freely - heartbreak, relief, sadness, happiness all rolled in together. Skylar will be ok. He is strong, independent and loved!

There is nothing for me to say... just watch and admire the magical bond between two seven year old boys who are wise beyond their years and have a friendship that will last beyond a lifetime...

Thank you to Marco, our family, and friends for keeping Skylar’s memory alive! This is what carries us forward on difficult days such as today.

Thank you for embracing our cause Reach4Sky to raise money for Families of Spinal Muscular Atrophy so that one day SMA will be eradicated and other families will not have to suffer the heartbreak and devastation of losing a child.
Early in the morning on 5/11/2013 Logan Red Ruth passed peacefully in Mommy’s arms with Daddy holding his hand and his armed wrapped around both of us. He was 10 months 5 days old. Our hearts are heavy as there is a giant void in our life now. However, Logan is running free, somewhere beyond the moon. This flawed body on earth no longer restrains his beautiful spirit. He is free to sit up, crawl, walk, run and play to his heart’s content. We already miss our baby boy terribly, but we will go forward in life better than before because he came into our lives.

**Special poem written just for Logan:**
My beautiful baby boy,
You left us much too soon
We know now you run wild and free
Somewhere beyond the silvery moon.
Although you never said a word,
Not even, “I love you”
We know the depth of love you had
For in your eyes, it sure rang true.
I’ll never forget the fun we had,
like dancing in the rain
or feeding giraffes at the zoo
You really loved their train!
We’ll always remember your radiant smile,
your little fingers and toes.
The way you laughed
at the breaking foam planes
erased our hearts of woes.
And now, until my time is done
Please find my soul within
And give me peace throughout the days
With the greatest love there has ever been

*By Tia Ruth - In Honor of Logan*
Rewind to July 6, 2010: I was getting Savannah dressed to go to the Pediatrician for her one month follow up. With matching watermelon outfit, shoes and bow, I was a proud mother ready to show off my daughter to the doctors and nurses. An hour later, I was hit with words I had never heard, Spinal Muscular Atrophy. All I remember hearing was my doctor say, “Savannah most likely will not live to see her 2nd birthday.” At that moment, well a little later after I scared the entire offices with my wailing, I made two decisions. One as Savannah’s mother, I would do anything and everything possible to give her the best quality of life despite this devastating disease. Two, I was going to make sure I had no regrets about the time and life I did share with her on this earth. Many would say to me in the two years following what a wonderful mom I was to Savannah. But the real truth was how gracious Savannah was to me. I learned so many things in my journey with her and SMA. I was humbled daily watching her endure such suffering. On the outside many would see the beautiful pictures of Savannah. However for a weak SMA Type I child daily life is full of machines and struggle to breath. Despite all that Savannah had a will of her own, she always managed a smile and in her precious way would let me know, it’s okay mom, I know you are doing your best.

Savannah loved unconditionally, she forgave instantly, she saw the good in everything and everybody. Being in her presence was like getting to experience a slice of Heaven. So it’s no wonder my life was changed in our walk together. I realized that this time we have on earth is too short for anger, for unforgiveness and wasting time and energy on worry or fear. I remember promising Savannah as I rocked her precious body the day she left me. I promised I would live my life from that day forward honoring her and to be the kind of person she lived her life being. For those of you reading this, perhaps you know the pain of loss or maybe you just recently got the shocking news that your child has SMA or your journey with your SMA child leaves you weary at times, wherever you are in your timeline with SMA remember this as I look at Savannah’s dates June 4, 2010 - April 12, 2013, what pops out at me the most is the dash between the dates. Such a simple symbol but so powerful because it’s how we live our life in between those dates that will ultimately count. Savannah’s time was short but her legacy; her dash will be forever remembered.

I love you sweet girl, you are gone from my sight but live forever in my heart.
IN MEMORIAM

HADLEY ROSE YATES
November 27, 2012 – April 13, 2013

Dear Families of SMA,
I really don’t know where to begin to thank you for all that you have done for our family since we got our precious Hadley’s diagnosis: the binder of information, the newsletters, the wonderful care package, the car bed and the beautiful book and Willow Tree angel. Your mission to provide a “family” of support throughout a time when our world was torn into pieces truly blessed us. Thank you so much for all you do!
Thank you for the passion and constant drive you have for finding a cure so no other family has to go through this and experience the agony and pain of losing a child. Our Hadley Rose was the greatest blessing and gift to our family and we are so thankful God entrusted her to us for 4.5 beautiful months. Thank you again for blessing our family as you have.
Love,
The Yates family from Alabama
David, Abby, Harper Ann & our angel Hadley Rose

IN MEMORIAM

BROOKLYN ELIZABETH BOUCHARD
September 11, 2012 – March 9, 2013

Brooklyn Elizabeth was born on September 11, 2012; a bright shining beautiful baby girl! She smiled from ear to ear and had the bluest eyes. When Brooklyn was 2 months old we got her diagnosis of SMA Type I. We kept Brooklyn home with no machines and promised to give her the best life while she was here and to live as normal as we could. We played, sang, danced, painted nails, played dressed up and even tried all kinds of food. Brooklyn was always happy. She was just two days shy of her 6 month birthday when she earned her angel wings on March 9, 2013. Brooklyn will always be our bright star; she will forever be loved and missed, but never forgotten. We love you muffin!

IN MEMORIAM

ISABELLA NICOLE KUREK
December 26, 2003 – October 13, 2010

“A Beautiful Life”
A beautiful life
that came to an end,
she died as she lived,
everyone’s friend.
In our hearts a memory
will always be kept,
of one we loved, and
will never forget.

You have given me love stronger
than any love I’ve ever felt
I will cherish those short months
we were together
The feel of your soft skin
tiny face, toes and fingers
Tears fill my eyes with joy and
sorrow, for I miss you
baby girl...
You must sleep now
so please forgive me
for I must say
goodbye.
Karson Michael Riggs was born on April 12th 2008. The day he was born was one of the happiest days of our lives. Shortly after he was born we knew something was wrong. He was diagnosed with Spinal Muscular Atrophy on June 2nd 2008. From that day forward we had to find our new “normal”. Karson was a very happy little boy and we could see it in his beautiful eyes and his quiet laugh (because it was hard to get enough air) he did have loud squeals though. SMA brought different challenges to Karson. He faced them with strength and perseverance and never gave up. He was such a happy little boy. I can’t even count how many times he stopped breathing. He could have easily given up but he didn’t. One of the many lessons that Karson taught us is that true strength has nothing to do with how strong your muscles are. He fought so hard to be with us, so we fought hard for him.

From the moment that Karson was diagnosed, we knew that our time with him would be short. We always prayed that when he did pass that it wouldn’t be in a traumatic way and that he would tell us when it was time. On January 13th 2011, Karson passed away in our arms peacefully.

Audrey was a beautiful little girl who lost her battle while at the Annual SMA Conference in Disneyland this past June. Her favorite song was “It’s a Small World” and she was so excited to be at Disney, to hear the song and be able to ride the It’s a Small World ride. After the Fireworks Show on Friday evening, Disney allowed our attendees to stay in the parks until their closing. Audrey’s parents made the decision to stay after and explore Disney. They headed for It’s a Small World, where Audrey was able to experience this very special ride and hear her favorite song. The next day, Audrey passed away. Audrey and her family were in the hearts and minds of all families and attendees.

Mighty Mathis Shakespaere went back to his home in heaven today. Growing a mustache was on his ‘hope list’. He is loved and will be greatly missed.
He was our hope and dream, our answered prayer, our beautiful boy, and our perfect son. He made us parents for the first time and taught us what it means to be responsible. His eyes pierced us immediately after birth, and comforted us until his passing. His innocence captivated our hearts and revealed to us what the purity of Love looks like. His personality was the perfect melody and captivated our hearts and revealed to us our days were always subject to the cadence of his heart. His morning smile was endless and his kisses unconditional. With the lifting of a brow he asked and commanded our attention he truly desired. Without ever saying a word, he delicately controlled our every step. It’s difficult to explain what it felt like to be told your world would end because that’s what if felt like when we received the doctor’s call, we can only share with you that we understand what it feels like to be crushed, silenced, numb. It took us hours of conversation in prayer to understand there has to be a test to be a testimony. We immediately began to see life differently. We’ve always been the type to expect miracles to look like mountains moving, crippled walking, the blind seeing. We’ve been missing the obvious, that life itself is a miracle. Yes, we briefly questioned God, only to feel cowardly because we know God gave his only son to take our place. As we sat alone in our son’s room crying out to God for direction, for peace and comfort for our family, we began to assume a responsibility that is our own as Christians. You see, God never promised there would be no hardships. It’s easy to get caught up in the difficult moment and miss all the details and purpose behind the experience. Our trust, relationship and Christian foundation was definitely tested. How we reacted was totally up to us. The only way we knew we would get through this is in prayer and by changing our perspective of the situation. We made a choice to love, sacrifice, and learn. In the 6 months my son lived in this world, he touched more lives than some of us do in a lifetime without saying a word. I like to say he was a man’s man, a Champion, not one who boasts physical strength, but one who holds no fear. He was a strong willed baby. It’s the little things he taught us, we should feel privileged to be able to hug or shake someone’s hand; I hope you’re understanding. My son never fussed about anything; he faced every one of his days with a smile first thing in the morning and kept it that way all day. He didn’t know anger, resentment, failure, he only understood love. It’s important we understand this about children. They are innocent in mind, spirit, and in truth. Everyone who met my son understood love, gave love, or received love. His eyes were different. I’ve heard from many people that his eyes were heavenly, that’s just confirmation that he came here with a purpose. I know a couple of people besides my wife and I who have confessed Taylor touched there lives and that’s what this is all about. We must understand there are things we cannot control and we leave that to Gods sovereignty, but there are things we can control. We can choose to love, forgive, and bless among other things. The life of Taylor has been a process of revelation and confirmation. Many of you may have questions, all I can say to that is I pray for wisdom and receive trouble, only to realize that’s what wisdom is all about. I can guarantee you that I am now that much more sensitive towards the needs of others, and wiser about many things. We are nothing without God, but an object. My son is in heaven with no worries, pain, anger, resentment, or regret. And if our mission is to one day make it to heaven, I don’t blame him for getting a head start. It’s a privilege to know my son is with his Father. Believe me, if our love could have saved my son he would still be here, but because His love saved my son, he will live forever. Yes, God heals, he’s deposited in us the power of prayer, but he also wants to know your heart and your actions when the healing doesn’t come. Will you praise him in the storm? God continues to be our healer, I fully trust that he will heal our hearts and we will always have happy thoughts when we think of Taylor. I don’t have questions for God; I only have questions for me. What did I learn from this? How can I be a better person because of this? How can I use this testimony for good? How can I love more? How will Taylor be remembered? I don’t have to understand the sovereignty of the Lord but these things I know for sure, God’s love is pure, His sovereignty is supernatural, His promises hold true. Matthew 11:28 “Come to me, all who are weary and heavily-laden, and I will give you rest.” God’s will is perfect! My son was tailored for us, he didn’t have excuses, and neither will we. Our little angel, Our pride and joy, your life blessed us. We miss you and will be better people because of you. I love you pa.

I pray that you all be blessed with love, kindness, and sensitivity for others.

Nieves Gomez &Valerie Gomez-Valdez
Our sweet, precious little girl was freed from Spinal Muscular Atrophy (SMA) and RSV while being held in her Mommy’s arms early Tuesday morning. She is now playing freely amongst the Heavens.

Blakely was such a blessing in our lives and to all who had the chance to share her with us. Her strength, determination, and courage have brought hope and optimism to so many around her.

Blakely loved watching Disney movies, especially Tangled and the TinkerBell series, also anything on TV with Mommy and Daddy (even soccer), she loved the outdoors and would often squeal with delight when we asked if she wanted to go outside or take a walk. She loved taking bashes and car rides, shopping, doing crafts with Mommy, helping Mommy and Daddy make dinner, swimming in her little pool, hanging out with her cousins, watching birdies from Daddy’s desk and watching her “sisters” Molly and Denver play, petting Pumpkin her favorite kitty and having her “big girl” comforter pulled over her for nite-nite. But her most favorite thing of all was to be held in the arms of her Mommy, whether it was being read to before bed or walking around the yard looking at flowers. Some of her adventures included: going to Jackson Hole and Yellowstone National Park, Daniel’s Summit Lodge, the Hogle Zoo, the Museum of Natural History, a private showing of Wreck-It-Ralph, visiting the Shark Reef in Las Vegas, driving on HWY 101 along the coast of Central California, stopping along the way to walk around Solvang and the Santa Barbara wharf.

We have so many incredible memories of this little princess, who has changed our lives forever.

We would like to give sincere thanks to the many people that made a huge impact on Blakely and our lives: the staff at Primary Children’s, especially her RT’s Kaylee, Tanner, Sandrelle and Michelle who fought so valiantly to keep her here with us; her nurses; the Rainbow Kids Team; Dr. Swoboda and the SMA team; her pediatrician Dr. Baranko; our therapist Leah Jaramillo, who helped us grow immensely as a family; Weber/Morgan Early Intervention Program, specifically her OT, Teresa, who she loved to play with and her PT Lisa for helping us get many of the things that made life easier for us; the thoughtful staff at the Ogden Main Branch of America First Credit Union and the South Ogden City Fire Dept.

Blakely is survived by her Mommy and Daddy, Janell and Elliot; her Great Grandma Bitner, her Great Grandma and Grandpa Milne, her Great Grandma Thompson; her Grandparents Greg and Barb Milne, Sally Lewis, and Chuck Lewis and Iran Lavasani; her Aunt Jaquelyn and Uncle Aaron Miller (Emma, Anna, Sara, Megan, and Madelyn), Aunt Jennifer and Uncle Duane Bickmore (Samantha, Sydney, Sari, and Hunter), and Uncle Scott and Aunt Carla Lewis; and her adopted Uncle Troy Thompson.
The International Spinal Muscular Atrophy Patient Registry

The International Spinal Muscular Atrophy Patient Registry was established in 1986 and is maintained at Indiana University. The purpose of the Registry is to function as an intermediary between individuals and families affected by Spinal Muscular Atrophy (SMA) and researchers who are interested in studying SMA. The Registry centralizes information on this rare genetic disease, allowing researchers to easily recruit participants for their studies and giving families a way to learn about new research studies. Currently, the Registry contains information from 2,800 families and over 3,200 individuals with SMA from all over the world and continues to grow. The Registry is funded by Families of SMA.

Because SMA is a relatively rare disorder, it is sometimes difficult for researchers to find adequate numbers of qualifying research participants to perform clinical trials and other studies. We believe that the greatest hope for treatment in SMA is through research. Individuals and families affected by SMA are invited to join the Registry. Participants are asked to complete questionnaires about the symptoms, treatment, medications, and other experiences with SMA.

In preparation for drug trials anticipated to start in the next few years, the Registry is stepping up outreach efforts. At the same time, we’re also supporting ongoing research efforts. The following studies are currently recruiting through the Registry:

• **Evaluating the Relationship between Fetal Movements in Pregnancy and Subsequent Diagnosis of Spinal Muscular Atrophy**
  This is a research study exploring prenatal factors of individuals with SMA, particularly decreased fetal movement during pregnancy and genetic screening. This study is currently enrolling women aged 18 and over who are the biological mother of an individual with SMA (living or deceased).

• **A Pilot Study of the Natural History of Infants with Spinal Muscular Atrophy (SMA) Type I**
  The purpose of this research is to learn more about how the disease progresses in first two years of life. This study is currently enrolling children with SMA Type I born on or after January 1, 2007.

• **Surface EMG Examination of Spinal Muscular Atrophy**
  This is a study on the efficacy of surface EMG examinations. Participants will have surface electrodes placed on their skin to record muscle signals in the biceps, hand muscles or leg muscles. This study is currently enrolling children, aged 3-18 years who are healthy (to serve as controls) or have a diagnosis of SMA Type III. Participation involves a single 60-90 minute examination at a Chicago, IL area facility.

• **Involvement versus Participation in Leisure Activities in Youth with SMA (I PLAY SMA)**
  This is a research study exploring the differences in preferences and actual involvement in leisure activities and participation in children with SMA. This study is currently enrolling children, aged 2-21 years with a diagnosis of SMA Type I, II, III, or IV. Eligible participants will be sent a 30-60 minute survey, which can be completed at home.

• **Isolation and Characterization of Human Embryonic Stem Cells Carrying Disease Genes Obtained from IVF Clinics**
  This is a study that derives disease specific human embryonic stem cell lines that can be used to study human genetic diseases. These cell lines are obtained by using embryos that have undergone a procedure called pre-implantation genetic diagnosis (PGD).

For more information about the Registry or the studies listed above, please visit [https://smaregistry.iu.edu](https://smaregistry.iu.edu), email smareg@iupui.edu, or call 1-866-482-0248.
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Have you ever thought about starting a chapter?

We want to hear from you.

Send an email to chapters@fsma.org to receive more information on how to start a chapter in your state.

FSMA Chapters

The mission of our Chapters is to support families and fundraise for SMA, giving hope to families in their community. Chapter fundraisers include Walk-n-Rolls, golf tournaments, gala events and more. 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.

Send an email to chapters@fsma.org to receive more information on how to start a chapter in your state.
Promotional Materials from Families of SMA
Make Planning a Fundraiser Easy and Fun

Fundraising Materials:

- Toolkits, Manuals and Sample Booklet (Walk-n-Roll, Golf Tournament, Dinner/Gala & Bowl-a-Thon)
- Promotional Tips
- Banners and Yard Signs
- Families of SMA “At a Glance” Flyer
- Purple & Orange FSMA Bracelets
- Plastic Event Bags
- Donation Cards
- Purple & Orange Golf Tees
- Temporary Tattoos
- Coin Canisters
- Families of SMA Pens
- Angel Tags
- Start Flags
- Pop-up Signs
- Tablecloths
- And more!

Merchandise

- Families of SMA T-Shirt | $12
  Youth Sizes: S M L
  Adult Sizes: S M L XL
- FSMA Bracelet | $2
- Golf Umbrella | $20
- Purple Grocery Bag | $4
- Orange and Purple Ribbon Lapel Pin | $5
- Cinch Bag | $10

To view a complete list of Families of SMA Merchandise visit www.fsma.org/Fundraising/Merchandise
New Event Website Upgrades

Quick and Easy Way to Invite Donors
Families of SMA recently added a feature to our event websites which allows you to upload e-mail contacts directly from your address book (Gmail, Outlook, Yahoo!, AOL and a number of other programs are supported). With this new feature, it takes just a few minutes to share your story with all of your friends and family when fundraising!

Easy Search Function to Find a Fundraiser
An easy drop-down list is now available as a search method to find teams or participants registered for an event.

Use Social Media to Share Your Page
On your personal fundraising page, you can click on the sharing buttons to post a link to your page on Facebook, Twitter and other social media sites. This is a quick and effective way of asking your friends and family to support your efforts to raise money to help find a treatment and cure for SMA!

Other Website Features
• Gifts made to an event or personal fundraising page can be made in honor or memory of a loved one.
• Donors can mark their contributions as “Anonymous” and remain anonymous on all event pages and personal fundraising pages.
• Participants can post their offline cash and check donations to appear online in their personal, team and event totals to showcase their fundraising success before event day!
ALASKA

The Alaska Chapter has been busy getting organized and spreading SMA Awareness. We have gained several new members and have contacted the medical community in hopes of generating more awareness. If you are interested in joining the chapter or have any get-together or fundraising ideas, please e-mail us at Alaska@fsma.org.

ARIZONA

KDK SMASH Golf Tournament

The 17th Annual KDK SMASH SMA Golf Classic held on May 3rd, 2013 in Gilbert, Arizona was another big success. Thank you to all of our Arizona Chapter Families of Spinal Muscular Atrophy supporters, golfers, hole sponsors, and volunteers of helping make this years event so advantageous. We raised over $22,500!

Thank you to our sponsors Brent and Pryl Erekson of Cochise Contractors and Rod Davis of Premier Building Group.

With the help and support of our Arizona Chapter, FSMAM, family and friends, we raised close to $25,000 this year. The AZ Chapter has been hosting this fundraising event since 1997. Over the course of time, we have raised over $600,000 to help support research and medical needs for Families of Spinal Muscular Atrophy.

Special thank you to Kasey, Steve, and Karey Kaler who work tirelessly to make this and all 17 years hosting this event possible. Your love and dedication to this event shows through in all that you do each year.

Diamondbacks – SMA Awareness Night

On April 13th, 2013, 60 chapter members and supporters attended the Arizona Diamondbacks vs. Los Angeles Dodgers at SMA Awareness Night in Phoenix, AZ – and raised $236 in doing so!

ASU Softball game – Cure SMA Awareness

On February 22nd, 2013, Kasey Kaler, SMA Type III, ASU student at the Walter Cronkite School of Journalism, coordinated Arizona State University Softball’s inaugural Cure SMA game. The Sun Devil team and staff wore bracelets reading “Cure SMA,” and the team also wore red ribbon in their hair to honor those living with SMA. Several families braved the Phoenix, AZ winter temps, enjoyed the game and helped educate the public about SMA. The Sun Devil Softball family proudly represented the Families of SMA community in a winning effort, beating Portland State University 6-5.

Field Trips

Several SMA families toured Southwest Human Development’s ADAPT Shop on March 26th, 2013. Our community is fortunate to have this resource, which allows for customized solutions to promote independence and success for children with special needs including made-to-measure foam seating that allows a child to sit upright for the first time, toy modifications that allow a child to play, and communication devices that allow children who cannot speak to express their needs.

Families also learned about mobility information and services in April at the “Permobil Power Trip” at the Disability Empowerment Center.
North Carolina

The 6th Annual LittleMan Memorial Golf Tournament
On February 1st, 2006, we welcomed a bouncing 8lb 4 oz baby boy named Joseph Blaine into our family. We were on top of the world. Little did we know that just eight short weeks later our happy world would come crashing down around us. When Joseph was just about two months old we noticed that something did not seem quite right. We were referred to one of the leading children's hospitals in the nation, Duke University, to have a neurological consult. Before we could even make that appointment we found ourselves sitting in the emergency department because Joseph had developed pneumonia. In less than 24 hours we had the devastating news that Joseph had SMA Type I. Even though we found ourselves with a lot of questions, we were fortunate to have a wonderful resource in the staff at Duke that helped us to learn about SMA, and we were welcomed into Families of SMA with open arms. The care package and information that came in the days after Joseph’s diagnosis helped us to better understand SMA as a disease and how to cope with having a sick child. For the next three months we were able to have few hospital visits and were able to keep Joseph at home as much as we could. Our little boy gained his wings on July 1, 2006 at the age of 5 months.

The 6th Annual LittleMan Memorial Weekend was held June 21st-22nd, 2013 in Macon, North Carolina. With 375 expected for dinner and the tournament having a confirmed 28 teams totaling 112 golfers we were excited, but nervous with the extra large crowd. The banquet kicked off that evening with an excited crowd that once again contained Dr. Jeremy Baker from Frisco, Texas. When the night was over we had found that the live auction had set the stage for another stellar year selling itself for nearly $20,000 with one item bringing in as much as 1/3 of the first year’s entire auction. Steve Owens and the Summertime Band finished the night off and set the stage for Saturday’s tournament. After Saturday’s golf tournament was over we once again crowned a first time winning team which we have done every year since we started. A new milestone was reached with the total money raised coming in at $40,000.

I found myself very humbled this year, much more so than years past, and I’m not sure exactly what it was. As the hour neared for the doors to open at the Amory I found myself sitting on the front steps with tears rolling down my cheeks. I sat and thought to myself, who would have ever figured seven years ago one little boy who had received his wings would be sitting in heaven smiling down on such a tight knit group of people? Thank you from the bottom of our hearts for being a part of our team.

Sincerely,
Blaine and Joanne Reese
Macon, NC

5th Annual Gray’s Gang Walk-n-Roll
This year’s Gray’s Gang Walk-n-Roll included Beat headphones, an iPad Mini, a Kindle Fire, and other fabulous prizes and of course the 2-mile Walk-n-Roll led by Gray and all his many friends and supporters all of whom wore “Gray’s Gang” t-shirts. Gray almost missed his event but made a miraculous recovery after spending three nights in the hospital with pneumonia followed by a staph infection just days before! He was determined to recover and not miss his event. He rallied and made it, and he and his “gang” had an absolute blast!

And to top it off, this year’s event raised over $36,000 for Families of SMA!

In its second year, the Loving Logan SMA Walk-n-Roll held on March 23rd, 2013 raised just over $5,700. This yearly one mile public walk at Smithfield Community Center in Smithfield, NC aims to raise awareness for SMA. This year additional activities were added for both children and adults to enjoy, including a generously donated bounce house. “Our goal is to grow the event into a day-long celebration,” states Dana Grimstead event organizer and aunt to Logan Moore, SMA Type I.

The event drew approximately 75 participants and active volunteers to the park early Saturday morning. Not only were families affected by SMA encouraged to participate,

Michael and Abbie Dougherty
Charlotte, NC

Stroller Warriors: Stroller Warriors H2H and C25K Graduation for SMA
Stroller Warriors Running Club and Atlantic Marine Corps Communities hosted and participated in a 5K/Half Marathon on April 13th, 2013 in Camp Lejeune, NC in honor of Cole Geraghty, SMA Type III. Participants took part in the Couch to 5K program and raised almost $7,300 for Families of SMA! Thank you to Stephanie Geraghty and the Stroller Warriors for organizing this event.

Loving Logan SMA Walk-n-Roll

Charlotte, NC
held on April 20th, 2013 was the best ever of the past five annual events hosted at AG Middle School! The event was hosted in honor of Gray Dougherty, SMA Type II. Gray was born with SMA in 1998 and every year since, a Gray’s Gang/SM A fundraiser has taken place.

About 400 folks came out on a glorious Saturday afternoon. The weather was sunny and cool, and the events were even cooler! There was a bounce house, a photo booth, a cake walk, a punt pass and kick event, the world’s best rockin’ DJ, awesome raffles including Beat headphones, an iPad Mini, a Kindle Fire, and other fabulous prizes and of course the 2-mile Walk-n-Roll led by Gray and all his many friends and supporters all of whom wore “Gray’s Gang” t-shirts.

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The event drew approximately 75 participants and active volunteers to the park early Saturday morning. Not only were families affected by SMA encouraged to participate,
but also community members and activists interested in raising awareness. One group tooted signs for their nephew, Connor, recently diagnosed with SMA. Other participants donated their time, including five volunteers from Kohl’s Cares for Kids.

This event is a celebration of Logan Turner Moore, who passed away from SMA Type I February 2012. “It is a celebration of Logan’s life of course, but it’s also about much more. It is about educating others. It is about awareness. It is about finding a cure, but most of all it is about hope.”

Like us on facebook.com/lovinglogansma for more information!

Dana Grimstead
Kill Devil Hills, NC

5th Annual JPL Golf Tournament

The 5th Annual JPL Golf Tournament was held on April 27th, 2013 at The River Golf and Country Club in Louisburg, NC. This year’s tournament, held in memory of Jocelyn Paige Lee, SMA Type I, was an amazing success in so many ways. With a smaller than normal golfer turn-out we were still able to have one of our largest amounts raised! With the help of our sponsors, friends, family and neighbors, the Jocelyn Paige Lee Foundation was able to raise $3,875 for Families of SMA! We can’t say thank you enough to all those that came out and donated to our cause. Jocelyn’s legacy continues to grow and inspire us all to be better people and help these children. We would like to thank the following companies/organizations for their support: Purdue Pharmaceuticals, Dixon Golf, Quick Print, Air Flow Design, American Legion Post No. 184 and Rock Spring Baptist Church.

Shane and Jennifer Lee
Louisburg, NC

South Carolina

1st Annual Rex’s Ride
Motorcycle Run

On October 8th, 2012 Kevin and Linda Arnold hosted the 16th Annual Arnold Family Golf Outing in Charleston, West Virginia in honor of their son, Eric Arnold, SMA Type II. Thank you to the Arnold Family for donating $7,000 to Families of SMA through your family event!
2nd Annual Cubby’s Run for FSMA

On May 4th, 2013 the 2nd Annual Cubby’s Run for Families of SMA was held in Ridgefield, Connecticut. The event was hosted by Ethan Hynes in honor of his best friend, Cubby Wax, SMA Type I. Ethan and Cubby met playing on the same baseball team at six years old and have been friends ever since. This year the event raised $25,000 for Families of SMA! Thank you to Ethan for all of his hard work and dedication in planning this event!

Saint Mary School Dress Down Day

On April 12th, 2013 Saint Mary School in Ridgefield, CT hosted a Dress Down Day for Families of SMA, organized by the student government. All students K-8 who dressed down had to bring in $1 to support the cause. From the dress down day, we raised $170 to support further research in helping to find a cure. We hope our donation assists you in all the hard work you do for the Families of SMA.

Sincerely,
The Saint Mary’s School government
Ridgefield, CT

Litchfield High School Dress Down Day

On May 3rd, 2013 Litchfield High School in Torrington, CT held a dress down day to support Families of SMA. The event held in honor of Eva Foell, SMA Type I, raised $133! Thank you to Lynn Scozzafava for organizing this event.

3rd Annual SMA 5K - Eat ‘n Run

The 3rd Annual SMA 5K – Eat n’ Run in Columbus, Georgia was held on March 16th, 2013 at Hardaway High School. The event was held in honor of Caleb Merriken, SMA Type II, by his parents, Kanaan and Kari Merriken. Thanks to all of the hard work of the Merriken family and many other family members, friends and volunteers, the event raised over $42,700 for Families of SMA!

Wyatt’s Wish 5K and Fun Run

Wyatt’s Wish 5K and Fun Run was bigger than we had ever imagined. We are hoping to make it an annual event. None of us had ever come close to putting something of this magnitude together, let alone in six weeks. I am still in awe of how the community came together to help in any way possible. They say it takes a village and it couldn’t be more true in this case. Many businesses and people don’t have a lot of disposable income, especially so close to the holidays, but so many gave what they could and it was amazing! It just proves that when a lot of people contribute a little, it can amount to big rewards! We could not have done it without the entire village including the organizers, volunteers, sponsors, donations, and runners. At the event on January 5th, 2013, in Bonaire, GA, we were able to raise $6,000 for Families of SMA!

I truly feel honored to have been able to be a part of such a wonderful event for such a meaningful cause. I had never heard of SMA until baby Wyatt Holter was diagnosed with SMA Type I. After educating myself about SMA I do believe it is a genetic disorder that more people need to be made aware of and it’s far more common than anyone would expect. Everyone at Families of SMA is doing a tremendous job at raising awareness and helping families with their individual special needs.

Treva Hennen
Bonaire, GA

Motorcycle Dice Ride in Honor of Wyatt Holter

The motorcycle ministry at our church, Harvest UMC in Warner Robins, GA held a motorcycle dice ride in honor of Wyatt Holter, SMA Type I. We donated $150 of the money raised to Families of SMA!

Tammie Litsas
Byron, GA
Greetings from the Greater Florida Chapter!

Stretching from south of Sarasota to the North Florida panhandle, our very active chapter covers a large and diverse geographic region and includes many amazing families. Make sure to stay connected by regularly checking our website at www.fisma.org/greaterflorida and joining our “Families of SMA – Greater Florida Chapter” group on Facebook (www.facebook.com/groups/79658598961). We also send a bi-monthly e-mail newsletter to our chapter members. Not sure if you are on the e-mail list? Send a message to greaterfl@fisma.org with your name and contact information today!

Board Update

This year, a few of our Greater Florida Chapter officers have stepped down from their roles on our executive board. They will continue to stay active in local chapter events, however, it is with sad hearts that we say good-bye to Tanya Krajewski, Shawn Santos and Laurie Sore. Thank you Tanya, Shawn, and Laurie for all of your hard work in helping to build the Greater Florida Chapter and in spreading SMA awareness! Your hard work and support means so much to our local families!

Also, Lisa Hoang Shockley was recently appointed as our Chapter Secretary. Join us in welcoming Lisa in her new role on the executive board! Katie Kerns continues to serve as your Chapter President and Audra Butler as Vice President.

If you have any questions for our executive board, please contact Katie Kerns, Chapter President, at (727) 388-1888 (office), (727) 512-4192 (cell), or greaterfl@fisma.org (e-mail).

Greater Florida Chapter Care Packages Groupon Campaign

The Greater Florida Chapter is excited to announce that our most recent event, a partnership with Groupon for an on-line campaign, raised an outstanding $2,690! This campaign ran on the Groupon Grassroots page from March 7th-13th, 2013, and the money raised will go to the Families of SMA Type II and III Care Package Program.

Type I Care Packages are generously funded by the Jacob Isaac Rappoport Foundation and Adi & Shaina Rappoport in memory of their son Jacob, SMA Type I. The Foundation has been generously funding the Type I Care Package Program since 2009.

Families of SMA sends hundreds of care packages each year so that every child diagnosed with SMA in the United States can receive one. Each care package is specific to the type of SMA that the child has been diagnosed with. These care packages include a range of items such as informational materials, toys appropriate for individuals with each type of SMA, books, blankets, hand-made quilts, and DVD’s. Many ideas for the items in these Care Packages come from SMA parents.

Thank you for supporting our chapter’s Groupon Campaign and for allowing funding to go towards this amazing program!

iParty Charity Days

iParty of St. Petersburg invited the Greater Florida Chapter to participate in its Charity Days event this spring. The store offered to give our organization FREE party goods to use for fundraisers and chapter events. We were able to fill up two shopping carts full of merchandise from their clearance section! Thank you to iParty for your generous donation!

iGive

Did you know that you can help support our chapter every time you shop online? With the iGive Button, you can do just that. When you join iGive, you can shop at your favorite stores and a percentage of your purchase will automatically be donated to our organization! This is a simple way to support our cause at no additional cost to you. Go online to www.igive.com/FamiliesofSMA-GreaterFloridaChapter to join today!

NYC Half Marathon

The NYC Half Marathon held March 17th, 2013 in New York, New York raised over $51,000 for Families of SMA! Ten people ran the race and raised funds to help fund a treatment and cure for SMA! Thank you to Lauren Bonelli, Claudine Campanelli, Steve Cannady, Douglas Erwin, Alex Kostyuchenko, Doris Linke, Danielle Nelson, Anthony Trama and Dmitry Vaysman for all of your hard work and dedication! Special thanks to the Erwin family for all of their help with this event!
New York Gala of Hope

The inaugural New York Gala of Hope modeled after the same event held annually in South Florida was held on June 6th, 2013 at the Ferrari-Maserati Dealership of Long Island in Plainview, NY. The event featured a silent auction, salsa style entertainment and an elite array of Ferraris and Maseratis on display. Guests also had the opportunity to meet with some local New York icons including Plaxico Burress, Jay Cardiello and Liza Huber.

Thank you to Michele Erwin and Julie Meriam for all of their hard work in planning this successful event that raised over $48,000!

A special thanks goes to Jennifer Miller Smith and Fiorenna Fuentes who organize the Gala of Hope in South Florida for all of their help!

2012 NYC Triathlon

In January 2010, soon after meeting my six month old niece for the first time (I live in Australia), I received news from my family that she, Amanda Shea, was diagnosed with SMA Type I. The realization that my niece will never reach the milestones that other children reach and would remain mostly immobile for the rest of her life made me appreciate the ability within my own body. I signed up for my first short distance triathlon immediately after her diagnosis. Every day that I trained, even to this current day, I think about Amanda and other SMA children.

After completing that first triathlon (a massive feat considering how I panicked in the open water and swam in the wrong direction!), I ramped up my training to include half-marathons and more triathlons. By 2011, as Amanda continued to grow, I started eyeing the NYC Triathlon as a goal. When it was announced that the 2012 triathlon would be held a week after her 3rd birthday, I signed up thinking the timing was perfect. I could return to NYC to celebrate her birthday with the family – something I had desperately wanted to do since she was born – but to also start a fundraiser in honor of my niece who had inspired me to do triathlons. Even though she never will be able to – I could, for her.

On July 1st, 2012 we celebrated Amanda’s 3rd birthday together as a family. By July 8th, on the morning of the NYC Triathlon, we raised over $5,300 for Families of SMA – over twice the amount that we originally expected to raise. It was one of the proudest moments of my life. Participating in the NYC Triathlon was one of the best moments in my life – not only for the physical challenge, surrounded by my family, but because of the funds and awareness we raised for SMA. It is something to feel truly proud of.

Sandy Lam
Australia

CA Montessori Children’s Center Dance Class

On April 30th, 2013 the CA Montessori Children’s Center in Oslandia, NY hosted a dance class to their families and donated the proceeds to Families of SMA. The event raised $2,550! Thank you to Maureen Ligouri for once again organizing this event!

Farm Family Insurance Jeans Day

Farm Family insurance companies, located in Glenmont, NY, recently raised $1,091 for Families of SMA in memory of Josie Lucy, SMA Type I. The company held a Jeans Day on June 17th, 2013, where employees who made a contribution to Families of SMA could wear jeans to work on a specific day.

According to Katie Piretti, a Farm Family employee and organizer of the event, more than 200 employees participated. “My cousin, Josie Lucy, is a beautiful SMA Angel. Families of SMA was there for Josie and her family as a resource for support, information and services. It was wonderful to have the opportunity to give back and support Families of SMA and to spread the word about this great organization.”

Katie Piretti
Lenox, MA

Alex and Ani Open House

Our Alex and Ani Open House event held on May 18th, 2013 was a great success! We had about 30 people come out to the Alex and Ani store and we raised $675 for Families of SMA.

Michele Rubenstein
Yorktown Heights, NY

Sean & Devin’s Birthday Gift Registry

Thank you to Eileen Prymaczek for collecting donations in lieu of gifts for your sons’, Sean and Devin, birthdays in April 2013. $135 was raised for Families of SMA!

New York Gala of Hope (cont.)
3rd Annual Tee Off with the Drive to Cure SMA in Honor of Ryan

Jeanne Emerson organized the event along with the Manfre and Fedea families in honor of their grandson, Ryan Manfre, who has SMA Type III. This year the event raised just over $50,000!

Thank you to Ken, Jeanne and all of the volunteers that helped make this year’s event such a success!

7th Annual Illinois Chapter Walk-n-Roll

The 7th Annual Illinois Chapter Walk-n-Roll was a great success! Hundreds of people gathered at Independence Grove Forest Preserve in Libertyville, IL on June 8th, 2013 for a day of family fun! Everyone enjoyed the DJ, face painter, balloons and magician. Thanks to the hard work of Chapter President, Janet Schoenborn, and many volunteers, the event raised $39,000 for Families of SMA!

Ross’ Holiday Beard for SMA

In February 2012, family friends of ours were told that their daughter, McKenzie Rainn, was diagnosed with SMA Type I. On April 7th, 2012 at the very young age of 4 months and 29 days, McKenzie sadly passed away in her parents arms due to complications from SMA.

Families of SMA is my charity of choice because of the amazing support they provided McKenzie and her family during her diagnosis. I cannot imagine receiving a more fulfilling gift than one that provides funding and resources to Families of SMA. Each person who donated had their name entered into a drawing. The winner created a beard design that I wore for a week.

Thanks to the support of family and friends $1,030 was raised!

Ross Herrick
Chicago, IL

The 3rd Annual Tee Off with the Drive to Cure SMA in Honor of Ryan took place on May 18th, 2013 in Lemont, Illinois. Ken and
Cure for Chloe Christmas Ornament Sales

Our daughter, Chloe, SMA Type III, designed a beautiful ornament again this winter for our annual Cure for Chloe Christmas Ornament fundraiser! We are excited that she raised $597 this year to give to Families of SMA for SMA research! Thank you FSMA for giving our family HOPE!

The Ochoa Family
Homewood, IL

Riverman Travel Goals Fundraiser

Thank you to the staff of Alexander Travel, Ltd. of Peoria, IL for raising $325! The staff made Families of SMA their charity of choice. For every goal that was hit by a staff member, $25 was donated.

Lee County Health Department Jeans Day Fundraiser

On February 15th, 2013 the Lee County Health Department in Dixon, IL held a jeans day to benefit Families of SMA in honor of Lisa Woods. Thanks to all of the department's employees and Kathy Ferguson $104 was raised!

IOWA

2nd Annual FSMA Spin 4 Quinn

On National Star Wars Day, May 4th, 2013, the 2nd Annual Spin 4 Quinn Bike Ride/Walk took place at Guthridge Park in Hiawatha, Iowa. After a week of pouring rain and 30 degree temperatures, the skies parted long enough to set up and ride the 20 mile route through Cedar Rapids to Tait Cummins Park. Though many were scared away by the looming clouds, 100 hardy souls showed up to ride and walk in honor of Quinn Anton Jensen, SMA Type II. Band members of PTO from Mt. Vernon United Methodist Church brought warmth to the morning with their music.

Quinn biked this year with his new hand-pedal tricycle attached to his dad’s tricycle. They had a fun, mud-splashed ride through downtown with friends! There were many activities taking place under the pavilion. Lizzy’s Creations returned this year with their amazing face paintings! Also present were Star Wars characters from the Iowa City 501st Legion to take pictures with event guests and participants. Quinn was even made Honorary Storm Trooper!

Thanks to all of our participants, donors and supporters, we raised about $8,500 for Families of SMA!

Nancy, Brice, Grant, Drew and Quinn Anton Jensen
Hiawatha, IA

Alan Shepard Elementary School Pink and Sparkles Day

On April 19th, 2013 the Alan Shepard Elementary School in Eldridge, IA held a Pink and Sparkles Day fundraiser in honor of Ella Christopher, SMA Type II-III, and Jack Lindaman. Thanks to the generosity of students and staff the fundraiser raised over $3,500 for Families of SMA!
**Chapter Updates**

**Kansas City**

**Board Update**

After outstanding service for more than a decade, the Kansas City Chapter is saying good-bye to two of its leaders, Natalie and Tim Gibbs. In June of 2013, Natalie and Tim stepped down from their posts as leaders in the Kansas City Chapter as well as the Chairs for the Kansas City Chapter Race-n-Roll. Vice President Kristal Wilson will be stepping into the role of President. Kim Sykora is stepping into the role of Vice President. The Chapter will hold elections in the near future to fill all vacant posts. Kristal Wilson, Kim Sykora and Tracy and Walt Cochran have taken the reins and will plan the 13th Annual Kansas City Chapter Race-n-Roll.

The Chapter would like to extend a heartfelt thank you to Natalie and Tim for their years of service, leadership and support. Their efforts to raise awareness and funds for SMA are unparalleled and their service to SMA families is inspirational.

If you have any questions or are interested in learning more about becoming a member of the Kansas City Chapter of FSM A Board, please contact Kristal Wilson, Chapter President, at (816) 529-1834 or kcfsm@ymail.com.

**Missouri**

**Gracie’s Second Annual Steak Dinner for SMA**

After the success of last year’s event, Gracie’s Annual Steak Dinner returned to Eugene, Missouri this year on April 27th, 2013. All event participants had a great time. In addition to the steak dinner, a silent auction and raffle were held. There was a clown in attendance who spent the night painting faces and creating balloon creatures for the children to enjoy.

Since many who came out to the event were unaware of what living with SMA means, a video presentation played throughout dinner in order to provide guests with some insight and hopefully inspire them to continue to work to fundraise for such an amazing cause.

In total, the dinner, silent auction and raffle helped to raise $8,787 for Families of SMA in honor of Gracie DeGraffenreid, SMA Type I. Thank you to all participants for your hard work and dedication to the cause.

Sherry DeGraffenreid
Eugene, MO

**Team Tilly Fundraiser**

Family and friends of Matilda "Tilly" McRoberts, SMA Type II, formed a team for the 2013 St. Louis Marathon on April 16th, 2013 in St. Louis, MO. Thank you to all of the participants for raising over $7,100 for Families of SMA!

**Phi Tau Omega Sorority Trivia Night (Delta Phi Chapter)**

The ladies of the Delta Phi Chapter of Phi Tau Omega sorority of St. Louis, MO raised $2,400 for Families of SMA at our annual trivia night! We hope this will help with the great work that you do for all the children with SMA. We have a friend, Brittany Carpenter, with SMA Type II who is always at our trivia events.

Sincerely,

Kathy Gradyear
Delta Phi Chapter
Arkansas

Families of SMA Receives an Amazing Gift from The Miller McNeil Woodruff Foundation

Families of SMA is thrilled to have received an incredible donation from The Miller McNeil Woodruff Foundation in the amount of $87,000! This exceedingly generous donation will go to support the FSMA funded Gene Therapy Program at Nationwide Children’s Hospital in Ohio.

This substantial gift will not only help continue the promising work on this research program, but will also serve as a reminder and beautiful memorial of a young life taken too early by SMA. Miller McNeil Woodruff lived for only 87 short days. This meaningful and significant gift of $87,000 symbolizes $1,000 for every day that Miller lived. It will ensure Miller’s precious legacy lives on.

On June 23, 2011, Miller McNeil Woodruff passed away from SMA Type 1 at just 87 days old. On that day, Miller’s parents, Patrick and Meredith Woodruff made it their mission to spread awareness, fund critical research and provide support to families faced with the challenges of living with SMA. Through the work of their foundation, the Woodruff family has supported organizations and research throughout the country for more than two years.

“Patrick and I promised each other AND Miller that we would make the best of this unfortunate situation and NEVER let Miller be forgotten. We spent 87 precious days with our angel, until his poor little body finally let go. We are at peace knowing he is breathing easy in Heaven and we have faith that we will see him again! As part of our commitment, we have started The Miller McNeil Woodruff Foundation. Our goal is to raise awareness, fund research and offer support to other families who are faced with this challenge.”

Patrick and Meredith Woodruff
Bentonville, AR

Everyone at Families of SMA would like to thank The Miller McNeil Woodruff Foundation for choosing to help us continue our mission to support cutting edge research to develop a treatment and cure for SMA. On behalf of the many families who will benefit from your generosity and Miller’s legacy, thank you!

To learn more about the Miller McNeill Foundation please visit www.imwithmiller.com.
Briley FAITH’S Walk & Run for a Cure

Briley FAITH’s Walk & Run for a Cure was held in Alexander, AR on June 1st, 2013. The event was held in memory of Briley Faith Turner who had SMA Type I. The event included lots of fun games and a balloon release. Thanks to the generous support of family and friends the event raised almost $6,500! Thank you to Diane Gardner for organizing the event.

MICHIGAN

Board Update

Ken and Cindy Armbrustmacher stepped down as Chapter leaders in 2012. Thank you to Ken and Cindy for all of your years of hard work building and growing the Michigan Chapter! The Michigan Chapter is excited to announce its new officers! Kevin Gorman of Orion, Michigan is the new Chapter President. Kevin was involved with the Michigan Chapter when it was first created back in 2004. After stepping away for awhile he is looking forward to being involved in the organization again and bridging local families together. Holly Schafer of Dewitt, MI will continue to be Chapter Vice President and Michelle Fox of Ada, MI will continue to be Chapter Treasurer.

Marianne Hunderman is also the new Chapter Family Support Coordinator. Marianne has a 5 year old daughter with SMA Type II. She is looking forward to creating a strong support network for Michigan families.

9th Annual Michigan Chapter Run, Walk-n-Roll & Family Fun Day

The 9th Annual Michigan Chapter Run, Walk-n-Roll & Family Fun Day held on May 4th, 2013 in Lansing, MI was a huge success! The event brought together families and friends for a day filled with great music, fun and clowns. Thanks to the hard work of Holly Schafer and the Michigan Chapter, the event raised almost $50,000 for Families of SMA!

2nd Annual Bowl with Brayden 4 SMA

The 2nd Annual Bowl with Brayden 4 SMA was held on March 17th, 2013 in Muskegon, MI. The bowling tournament, held in honor of Brayden Janetzke, SMA Type II, by his mother, Christina, raised over $2,500 for Families of SMA!

9th Annual Michigan Chapter Hot Dog Fundraiser

The 9th Annual Michigan Chapter Hot Dog Fundraiser held May 4th – 5th, 2013 raised over $1,000 for Families of SMA in honor of Malorie Fox, SMA Type II! The event, hosted by Michelle Fox and friends, was held in Ada, MI.
Minnesota

William Anco Memorial 5K (W.A.M. 5K)
The William Anco Memorial 5K held on May 4th, 2013 in Lindstrom, Minnesota was a huge success. The event, organized by Scott Buchkoski, raised over $3,200 for Families of SMA!

North Dakota

Jack Attack on SMA: Round 9
On June 9th, 2013 Kristi and Rod Gellner hosted Jack Attack on SMA: Round 9 in honor of their son, Jack, SMA Type II, in Fargo, North Dakota. Thanks to the generous support of family and friends the event raised over $15,000 for Families of SMA!

New England

2013 Rogo Cup SMA Golf Tournament
The 2013 Rogo Cup SMA Golf Tournament was held June 3rd, 2013 at the Atlantic Country Club in Plymouth, MA. The Rogowicz, Speigel, Butler and Scully families joined together, along with many family members and friends, to raise money for Families of SMA. The tournament is held in memory of Mark Butler, SMA Type I, in honor of Billy Speigel, SMA Type II, and Lauren Rogowicz, SMA Type III. Although the weather wasn’t very cooperative, everyone showed up with smiles and umbrellas and had a great time. The families can’t thank the participants and all the donors enough for the great support helping us raise over $38,000 for Families of SMA. With this incredible support we are looking forward to many more years for the Rogo Cup!

Joe Sculley
Marshfield, MA

Families of SMA Yard Sale and House Party
Under sparkling blue skies, we hosted our 9th Annual FSM A Charity Yard Sale. With the community’s amazing spring cleaning donations, our incredible day of help from family and friends, a last minute lemonade/bake sale set-up and run by some special 7th grade lady-friends of William’s, and lots of very generous shoppers, we were able to raise nearly $3,000 for our Walking for William pledge card in honor of William Johnson, SMA Type II, for the 13th Annual New England Chapter Cure SMA Walk-n-Roll that took place on May 18th, 2013. Extra special thanks to the Cook Family, Tim Jones, Hall Family, Robin & Mike Petit, Tommy Dugan, Neal O’Connor, Sarah Murphy, Heidi Oppel, Gramps, Michelle, Debby, and Carol. We could never pull this sale off without the community behind us!

We also held a house party at our home in Medfield, MA to raise funds for our team on May 11th, 2013. Our party raised $2,400!

Heidi Johnson
Medfield, MA

Boston Marathon
My daughter, Joanna Buoniconti, suffers from SMA Type II. On April 15, 2013, I ran the Boston Marathon in her honor. This is the ninth year I have participated in the race. I am so proud to have done this as a tribute to Joanna. I am most grateful for the support of my family and friends and I am most hopeful that their kindness and generosity will help find a much needed cure for SMA. This year I was able to raise $5,230 for Families of SMA.

Stephen J. Buoniconti
Springfield, MA

may differ from current fundraising totals by the time you get this newsletter.
Massachusetts

13th Annual New England Chapter Cure SMA Walk-n-Roll

The 13th Annual New England Chapter Cure SMA Walk-n-Roll took place on May 18th, 2013 at DCR Wompatuck State Park in Hingham, Massachusetts. It drew a record crowd of 900 and raised more than ever before, $140,000. The walk was organized by Silvia Murphy, Stacey Farrell, and Kate Norton, who were grateful for the assistance of many volunteers and for the donations, sponsorships, and raffle items from family, friends, and local businesses.

The weather was perfect, making it possible for over 30 SMA families to attend, including participants from Maine, Vermont, New Hampshire, Massachusetts, Rhode Island and Connecticut. This year the trademark T-shirts had bright orange ink and the initials “KMN” in memory of Kathy Norton, an SMA mom and dedicated FSMA supporter who recently lost her battle with cancer. Kathy helped organize this event for many years; her supporters, including a team of her co-workers from the Boston Globe, turned out in full force to carry on her mission to cure SMA.

The raffle had over forty items and was a huge hit, as usual. Due to the Bruins winning streak the crowd received a visit from their mascot, Blades. The children also enjoyed face-painting, balloons, games and prizes, and a fire truck. The favorite refreshment item was the Hoodsie ice cream cups!

The incredible amount raised was a result of significant fundraising efforts by many different participants. The top fundraising teams this year were: Team Murphy, Team Norton, Mac’s Pack, Team BraeKer, and Walking for William; followed by Team Farrell, Wizards of Oz, Team Mirabile, and Team Nico. Other teams supporting the walk were: Diego Alejandro, Vivienne for a Cure, Team Addison Barrett, Team Pip, Team Melanie Lee, Glen’s Gang, Team Ian, Team Goldman, Angelica, Team Greer, Team Ashdon, Chloe’s Club, Team Brendan, Fight for Ford, Team Sammy, Team O’Neill Boys, Team Kamdyn, Team Kristin Lee, and Boston Children’s Hospital. Other SMA families that participated or were represented included: the Butlers, Carters, Cederlunds, Costas, Cutrones, Hoadleys, Lucy, McDonalds, Nguyens, Pimentels, Reillys, Rogowicsz, and Venezias. Although they couldn’t attend the event, the Kulis family helped with the donation of water and the Rose family sponsored the t-shirts. Several teams used mini-fundraisers instead of asking for pledges, including Team Walking for William -- who held a yard sale, house party, and quilt raffle. Other creative contributions included “Bootcamp for a Cure,” lemonade stands, bake sales, and a handmade accessories sales.

We look forward to seeing everyone again in May 2014. Please contact Silvia Murphy at brianandsilvia@verizon.net for questions or more information.
Medfield High School Charity Baseball Game

On March 12th, 2013 the Medfield High School baseball team from Medfield, MA held a charity game to benefit Team Walking for William for the 13th New England Walk-n-Roll in honor of William Johnson. Thanks to Matt Pembroke the game raised $2,840!

SMA Day at the Rockhoppers

On January 12th, 2013 the new North American Lacrosse League Boston Rockhoppers hosted their first home game of the season against the Baltimore Bombers at the New England Sports Center in Marlborough, MA. They may have suffered a tough overtime loss to the Bombers, but the game was a win for SMA!

The team’s Co-President, Tyler Low, was contacted by the Johnson family last summer to consider making a ticket donation to the family’s annual Fall Classic FSMA Charity Golf Tournament in honor of their son William Johnson, SMA Type II.

Not only did Mr. Low put together donations of two exciting spectator packages for the home opener, one including a “Captain for the Day” on-field experience for a lucky youth, he also offered something extra special to William and the Johnsons. During the home opener, they were able to set up a 50/50 raffle table where proceeds would benefit Families of SMA. William, along with his cousin Julian and other family members went around selling tickets in the crowd. Who could say no to William?

The wonderful experience not only raised $215 for the New England Chapter of Families of SMA, but also raised SMA awareness in the huge home crowd. The Rockhoppers’ Organization should be applauded for their willingness to support the effort of a local charity. Again, a big win for the home team despite the end scoreboard.

Heidi Johnson
Medfield, MA

Aaron’s Animal Angels – SMA Strides for Life

On April 21st, 2013, Aaron Phelps, SMA Type I, held a walk in Fairfield, CA, and was joined in a virtual walk by his Aaron’s Animal Angels friends from across the country. Aaron asked everyone to grab their sneakers and pet(s) to walk with him and his therapy dog, Lindy, to celebrate his 5-year D-Day anniversary, and to raise funds to support Families of SMA. With everyone’s support and enthusiasm Aaron raised $10,023 for research and awareness of SMA. Walkers joined in from 26 states, and even as far away as England! A wonderful morning, afternoon and evening was had by all. Special thanks to the San Pasqual Band of Mission Indians and Platinum Performance, Inc. for their outstanding support of this event. Aaron and Lindy are looking forward to walking with everyone again soon!

Santa Cruz Marathon in Memory of Fletcher Krider Koll

I have been training to run a marathon for the last 15 years. Each year there was a reason to not follow through. Mostly work related, the odd injury and then just good old-fashion laziness.

This all changed when two of the people I work with at Goodby Silverstein & Partners learned that their son, Fletcher, had been diagnosed with SMA Type I. I didn’t know much about SMA. I didn’t know that one in every 6,000 babies would be diagnosed with SMA this year. I didn’t know the disease destroys the nerves that control voluntary movement. I didn’t know that children with SMA may never crawl, walk or even lift their head. All I saw was a little smiling baby boy called Fletcher.

This was the motivational push that I needed. I was going to run the 26.2 miles and raise some cash in the hope that it would make Fletcher’s life a little easier and would help others with the condition.

On April 12th, 2013 little Fletcher beat me to the finish line as he lost his battle with SMA and passed away. Both of his parents, Sara Krider and PJ Koll were with him, saying that it was peaceful and painless and love was all around.

I finished the Marathon in Santa Cruz, CA on May 19th, 2013 and was propelled by his spirit. Overall, I raised $7,765 for SMA research and awareness. Thank you to everyone that supported me.

Derek Robson
Tiburon, CA

4th Annual Yard Sale

The Gutierrez family held their 4th Annual Yard Sale on June 22nd, 2013 in honor of Jessica and Jaylin Gutierrez-Gayle (our lovely angels). The yard sale assists in raising awareness and funds to help find a cure for SMA. Our local TV station and Anchor Alan Sanchez aired a segment and it helped us bring more customers in and spread the word. We raised close to $800 for our Northern California Walk-n-Roll team. We will continue to fundraise for SMA! Thank you to our friends, family and community for your donations and your support!

The Gutierrez Family
Shafter, CA
Concert for a Cure

The 13th Annual Concert for a Cure was held on May 11th, 2013 at the Blackhawk Country Club in Danville, California and was a huge success! This year’s event raised over $142,000 with a fun filled theme of “Cruising for a Cure.” This memorable evening was made especially rewarding for donors when Kenneth Hobby, President of Families of SMA highlighted the exciting advancements in SMA research taking place now in California. Dr. John Day, Stanford Professor of Neurology, Pediatrics, and Pathology, was our special guest of honor. Dr. Day had just that week performed the first infant clinical trial for ISIS at Stanford and was pleased to share his experience. Additionally, clinical researchers Adam Kennedy and Darren Hwee from Cytokinetics were in attendance. Cytokinetics was a first time sponsor and is working with FSMA on a new clinical trial of Tirasemtiv in SMA mice.

Guests were deeply moved by the show of support by Danny McHale’s classmates from Los Cerros Middle School who presented the McHale and Dindzans families with a check for $1,838. Led by Hannah Doris and Brady Martin, these friends organized lemonade stands and school bake sales so they could make their own contribution as a surprise to the McHale family. The 20 seventh graders and their siblings were a big hit as this marked their fourth year serving at Concert for a Cure selling raffle tickets, running games and encouraging donors to give generously.

Mary and Joe McHale founded the Concert for a Cure in 2001 shortly after the diagnosis of their son, Danny, with SMA Type II. Nancy and Andris Dindzans joined the efforts in 2003 when their daughter, Ariana, was diagnosed with SMA Type III. Both Danny and Ariana will turn 13 years old this year. Over the years, this event has raised over $1.14 million dollars to fund Families of SMA research programs.

Thank You!

The McHale and Dindzans families send out their heartfelt thank you to our friends, families, and the local community for their generosity in their continuous support of SMA families and children everywhere.
The 15th Flying Pig Marathon
On May 5th, 2013, 25 dedicated runners laced up their shoes to run the 15th Flying Pig Marathon for Families of SMA in Cincinnati, OH. Some ran the full marathon, while others completed the half marathon, 10K, 5K or 4-person relay. Thank you to all of our runners for their hard work and fundraising efforts. The event raised over $5,600! A special thank you to Beth Lockwood for helping organize the event.

Pigs’n Pins Bowling Fundraiser
The Norwood Police Department hosts a fundraiser every year and donates the proceeds to a different charity. This year they chose to fundraise for Families of SMA in memory of Madison Vickers, SMA Type I. The event took place on March 9th, 2013 in Cincinnati, OH, and was a complete success. We are so appreciative that they chose to honor the memory of Madison by donating a whopping $4,700 in her memory to Families of SMA! We could not be happier or more proud to be on their team.

Thank you to everyone who donated baskets to the raffle, purchased raffle tickets, split the pot, paid for lane sponsorship, and to the officers at Norwood Police Department for being who you are and doing what you do!

Michelle Vickers
Cincinnati, OH

Garage Sale Fundraiser
On May 9th, 2013 Greg and Andrea Hill hosted a garage sale in Salon, OH in honor of their nieces, Emma and Ruby Cannady, both of whom have SMA. The event raised $2,400 for Families of SMA!
10th Annual OKI Chapter Walk-n-Roll
A great time was had by all with carnival games, face painting, PB & J sandwiches and amazing raffle prizes at the 10th Annual OKI Chapter Walk-n-Roll in Cincinnati, Ohio on May 5th, 2013.
In the past 10 years, the OKI Chapter has raised close to $700,000 through their annual Walk-n-Roll, which has had an outstanding impact on the entire SMA community. A special thanks to the Lockwood Family, OKI Chapter leaders and board members, Walk-n-Roll committee members and everyone who has helped to organize and build this special event over the past 10 years! Without you and the dedication of all our Ohio, Kentucky and Indiana families, this amazing chapter and event would simply not be possible. Thanks to our generous donors and the hard work of all of our fundraising teams, we raised $80,000 for Families of SMA!

A Special Note From Caitlin Brown’s 10th Annual OKI Chapter Walk-n-Roll Fundraising Page
Everyday in our tech modern world we use all kinds of acronyms such as LOL, LMAO, TTFN, TTYL, U, R, but do you know what SMA stands for? These days we know all about so many diseases such as Breast Cancer, Alzheimer’s, and Parkinson’s disease, which are all worthy causes, but Spinal Muscular Atrophy is hardly ever known unless a person in your family or someone around you has it. Even then, some of those people still do not understand what the disease does to not only the person who has it, but their caregivers as well.
Imagine breathing everyday like it is the hardest thing you’ve ever had to do. It feels like an elephant is sitting on your chest and you feel like if the elephant got up you could finally feel what it is like to breath normally for the very first time in your whole life, but no matter what you do the elephant just won’t get up. Now remember being a child and not being able to go to school and have the socialization with your friends because there are too many germs especially, in the winter time so you have to be home schooled so you don’t get to hang out with all of your friends and make new friends because even if you get just a mild cold it can turn into pneumonia and you can die from it.
That is what I went through as a child, even though eventually in high school I went to school everyday I still have to be cautious who I am around if they are sick. I love being an Aunt to my two nieces and my nephew but when they are sick even if I am already in the same room as them I cannot get too close to them just in case. Also, imagine having so many doctors appointments and medicines that sometimes it feels like that’s all you do. Often the little SMA kids and even the adults do not understand why other little kids and adults stare at them because even though we think we are perfectly normal that is not seen by a lot of other adults and especially kids.
Even if you are born with the most severe type of SMA (type 1) like I have it just keeps getting worse over time. It is a progressive disease so at one time you can write a whole paragraph with a pencil and then maybe six months to a year later you can only write a couple sentences and then a couple years after that you can only write your name and then a few years after that you can’t even write your name. I used to have no problem using my computer and now the last few years particularly the last year I am having more and more trouble using my mouse pad. This means that it can often be discouraging to people with SMA because we are not quite sure what ability we are going to loose next or how bad the severity of this disease can get or how quickly we are going downhill. I was not supposed to live past two years old and yet I am almost 25. Unfortunately, other babies and kids have not been as lucky. We have already lost enough kids to this wretched disease.
Praying for a cure!
Caitlin Brown, Cincinnati, OH
Kentucky

The Schwegmann Wedding
Erica and Nick Schwegmann of Covington, Kentucky gave a $380 donation to Families of SMA in honor of Emma and Nicholas Lockwood, who both have SMA Type I, in lieu of wedding favors on May 4th, 2013. Thank you both for your generosity and support!

Indiana

Home Laureates Homemaker’s Club Holiday Candies and Nuts Sales
The Home Laureates Extension Homemakers Club in Muncie, Indiana sells holiday candies and nuts each year. All proceeds raised are given to non-profit organizations. This year the club voted to donate to Families of SMA in honor and memory of Katie and Betsy Swetnam. This year we were able to raise $100 in their memory!

Jackie Johnston, Treasurer
Muncie, IN

Oregon

Portland Gathering
In January 2013, families in the Portland, Oregon area gathered for a fun afternoon. Several families were able to attend and get to know each other. We would like to continue holding events in Oregon, so please let us know if you are interested in organizing something. Thank you to Kathleen Wilson for planning this event.

Washington

Wreaths of Hope Fundraiser
In November 2012, Sybil and John Kuhn hosted a Wreath’s of Hope Fundraiser in honor of Kiley McClay, SMA Type II, in Centralia, Washington. Thanks to all of their hard work $945 was raised for Families of SMA!

Gymnastics Camp Fundraiser
My name is Paige and I am a senior at Naches Valley High School. For my senior project, I held a gymnastics camp at my place of work in Naches, WA in December 2012 to raise money for your organization. My good friend Peyton Yates is a very special six year old. She was diagnosed with SMA Type III about two years ago. She continues to amaze me with her bravery and strength. The $400 I raised at the event is in honor of her. Thank you for all the support your organization to offers families affected by SMA. I hope this donation will help continue all the amazing work Families of SMA does to find a treatment and a cure.

Paige Engquist
Naches, WA

Mariner’s Game
In Seattle, WA on June 8th, 2013 we held the 5th Annual Mariners Day in memory of Gabby Stack, SMA Type I. Another fun day at the ballpark with friends and family while raising funds and awareness for Families of SMA. Over 75 tickets were sold this year with many new families joining us!

PACIFIC NORTHWEST

Bowling Party
In March 2013, FSMA families got together for an afternoon bowling party. We had lots of fun bowling, playing games and sharing lunch.

may differ from current fundraising totals by the time you get this newsletter.
Families of SMA PA Chapter 10th Annual Cure SMA Walk-n-Roll

The 10th Annual PA Chapter Cure SMA Walk-n-Roll, held May 19th, 2013 at Lloyd Hall Recreation Center in Philadelphia, Pennsylvania was a tremendous success! Over 800 “walkers and rollers” turned out for a beautiful day of family fun. Seventeen teams contributed to the success of this year’s walk with their incredible fundraising efforts. In total, this year’s event raised almost $100,000! The fun-filled day included entertainment provided by fire throwers, adaptive games, and the chance to connect with other SMA families.

Because of the PA Chapter’s commitment to raising funds for our SMA research and family support programs, we are making tremendous strides toward achieving our vision of a world where SMA is treatable and curable. Thanks to the funding of this Walk-n-Roll over the past 10 years, we’re now funding 15 drug development programs within our pipeline, 3 of which are currently in clinical trials. Thank you again to all of our amazing SMA families, participants and donors for helping make this possible.


A grateful shout out goes to our in-kind donors who help to make the day so special: Walter Perez from Channel 6 ABC News, Susan Pardys Photography, MicDuff Photography, Chuck Adams Photography, Staples, and TAG DJs.

Thank you to Karen McRory-Negrin, Allyson Henkel, Tara Maida, and Paula Saxton for all of their hard work in planning this event year after year! Special thanks to former Chapter Treasurers Connie Smith and Josephine Tripodi for playing key roles in this event for eight years.
Muscles for Mckenna Gala

My sister Amy and her husband Jim became the parents of beautiful twin girls 4 years ago. Last year, my niece, Mckenna Ellixson, was diagnosed with SMA Type III. Since they were able to walk, they noticed that Mckenna had difficulty ambulating, fell frequently and developed a slight tremor in her hands. They had many visits with her pediatrician as well as pediatric subspecialty physicians but were never given a definitive diagnosis. She was involved in physical therapy, but it was not until she was evaluated at the neuromuscular clinic at CHOP that the diagnosis of SMA was discovered.

Mckenna is an amazing little girl with great energy and enthusiasm for life. I had to change my defeated attitude and do whatever I could for my precious niece, as I know my sister would do the same. Therefore, with the help of friends and family, I planned a gala in her honor.

We held the first Muscles for Mckenna Gala on March 15th, 2013 in Philadelphia, PA, with nearly 300 attendees, and raised over $96,000! We had an amazing party and are already planning our 2nd gala on March 7th, 2014! Next year’s event will also include food, dancing and live entertainment at the elegant Crystal Tea Room in Center City Philadelphia.

Our family is dedicated and determined to help find a cure for Mckenna and everyone affected with SMA. Thank you to everyone who helped plan the event and all of our generous supporters!

Kellie Keenan
West Chester, PA
Kears Guest Bartender Fundraiser
On March 23rd, 2013 Kristen Kears was a guest bartender at the Overbrook Golf Club in Bryn Mawr, PA. Kristen gave her tips and donations to Families of SMA in memory of Zane Schmid, SMA Type I. Thank you Kristen for raising almost $1,200!

Shoot for a Cure

We would like to thank everyone who has helped make this year’s “Shoot for a Cure” in honor of Tiernan James Conner Park, SMA Type I, a great success. This year we managed to raise $1,025 to help find a cure for SMA at our event in June 2013 in Dubois, PA. We need to thank the following companies and groups for their help: All Season Trap & Skeet, Ruger Firearms, Fat Boy Game Calls, Reynoldsville Riverside, Jim Schaedler, Adam & Susan Hollobaugh, Virginia & Jim Conner, Josette Harbison, Josette Harbison, Stacy Park, Kelly Park, Tom Dinger, Greg & Marcie Dixon, Steve Meholic, Lisa Schaffer, Dave Larko, Larry & Amy Way and James Park.

Sincerely,
Andrew and Erin Connor-Park
Dubois, PA

Thirty-One Party Fundraiser
Laurie Peters held a Thirty-One Party Fundraiser on February 10th, 2013 at St. Luke’s Church in Stroudsburg, PA. Thanks to all of Laurie’s hard work, $510 was raised for Families of SMA!

Lincoln Jeans for a Cause Days
On February 26th, 2013 a jeans day was held to benefit Families of SMA in Exton, PA. Thank you to Pam Cummins for her work in planning this event and raising $440!

Whitfield Elementary Jeans Day
Teachers from Whitfield Elementary School in West Lawn, PA each donated $15 to wear jeans for a cause on May 17th, 2013. Two students that attend Whitfield Elementary have SMA. Teachers wore their bracelets and either purple or yellow shirts to support the cause and raise awareness. More than $235 was raised for Families of SMA!

Regina Bell
West Lawn, PA

Harriton High School Casual for a Cause
Students and staff at Harriton High School in Rosemont, PA held a Casual for a Cause dress down day on May 3rd, 2013. The event raised $150 for Families of SMA in memory of Billy Kanehann.

1st Annual Christmas Cupcake Sale in Honor of Quinn Comer
On December 23rd, 2013 Juanita Comer held a cupcake sale in honor of her child, Quinn Comer, SMA Type III, in Reading, PA. Thank you to Juanita for all of her hard work and raising $151 for Families of SMA!

Northampton County Dress Down Day
On December 3rd, 2013 employees of Northampton County in Easton, PA raised money for Families of SMA through a dress down day. Every Friday, employees pay $1 if they wish to ‘dress down.’ Our donations are given to those organizations that are suggested by the employees. Although we wish this amount were larger, we hope that this gift from the employees of Northampton County will begin to address some of the challenges that you face. More than $100 was raised for Families of SMA.

John Stoffa, County Executive
Easton, PA

Note: The amounts raised and shown are totals as of June 21st, 2013 and
10th Annual Charity Golf Tournament 2013

The 10th Annual Charity Golf Tournament was held on May 20th, 2013 at Pradera Golf Club in Parker, Colorado and was another well attended event! The morning started off with some unexpected rain showers, but everyone still completed an enjoyable round of golf! We appreciate the ongoing support of the golfers who come out to play every year and we love to welcome new golfers to the tournament!

Our winners this year were:

1st place: Lewis Padilla, Larry Subia, Marcus Cordova, Andy Vigil
2nd place: Dave Moorehead, Martin Faith, Blake Anderson and Thom Mattson
3rd place: Ian Culverhouse, Josh Herrick, Kyllion Chaffin, Make Saliger

Congratulations winners and thank you to all of our generous supporters! This year we were able to raise over $20,000 for Families of SMA.

Gillian Faith
Centennial, CO

Rocky Mountain Beer & Cheese Pairing

On June 6th, 2013 the Rocky Mountain Chapter hosted the Evening of Hope Beer & Cheese Pairing in Denver, CO. This year’s event featured an array of beer and cheese pairings from some of Denver’s finest producers while attendees viewed some of the finest art in the city at the Artwork Network Gallery. Thank you to Marla Marlow and Joy Spellman for all of their hard work in planning this event. This year’s Evening of Hope raised over $5,600 for Families of SMA!

Dave & Buster’s Night for SMA

On April 16th, 2013 families and friends came together for an evening of fun, food and games at Dave & Buster’s in Garden Grove, California. Everyone had a great time meeting new families and seeing old friends. Dave & Buster’s donated a percentage of sales to Families of SMA that totaled $128. See everyone next time!

Rickk and Autumn Montoya
Garden Grove, CA
The 10th Annual Jacob’s Run, Walk & Roll to Cure SMA

The 10th Annual Jacob’s Run, Walk & Roll to Cure SMA was held this year on April 21st, 2013 in Boca Raton, Florida. At this landmark event that has become a staple to local south Florida residents, the Jacob Isaac Rappoport Foundation celebrated a gathering of friends and families for ten years in a row. Many of the close friends in attendance at the beautifully sunny Sunday morning walk have been coming to the event for the majority of those ten years. Some have even been there for all 10 walks! Additionally, last year, the event reached a total cumulative fundraising milestone of $1 Million for Spinal Muscular Atrophy (SMA) research and support. This year’s event added another $95,000 towards SMA research and support programs.

A special part of the event this year included Jordan, Jacob’s older sister, reading a letter that she wrote to Jacob on what would’ve been his 11th birthday last December. She wondered aloud what type of brother he would be, if he were still alive: “I only wish that instead of sitting around wondering what you would have wanted your birthday to be like, we could actually be out celebrating. Every day, I wonder what you’d be like and what kind of stuff you’d be into. […] I’m sure that on your birthday, all the people in Heaven give you a humungous party. I bet that you get a super big cake with lots of chocolate frosting, and you get to hang out with Sawyer, Peyton, Shelly, Tyler, and all your angel friends. Last night, I was reading mommy and daddy’s journal on the website. The daily struggle that Mommy and Daddy had to go through made me sad, but I was happy that your life was special to so many people. The amount of lives you touched and amount of people you filled with joy is endless.”

The Jacob Isaac Rappoport Foundation was founded in memory of Jacob, Shaina and Adi Rappoport’s son and shooting star, who had SMA Type I. Jacob was born December 27th, 2001 and was an alert and happy baby. When he was nearly four months old, Jacob was diagnosed with Spinal Muscular Atrophy. Soon after Jacob’s diagnosis, Adi and Shaina quickly connected with Families of SMA and many other SMA families. Jacob passed away on October 1st, 2002 at the age of nine months. Through Jacob’s Run, Walk & Roll, The Foundation’s main fundraising event, his memory lives on in the hearts of hundreds of family members, friends, colleagues and local SMA community members.

Thank You!

In ten years, The Jacob Isaac Rappoport Foundation has made a tremendous impact on the entire SMA community. In raising over $1 Million, The Foundation has supported many diverse programs, including:

- Multiple basic research awards which led to breakthrough discoveries on the genetic cause of SMA.
- New drug discovery programs, including the first ever new SMA drug program to be approved to begin clinical trials.
- Type I Care Packages
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**SOUTH JERSEY/DELAWARE**

**Delaware**

2013 Delaware Marathon

On May 12th, 2013 Team Families of SMA had an amazing 49 participants run in the 2013 Delaware Marathon! Some ran the full marathon, while others completed the half marathon, 4-person relay, or 8-person relay. Thanks to the incredible efforts of all the participants, more than $21,000 was raised for Families of SMA! Thank you to Jessica Moyer and the Cheslock Family for helping organize the event again this year.

**Zumba Away SMA**

On April 13th, 2013 Zumba Away SMA was held in Dover, DE and raised almost $3,800 for Families of SMA! The event was held in memory of Steven Moyer, SMA Type I, and Eden Cheslock, SMA Type II. Thank you to Jessica Moyer and Nicole Cheslock for organizing the event.

**Christmas Shopping for a Cause**

During the month of December 2012 representatives from Lia Sophia, Thirty-One, Scentsy and Arbonne Cosmetics teamed up for Christmas Shopping for a Cause! Up to 35% of profits from each sale was donated to Families of SMA in memory of PJ Desroches, SMA Type II! Thank you to Caroline Desroches, of Wilmington, DE, for all of her hard work on this event and for raising over $880 for Families of SMA!

**New Jersey**

8th Annual Steven’s Walk to Drum Out SMA

The 8th Annual Steven’s Walk to Drum Out SMA held at Newton Lake Park in Haddon Township, New Jersey on May 4th, 2013 was a great success! The event held in honor of Steven Potter, SMA Type III, raised over $18,000! We greatly appreciate the continued support of the participants and donors. Thank you to Terri Potter and all of the amazing volunteers for organizing the event again this year!

6th Annual Cure SMA Walk-n-Roll in honor of Katherine Santiago

On May 11th, 2013 the 6th Annual Walk-n-Roll in honor of Katherine Santiago, SMA Type II, was held in Milburn, NJ. The event raised over $12,300 for Families of SMA this year! Thank you to Allie Mazzella, the Mazzella Family and the Santiago Family for all of their hard work in putting together this event.

Walk-n-Roll in Honor of Alexus & Zane

My name is Alexus Dick and I have SMA Type II. On April 27th, 2013 we had our Walk-n-Roll in Honor of Alexus & Zane to help find a cure for SMA in Cherry Hill, NJ. We raised over $10,000!

I had so much fun, it was a huge park and handicap accessible. We had a bake sale, face paintings, water ice, balloon animals, and food.
We also raffled off gift cards and baskets from our local community. All of my friends came out to walk and support me. I am happy we raised money to help find a cure to help kids that suffer from SMA. I will never forget that day and the memories that we made.

I just want to say thank you to everyone that came to support Zane and myself. I also would like to thank my mom, Becky Dick and my 5th grade teachers, Mrs. Stephanie Rupin and Mrs. Kathi Niewochner who help me and support me in my everyday life. It means so much to Zane and me to have them in our lives. I am looking forward to next year’s big event!

Alexus and Zane Dick
Franklinville, NJ

Best Meatball in Secaucus
The Best Meatball Contest took place on April 28th, 2013 in Secaucus, NJ and drew awareness to SMA. The Secaucus-based relatives of the late Daniel Cevallos, SMA

Type I, along with the Immaculate Council Knights of Columbus, invited the community to help raise funds in the hopes of finding a cure for SMA. Nearly 100 people attended the event and raised over $3,000 for Families of SMA!

Giovanni Recalde
Secaucus, NJ

Wine Tasting Party
A member of our Knights of Columbus Council #542, George Kuzma, requested you receive the profits from a fundraising social, our annual Wine Tasting Party held in Township of Washington, NJ on May 11th, 2013. We are pleased to be able to donate $2,500 to Families of SMA for research and support for patients suffering with SMA.

Kenneth Kaphammer
Township of Washington, NJ

Remembering Camryn…Balltown Comes Alive
On November 30th, 2013 Douglas and Marci Kepple hosted Remembering Camryn…Balltown Comes Alive in memory of their daughter, Camryn Kepple, SMA Type I-II. The event, held in Marlton, NJ, included bands, a baseball skills competition, basketball and good ole’ fashioned vinyl records. Everyone had a great time and raised almost $2,500 for Families of SMA!

9th Annual SMA Charity Golf Classic
The Tennessee Chapter’s 9th Annual SMA Charity Golf Classic was held on May 13th, 2013 at Gettysvue Country Club in Knoxville, Tennessee. Fifteen teams competed in the 18 hole, four person scramble. During the tournament individuals competed in closest to the pin, longest drive and Happy Gilmore putting contests. A delicious lunch was provided by Ruby Tuesdays and dinner was courtesy of Papa Murphys. We can’t thank these donors enough for their generous contributions. Diners enjoyed the music of Andrew Strader and Taylor Kress of The Collies and everyone had a great time at the photo booth (thank you Randy Zinck). A silent
Auction, gamblers raffle, iPad raffle and live auction for a Big Green Egg smoker capped off a fun filled day that raised over $30,000! Once again Louise Ball, our fearless leader, organized an amazing event. A huge thank you goes out to all of our volunteers. We couldn’t have done it without all of you!

On a more serious note, we also brought hope to families who are just beginning this journey. The Hodgson family came to introduce themselves after the recent diagnosis of their son and ended up staying all day! One of the most rewarding moments of participating in fundraisers is being able to meet with families and spread awareness and hope. A radio interview earlier in the day with Louise Ball resulted in another family touched by SMA reaching out for support. As much as we hate hearing of a new diagnosis, we want these families to know that they are not alone.

Sarah Boggess
Knoxville, TN

3rd Annual Jellico High School Rubber Duck Race

The Jellico High School Student Council held their 3rd Annual Jellico High School Rubber Duck Race on March 21st, 2013 in Jellico, TN. Students sold duck numbers throughout March and then raced the ducks at nearby Indian Mountain State Park. The duck that made it to the spillway first won a Kindle Fire. This year’s race raised $1,500 for Families of SMA. Despite it being a cool dreary day many local people came to watch the race and to see how their ducks fared.

The event has been held the past three years following the annual Easter Egg Hunt. The race was started in honor of Casey Douglas, who was a member of the Council for four years and has SMA. She has gone on now to a local college and is proud to have started the race. She is making the council proud by doing well in college. Though Casey has moved on, the students still attending Jellico High School want to and will continue to host the race for years to come!

Barb Carter
Jellico, TN

InsideSMA.com Proceeds

We decided to donate a portion of InsideSma.com sales from 2011 and 2012 totaling $590 in honor of your daughter, Nora Gooden, SMA Type I. We plan to continue offering arm and leg slings to children with a percentage of sales going to Families of SMA in 2013. We can’t thank you guys enough for the love and support you have shown our family.

Love,

TJ, Jaime and Nora Gooden
Knoxville, TN

The Collies and Sea Legs Benefit Concert

The Historic Grove Theater in Oak Ridge, TN was the place to be on May 24th, 2013 for the Concert for the Cure. The Collies, a very talented local band, hosted the event with opening act The Sea Legs. Over 100 were in attendance to enjoy an evening of music, raising awareness to a younger audience and over $500 for Families of SMA. Thank you to Jana Strader and Lise Murphy for organizing the concert and to The Collies and The Sea Legs for a wonderful performance.

Sarah Boggess
Knoxville, TN
On May 11th, 2013, over 400 registered runners and walkers gathered at the Villagio Center in Katy, Texas for the Savannah Smiles Fun Run and 5K. With a beautiful start to a great day, Maci Medford, an upcoming national artist, sang the Star Spangled Banner. It was humbling to see so many families dedicated to bringing awareness to SMA in honor of Savannah Sue Norton, SMA Type I, who became an SMA angel on April 12th, 2013. The event had a great play area for kids, with a jump house and a kid’s tent offering face painting and special tiles where they painted butterflies on them for a future bench in Savannah’s memory. Several local food vendors came out in support offering massages, delicious food and beverages, and upbeat music to keep the celebration going. In addition the silent auction and door prizes were a hit. Added to this event was a couple of lemonade stands by a group from our elementary school and a few of Savannah’s friends that wanted to help raise funds. Bonnie Holland Elementary also raised $1,323 by sponsoring Bubbles for Savannah where the kids could buy bubble gum for $1 and chew it at school. All in all our goal was set for $15,000 and amazingly we raised over $43,000. Unbelievable support and love in this Katy community. A huge thank you to all the close friends, family members and the incredible Katy area for making this event such a success. I always say it takes a village and I have a great support system here in Katy, TX where once no one new what SMA was now thousands are bringing awareness and fighting for a cure.

Sheree Norton
Katy, TX

Kadence’s Krew to Kure SMA

Kadence’s Krew to Kure SMA ran in the Corpus Christi Beach to Bay Relay Marathon in Corpus Christi, TX in May 2013 to raise funds and awareness for SMA! The team raised $300 for Families of SMA! Kadene’s Krew to Kure SMA will participate again in the BrAvery 5K Run in Fredreicksburg, TX on August 17th, 2013. They had 24 on the team last year and expect almost 40 this year!
Pig Roast Fundraiser

Brad and Peggy Shiesley invited area friends and SMA families to our pig roast in honor of Kale Shiesley, SMA Type II, on June 22nd, 2013. The weather was threatening rain most of the afternoon, but the day turned out awesome. The kids, young and old, participated in a balloon toss, kickball, and fishing contest. Jenna Boguhn won the contest with the biggest and the most fish caught. There were big smiles everywhere.

Everyone brought a donation and a dish to pass. The pig was ready at 5:00 p.m. and there were plenty of salads, fruit, desserts and drinks to make a great feast. A total of $1,759 was raised to “roast” SMA.

Thank you Brad and Peggy for a great time!
Karen Shiesley
North Towanda, NY

PCNS 1st Annual Trike-a-Thon

The Pittsford Nursery School in Pittsford, New York held its 1st Annual Trike-a-Thon in May 2013 in honor of two former students affected by SMA, Emma and Ruby Cannady. The event raised $185 for Families of SMA! Thank you to Lisa Foster for organizing this event.

Radzik’s Birthday Party in Memory of Kennady Quinnell

Donations were collected in memory of Kennady Quinnell, at her Great Grandmother Raszik’s birthday party this year. Over $520 was raised for Families of SMA. The organization has been a blessing for our family, and we thank everyone for the support and love they have given us through the past couple years.

Love,
The Quinnell Family
Milwaukee, WI

For the latest news visit the FSMA website:

www.curesma.org
Hello Everyone,

Jenica has been keeping a very big secret since February and we finally get to tell! Jenica is being awarded the Medal of Honor by Girl Scouts USA! This is a very rare award and our local council believes she is the first recipient from Indiana.

Last fall Jenica’s morning nurse had a severe case of vertigo (didn’t know that was it at the time). Her nurse was physically unable to care for her. Jenica stayed calm and asked her nurse all of the right questions to make sure it wasn’t her blood sugar as she is an insulin dependent diabetic. She had to work to get the deadbolt undone on our front door for the back up nurse that was coming. She stayed right by her nurse and talked to her to keep her calm and alert. When the back up nurse arrived she debriefed him and then told him where her equipment was he would need to check the sick nurse. She also had to walk him through getting her ready for the bus.

To get the actual award, we had to fill out an application that included an essay from Jenica on what she did and letters from the 2 nurses involved. In addition to the medal, Jenica will also get a letter of recognition from the GSUSA CEO.

We are in the process of planning the awards ceremony with Girl Scouts of Kentuckiana. Their PR department is getting everything ready for a press release.

Julie Barksdale of Indiana

My daughter, Julia Carrington, started a club this year in her middle school in Pike County, GA. The SMACK club (SMA Cure Krusaders) raised a little over $2,000 in honor of Julia’s little brother, Jamey, who has SMA Type II. The group sold donuts and beanie babies, hosted a skate night at the local roller rink and sold donated T-shirts. I am enclosing a pic of the check being presented to Rio at the awards ceremony in May.

Thank you so much!

Rachel Carrington of Georgia

In July 2013, Bryson Crippen, from Weatherford, Texas, requested his friends bring a donation for FSMA instead of gifts to his 10th birthday party, in honor of his friend, Seth Gilley. He and his friends raised $200 for FSMA! They sent him a picture of all of them posing with Seth’s favorite superhero, the HULK. Thank you, Bryson, for being such a terrific and thoughtful friend to Seth and for helping FSMA find a cure!

Race in Honor of Victoria Meneghini

On February 10th, 2013 I ran a race in memory of my sister Victoria Meneghini who died from SMA Type I at an early age. I raised money for Families of SMA and donated $200 in her memory. I am always happy to help and I hope this money is used well. I also hope you will find the cure to this disease. This money should help. Good luck!

Philip Meneghini
Ipswich, MA

Joey Romanowski’s Birthday Fundraiser in honor of William Johnson

This fantastic kid in our community, Joey Romanowski, has done it again! For 4 years in a row (since the age of 8) he has decided to give up asking for birthday presents at his party and instead asks his friends to make a donation to Families of SMA to honor him and his buddy William Johnson, SMA Type II. Joey’s party raised close to $300 this year! All in all it was a great success and Joey’s mom said all of his friends felt great contributing to our cause.

Heidi Johnson
Medfield, MA

Lemonade Stand in Honor of Ayden Trammell

Arien Pool raised $106.33 by selling lemonade and cookies on June 7th, 2013. Arien, our granddaughter, lives in Fairbanks, Alaska. We were looking at Directions and reading the stories and she decided to raise some money for SMA research. Her cousin (my great nephew) is Ayden Trammell from Lincoln, Nebraska. Ayden will turn 7 this month and has SMA Type I. Thank you for all you do.

Sincerely,
Deb LeBlanc of Kansas

Piggy Bank Donation in Honor of Ariana Dindzans

Emma Rodrigues is my daughter, Ariana Dindzans’s best friend. Ariana has SMA Type III. Emma emptied her piggy bank one day and gave Mrs. Dindzan $32.21 in cash to “help find a cure for SMA, so Ariana could walk again someday.” Emma is a young girl with a HUGE heart!
Morgan Turner’s Birthday Party
My daughter, Morgan Turner, asked friends at her birthday party on January 15th, 2013 in Maple Grove, Minnesota to bring donations for SMA in lieu of a gift. Morgan was able to raise $75 in honor of Angie Lee, SMA Type II, who lives in Naperville, IL.

Jen Turner
Maple Grove, MN

Hamman Family Bake for a Cure Bake Sale
My daughter Savannah and her friend Sarah Lindflott held a “Bake For A Cure” bake sale this past April 24th, 2013 in Naperville, IL. The event was a huge success and they raised $151 in memory of Henry Hamman, SMA Type I!
Savannah and Sarah have been friends since the day they were born. As part of their 4th grade Sunday School curriculum they learn about service and how God wants us to do things for others. Thus hatched the “Bake For A Cure” idea. They did most of the baking themselves, and did an incredible job marketing their sale. They held the bake sale after their regular church choir practice, a great audience for baked goods! Best of all, they had a great time doing it and were truly amazed by their success. They also were building awareness of SMA with materials they had displayed and by talking with their customers. They have both actively helped us with our Luau’s in the past. Neither of them knew Henry yet they have such a great understanding of SMA and the great work Families of SMA is doing to find a cure!

Lisa Hamman
Naperville, IL

Make-A-Wish App

When children with life-threatening illnesses are granted a wish through the Make-A-Wish® Foundation, most choose to meet someone famous or visit Disneyland. But when Ryan Cottor was offered a wish, he picked something a little more unusual.
The 12-year old has spent his life battling Spinal Muscular Atrophy (SMA), a neuromuscular disease that has left him limited use of his body. But with his hands, he can play video games. So he made a decision. Why not create a game of his own?
The Make-A-Wish® Foundation contacted Pixel Jinn, a collective of video designers from throughout the West. The group worked with Ryan to create the game "Monkey Wars," which was made available in the iTunes store in July. The whole process took about four years!
Ryan, who loves video games, was the creative director, which means he called all of the shots. Monkey Wars is his vision brought to digital life.
It’s a challenging game. The goal is to defend "monkey islands" from humans who want to take over. The monkeys must throw "banana-rangs" to protect their home and fend off the humans.
Monkey Wars is available for iPhone, iPod touch and iPad. It requires iOS 6.0 or later and is free to download.
Check it out at:
Hi. My name is Jake. I am 10 years old and I am a hockey player. I play Power Wheelchair Hockey for the Philadelphia PowerPlay.

Power Chair hockey is almost like ice hockey except we play on a gym floor and use lightweight sticks and a wiffle ball instead of a puck. If a player has difficulty holding a stick they can tape it to their chairs. There are no age limits but most players are teenagers or older. Players must use power wheelchairs for most of their daily lives to be considered for the team.

This year my team hosted the 2013 US PowerHockey Tournament from August 2nd - August 4th at Neumann College in Aston, PA.

Teams from Michigan and North Carolina competed to see who would become this years Championship team.

The Philadelphia PowerPlay ended up playing the reigning champs, the Michigan Mustangs in the final round. I am happy to tell you that my team, for the very first time, won the championship by a final score of 9-4. It was awesome!

Our regular season will be starting up in September. If you are interested in playing PowerHockey please visit www.philadelphiapowerplay.com. There are also teams in Michigan, Minnesota, North Carolina and Canada. It’s a great sport and you will meet amazing people but most importantly you will have a lot of fun!

Jake Saxton, #18
Philadelphia PowerPlay
Isis Reports Follow-Up Data From Phase I Study In Children With Spinal Muscular Atrophy

Isis Pharmaceuticals, Inc. announced that follow-up preliminary data from a single dose, open-label Phase I study of ISIS-SMNRx in children with spinal muscular atrophy (SMA), show that most SMA children receiving the two highest doses of the drug (6 mg and 9 mg) continued to show improvements in muscle function tests up to 14 months after a single injection of the drug. The Phase I data, including these preliminary follow-on data, will be presented at the International Congress of the World Muscle Society by Dr. Kathy Swoboda on Oct. 3, 2013. SMA is a severe and rare genetic neuromuscular disease characterized by muscle atrophy and weakness and is the most common genetic cause of infant mortality. ISIS-SMNRx is an antisense drug designed to treat all types of SMA.

FDA Approval to Nationwide Children’s Hospital for Phase I Clinical Trial of Systemic AAV9-Delivered SMN Gene For Spinal Muscular Atrophy

Following the submission of an Investigational New Drug application, the FDA has given its approval to physician-scientists at Nationwide Children’s Hospital to begin a Phase I clinical trial of a systemic AAV9-delivered human SMN gene.

The clinical trial is expected to begin in early 2014 and will be limited to SMA Type I patients, ages birth to 9 months.

Previous research from Nationwide Children’s Principal Investigator Brian Kaspar, PhD, demonstrated the AAV9 viral vector crossing the blood brain barrier. Based on this research and additional preclinical studies, the SMN gene will be delivered by injection into the bloodstream as part of this Phase I trial. Neurologist Jerry Mendell, MD, director, Center for Gene Therapy at Nationwide Children’s, will lead the study.

Pfizer RG3039 Program for Spinal Muscular Atrophy Reaches Development Milestone

Reaching the first milestone was triggered by completion of specific program activities and coincides with the successful completion of all transition obligations to Pfizer by Repligen. Repligen Corporation announced that it has achieved a milestone from Pfizer, Inc. under the terms of the companies’ exclusive worldwide licensing agreement for the development of compounds to treat spinal muscular atrophy (SMA). RG3039 is orally bioavailable, brain-penetrant small molecule that has been shown to be an inhibitor of the mRNA decapping enzyme, DcpS. The pharmacological characterization of RG3039, reported recently in new studies in Human Molecule Genetics, demonstrates RG3039 can extend survival, improve function, and impact neuromuscular pathology in three SMA mouse models of varying disease severity.
Families of Spinal Muscular Atrophy (SMA) funded and directed the preclinical development of RG3039 with an investment of more than $13 million. This was the first drug discovery program ever conducted specifically for SMA. Repligen licensed RG3039 in 2009 from Families of SMA for clinical development. In January 2013, Pfizer licensed the program, marking a significant advance for the SMA community by securing the commitment of one of the world’s largest pharmaceutical companies to develop potential treatments for SMA. It is currently being tested in Phase 1B human clinical trials.

RG3039 is orally bioavailable, brain-penetrant small molecule that has been shown to be an inhibitor of the mRNA decapping enzyme, DcpS. The pharmacological characterization of RG3039, reported in the two new studies in Human Molecular Genetics, demonstrates RG3039 can extend survival, improve function, and impact neuromuscular pathology in three SMA mouse models of varying disease severity.

The first paper from the laboratory of Dr. Christine DiDonato at the Lurie Children’s Hospital in Chicago is entitled “The DcpS inhibitor RG3039 improves survival, function and motor unit pathologies in two SMA mouse models”. It shows that when administered to an intermediate mouse model of SMA called 2B/−, the drug RG3039 provides >600% survival benefit (median 18 days to >112 days) when dosing began at postnatal day 4. These mice are presymptomatic on day 4 of life, highlighting the importance of early intervention. RG3039 also significantly increased the number of gems and cells with gems in motor neurons, which is used as an indirect measure of SMN.

A second mouse model was also tested in this study. Taiwanese hemizygous SMA mice were used to test RG3039. This model was generated and described by Dr. Hong Li and has a similar survival time as Delta7 SMA mice. RG3039-treated SMA mice had a median survival that was 38% greater than vehicle-treated SMA mice, when dosing started at postnatal day 4. Furthermore, when placed on their back RG3039-treated SMA mice were able to right more quickly. Therefore, these collective results suggest RG3039 is able to slow disease progression in these mice even when the drug is administered after symptoms have emerged.

“This is another very exciting step on the path toward developing treatment for SMA patients,” said Charlotte Sumner, MD, Associate Professor Of Neurology at John Hopkins University. “Families of SMA particularly deserves huge congratulations for initiating the development of this novel drug, which has now been licensed to Pfizer. It was a pleasure to work together with the teams from Repligen, Dr. DiDonato, Dr. Ko and Dr. Pellizzoni’s laboratories on this project. We believe that the improvements in neuromuscular junction structure and function observed in SMA mice after treatment with RG3039 are likely to be relevant for SMA patients and the possibility that different types of treatments could work additively in patients is also exciting.”

RG3039 Background:

FSMA began the Quinazoline/RG3039 program in 2000 at the very initial stages of drug development, when risk is the highest. It was the very first industrial drug program for SMA ever conducted. The direction from FSMA provided the positive results necessary to license the program to Repligen and now Pfizer to leverage resources and expertise for clinical development. Through FSMA’s leadership and research funding of over $13 million, a drug candidate has been created that treats the underlying cause of SMA.

“There is a critical need to expedite potential treatment solutions for rare diseases such as spinal muscular atrophy, where patients have such limited options,” said Jose Carlos Gutierrez-Ramos, Senior Vice President, Pfizer BioTherapeutics R&D. “This partnership will combine our expert capabilities in advancing molecules for genetic diseases with Repligen’s leading SMA program.”

The FSMA research model is to fund early stage drug discovery programs for SMA and then partner with companies for later stages and accelerated clinical development. At the earliest stages of drug development, programs have less than a 1% chance of success. This inherent risk along with small patient populations has traditionally hindered industry from working on orphan diseases.
“We are very happy that our research was able to assist in the pharmacological characterization of RG3039 and contribute to dose selection and exposure estimates for the first studies with RG3039 in human subjects,” said Dr. Christine DiDonato from Ann Robert H. Lurie Children’s Hospital of Chicago Research Center. “We are indebted to my colleague, Nancy Kuntz, MD, and co-author laboratories of Drs. Jasbir Singh and Mike Kiledjian along with Repligen and Families of Spinal Muscular Atrophy (SMA), who initiated the development of this drug candidate. The depth of our manuscript would not have been possible without this group collaboration. Also, it is reassuring that our results of improved motor unit dysfunction in SMA mice treated with RG3039 mirror the independent findings by the laboratories of Drs. Sumner and Ko. Improved neuromuscular function is an obvious goal for SMA therapies. Overall, this is an exciting time as we take another step forward towards developing therapies for SMA patients.”

The second paper from the laboratories of Dr. Charlotte Sumner at Johns Hopkins University and Dr. Chien Ping Ko at the University of Southern California is entitled “The DepS inhibitor RG3039 improves motor neuron function in SMA mice”. Dr. Livio Pellizzoni’s group at Columbia University also contributed to this study. In this study of the effects of RG3039 on the Delta7 mouse model, drug distributed to CNS tissues where it robustly inhibited DepS enzyme activity, but minimally activated SMN expression or snRNP assembly. Treated SMA mice showed a dose-dependent increase in survival, weight, and motor function. Drug-treated SMA mice demonstrated a 26% increase in median survival compared to vehicle-treated mice and a 19% increase in maximal weight achieved. Improved righting times were evident and improved ambulatory index scores observed, which indicates significant improvements in motor function. This was associated with improved motor neuron and neuromuscular junction (NMJ) synaptic innervation, NMJ function, and muscle size.

RG3039 also enhanced survival of conditional SMA mice in which SMN had been genetically-restored to motor neurons. These data indicate that RG3039 treatment when combined with motor neuron restoration of SMN had better than additive effects, suggesting that while RG3039 can improve motor neuron function, it has beneficial effects that are independent of motor neurons.

Families of SMA Awarded Grant from the National Institute of Health for The Annual Spinal Muscular Atrophy Research Group Meeting

Families of SMA has been awarded a grant from the National Institute of Neurological Disorders and Disease (NINDS) at the National Institutes for Health (NIH). We thank them for the fantastic support that helps bring together the SMA research community along with our families.

The SMA Research Group Meeting is the largest research conference in the world for SMA. It was held June 13, 14, 15 at the Disneyland Hotel in Anaheim, California. Over 225 researchers gave 110 updates on the latest breakthroughs in SMA research. Researchers are registered from 15 different countries, 70 different organizations, and 17 biotech and pharmaceutical companies.
New Funding of SMA Patient Care Research Announced at The Annual SMA Conference.

Families of SMA launched a new program to fund care research to drive improvements in patient care in Spinal Muscular Atrophy. Four new research grants were awarded at The 2013 Annual SMA Conference:

- $50,000 to Oscar Mayer, MD at The Children’s Hospital of Philadelphia to study Decisions Related to Goals and Limitations of Care and the Challenges in Making Them for Parents of Children with SMA
- $49,912 To Timothy Lotze, MD at Texas Children’s Hospital to study a Quality Improvement Project to Reduce Gaps in Care in the Hospital Setting for Children with SMA Type I
- $50,000 To Matthew Halanski, MD at University of Wisconsin to study a Spinal Muscular Atrophy Database: a Multicenter Multidisciplinary Assessment
- $49,980 To Kathryn Swoboda, MD at University of Utah to study Glucose Load Tolerance and Fasting in SMA type II

Families of Spinal Muscular Atrophy has been funding critical research to develop a treatment and cure for the disease since 1984, along with providing important resources and support for families affected by SMA. This new research funding program announced is focused on improving care and the quality of life for SMA patients.

This funding will build on the consensus statement for the standard of care for SMA with data driven results on specific areas of SMA care. These results will be used to provide:

- Educational programs for professional medical providers, such as the FSMA CME day
- New family-focused care publications, such as the FSMA Care Series Booklets
- Peer reviewed journal publications to influence insurance coverage

The results of these projects will build an evidence base and demonstrate measurable, positive effects on the clinical management and lives of patients with SMA. This initial round of funding is designed to fund pilot studies which will demonstrate the feasibility of data collection, and which will then support submission of a larger study for FSMA or government funding.

This new funding program complements the three established FSMA research funding programs for basic research, translational drug discovery, and clinical trials to provide a comprehensive approach to developing a treatment and cure for SMA and improving care for all SMA patients:

1) Basic Research Funding to generate new ideas and approaches to treat or cure SMA
2) Translational Drug Discovery Funding to turn ideas into practical, safe and effective therapies
3) Clinical Trials Funding to establish resources and tools to test new drugs in humans
4) Clinical Care Funding to drive improvements in patient care

All applications were reviewed by the FSMA Medical Advisory Council, using a NIH-like scoring system based on both scientific quality and relevance to the Families of SMA research mission of improving care for all SMA patients.

Recruitment Update on the NeuroNEXT Trial on Spinal Muscular Atrophy Biomarkers in the Immediate Postnatal Period of Development Study.

The NeuroNEXT Network for Excellence in Neuroscience Clinical Trials (NeuroNEXT), Spinal Muscular Atrophy (SMA) Biomarkers in the Immediate Postnatal Period of Development study, led by Stephen Kolb, MD, PhD, has announced that as of June 1, 2013, 20 infants with no medical condition have enrolled in the study to support their enrolled infant peers with SMA. Within 4 months of beginning study enrollment, we were half-way to the recruitment goal of enrolling 27 children that do not have SMA. “We are thankful for the support of the SMA community. We recognize the commitment this study entails for the families and greatly appreciate the support. We are enrolling the children without SMA much quicker than we had originally anticipated. Exceeding a recruitment goal timeline is not something you get to claim in research very often. I know this achievement is because of the support of family and friends of the amazing SMA advocacy networks.” stated Dr. Stephen Kolb study Protocol Principal Investigator. This study is still recruiting 7 additional infants with no medical condition and 20 infants with SMA between 0-6 months of age at the time of enrollment.

We hope people will continue to spread the word with the YouTube™ video www.youtube.com/watch?v=f8xGDFjDLU to help us fulfill enrollment in record time.

For more information please call Amy Bartlett at 1-855- SMA- BIOM (1-855-762-2466) or Amy.Bartlett@osumc.edu.
Congratulations to Ta’Bria Neosha Collier of Roanoke, Indiana who has SMA Type I and is 19 years old, on your exciting accomplishment of graduating from Homestead High School!
Congratulations to proud parents, Dorothy and Jon, big brother, Owen, and big sister, Natalie Shuler on the birth of Jack Owen, born October 27, 2012!

Congratulations to Sarah and Chris Bonelli on the birth of their new daughter, Melia Jack, born on January 14, 2013!

Congratulations to the Granger family on the birth of their new baby boy, born on April 16, 2013!

Congratulations to the Patrick family on the birth of their newest addition, Grant Patrick, born on July 26, 2013!

Congratulations to James, Jennifer and big sister Addy Garner, on the birth of their new baby boy, Bryson William, born on July 31, 2013!

Congratulations to Peter Voskovitch, age 16, SMA Type III from Warrensburg, Missouri who recently received the National Wheelchair Basketball Association First Team Academic All American Award while competing at the National tournament this April. This award goes to the top ten academic students among all wheelchair basketball players across the nation. Peter was also recognized at the conference level. He competes with the Kansas City Pioneers wheelchair basketball team.

Congratulations to Olivia LeVoy from Flint, TX, who has SMA Type III, on being chosen to represent Texas in the national Ms. Wheelchair USA competition! Olivia is a 20 year old who is attending Tyler Junior College and majoring in agriculture. She has a passion for animals, and is the first Ms. Wheelchair USA finalist to be involved with the agriculture industry. Olivia currently has a herd of Lowline Angus and owns Rhinestone Cattle Company.

Congratulations the Butler family on the birth of their little boy, William McLean, born on May 6, 2013!
Congratulations to Mallory Armbrustmacher, 17, who has SMA Type III. Mallory was awarded the People’s Choice Award from the Governor’s Council on Physical Fitness for accepting the challenge. The Governor’s Fitness Awards serve as a platform for inspirational individuals and organizations for their pursuit, commitment and dedication to healthy lifestyles. Each award recognizes the many different ways the winners are inspiring and influencing Michigan residents to live a healthy lifestyle. The purpose of the Governor’s Fitness Awards is to recognize individuals, organizations and events that encourage others to make healthy choices by teaching, volunteering, role modeling or other community involvement. The awards event serves as a platform of appreciation and public recognition for these deserving citizens as well as a fundraiser for the Michigan Fitness Foundation. To learn more about the Governor’s Fitness Awards visit http://www.michiganfitness.org/governors-fitness-awards.

Mallory is a dedicated member of both the DeWitt Area Swim Team and the DeWitt High School Girls Swim and Dive Team. She swims year-round because of a deep love for the sport as well as the time she gets to spend with her friends and teammates at meets and practices. She inspires her team with her positive attitude and work ethic. The sport has provided her with the opportunity to form friendships with, as well as receive support from, coaches and teammates. Opponents often approach her after the race to congratulate her and tell her how she has inspired their fitness journey. Everyone in the stands cheers when she swims, regardless of what team they are there to support.
SHARING PHOTOS

Cameron Williams

Kate Saxton
Kale Shiesley
Kyler and Kale Shiesley

Ember Hinson

Maeve Coffey, Emily Lozina and Eva Grace Kelly

Oskar Karlsson
Katie Miller

Grace DeGraffenreid
Journee & Jace Weiberg

Conner Smith

Eva Grave Kelly
Brooklynn Santos
Grace DeGraffenreid with Grandma Sherry
The DeGraffenreid Family

The Kelly Family

The Patrick Family

The Grindle Family

The Smith Family

The Davis Family

The Williams Family

The Manfre Family
Isis Pharmaceuticals Initiates Clinical Study in Infants with Spinal Muscular Atrophy

Isis Pharmaceuticals, Inc. (NASDAQ: ISIS) announced in April the initiation of a Phase II study of ISIS-SMNRx in infants with spinal muscular atrophy (SMA). The study, which will begin enrolling patients soon, is a Phase II study in eight patients with infantile-onset SMA. The study, which initiates the Phase II/III program for ISIS-SMNRx in infants, is designed to provide data to allow Isis to define the optimal dose for the larger planned Phase II/III study in infants and to provide safety and tolerability data.

The Phase II study of ISIS-SMNRx is an open-label, multiple-dose, dose-escalation pilot study, which will include eight infants who have been diagnosed with SMA. To meet enrollment criteria, infants must be between the ages of three weeks and seven months, live in close proximity to a study site and pass screening evaluations conducted at study sites. The study will be conducted at centers in the United States and Canada.

For further study information, please visit www.clinicaltrials.gov and search for ISIS-SMNRx.

Isis completed an open-label Phase I study evaluating ISIS-SMNRx in children (age 2 and older) with SMA, in which ISIS-SMNRx was well tolerated at all dose levels when administered intrathecally as a single dose directly into the spinal fluid. Although the study was not designed to provide evidence of functional activity improvements in the Hammersmith Functional Motor Scale-Expanded, a measure of muscle function, were observed in a number of these children. In addition to the infant pilot study, Isis is also completing a multiple-dose, dose-escalation Phase Ib/IIa study of ISIS-SMNRx in children with SMA. Data from the Phase Ib/IIa study will provide information to determine the dose for the Phase II/III registration-directed study in children (age 2 and older) with SMA.

“SMA represents a serious unmet medical need with no currently available treatments. Based on its mechanism of action and encouraging preclinical and clinical data, ISIS-SMNRx could be an effective treatment for these very sick children, though additional work still needs to be done. The rapid advancement of this drug to this stage in development reflects the support from the SMA community and the success of the collaboration between Isis and Biogen Idec. Isis and Biogen Idec are committed to advancing the program for children with SMA,” said C. Frank Bennett, Ph.D., senior vice president of research at Isis. “ISIS-SMNRx is our first drug designed to intervene in the splicing of RNA to increase the production of a normal protein, SMN. Antisense drugs could offer novel new therapeutics for a number of severe neurodegenerative diseases. The encouraging safety data from this program and our preclinical and clinical experience in other neurodegenerative diseases support the broadening of our efforts to develop antisense drugs to treat such diseases.”

In October 2012, Isis Pharmaceuticals began their second SMA clinical trial, which is ongoing. It is an open-label, dose escalation study to assess the safety, tolerability, and dose-range finding of multiple doses of the drug delivered intrathecally to 24 patients with SMA. Click here for more information about this study.

Isis Pharmaceuticals exclusively licensed intellectual property from the University of Massachusetts to develop this new drug for Spinal Muscular Atrophy. Families of SMA provided over $500,000 in funding support for the University of Massachusetts’ research program responsible for creating this intellectual property. Click here to read about this licensing agreement and FSMA’s involvement.

ABOUT ISIS-SMNRx

ISIS-SMNRx is designed to alter the splicing of a closely related gene (SMN2) to increase production of fully functional SMN protein. The United States Food and Drug Administration granted orphan drug status and fast track designation to ISIS-SMNRx for the treatment of patients with SMA. Isis is currently in collaboration with Biogen Idec to develop and potentially commercialize the investigational compound, ISIS-SMNRx, to treat all types of SMA. Under the terms of the January 2012 agreement, Isis is responsible for global development and Biogen Idec has the option to license the compound until completion of the first successful Phase II/III study. ISIS-SMNRx is currently being evaluated in a Phase 1b/2a multiple-dose, dose-escalation study in children with SMA. In this study, children will either receive two or three doses of ISIS-SMNRx over the course of the study.

Isis acknowledges support from the following organizations for ISIS-SMNRx: Muscular Dystrophy Association, SMA Foundation, Families of SMA and intellectual property licensed from Cold Spring Harbor Laboratory and the University of Massachusetts Medical School.

“We are very pleased with the achievement of this clinical milestone of advancing a potentially disease-modifying drug treatment into later-stage clinical trials. This achievement is a culmination of the close interactions between basic researchers, families, clinicians and industry. Families of SMA applauds Isis for investing in and leading drug development efforts for this devastating, orphan disease,” said Jill Jarecki, Ph.D., Research Director of Families of SMA.
PTC Therapeutics Announces Selection of a Drug Development Candidate for SMA.

PTC Therapeutics, Inc. (NASDAQ:PTCT) announced on August 8 the selection of a development candidate in its spinal muscular atrophy (SMA) collaboration with Roche and the SMA Foundation.

“We are excited about this important achievement in our SMA program”, stated Stuart Peltz, Ph.D., Chief Executive Officer of PTC Therapeutics. “This program exemplifies PTC’s technology platform and its chemistry and biology expertise applied in an area of great unmet medical need. We are grateful for the exceptional commitment and dedication from our PTC team, as well as from our partners, Roche and the SMA Foundation. The discovery and advancement of a potential new treatment for SMA is a significant milestone for SMA patients and their families.”

SMA is caused by a missing or defective SMN1 gene, which results in reduced levels of the survival motor neuron (SMN) protein. It is a genetic neuromuscular disease responsible for the early death of motor neuron cells within the spinal cord leading to muscle atrophy and eventually death in the most severe form of the disease. The oral small molecule compounds in the program target the underlying cause of the disorder by increasing SMN protein levels in the nervous system, muscles, and other tissues. It is estimated that SMA affects approximately 10,000 to 25,000 children and adults in the United States, and that between one in 6,000 and one in 10,000 children are born with this rare disorder.

Luca Santarelli, Head of Neuroscience and Small Molecule Research at Roche commented: “The compelling science behind this project and the highly synergistic alliance between Roche, PTC Therapeutics and the SMA Foundation are the groundwork for this potential therapeutic program. We consider every step towards a treatment option in SMA of high importance for patients and families affected by this devastating and currently incurable condition.”

The SMA program was initially developed by PTC Therapeutics’ in partnership with the SMA Foundation. The SMA Foundation was established in 2003 to accelerate the development of a treatment for SMA. In November 2011, Roche gained an exclusive worldwide license to PTC’s SMA program.
Reliable tests to determine if a new therapy is working are critically important for future SMA clinical trials. The Project Cure SMA study “Reliability and Validity of the Test of Infant Motor Performance Screening Items (TIMPSI) for Infants with Spinal Muscular Atrophy Type I” recently published in Pediatric Physical Therapy evaluates a possible clinical endpoint for SMA trials. Currently a limited number of tests are available to measure motor function during clinical trials of infants with SMA.

The purpose of this study was to examine the reliability and validity of the Test of Infant Motor Performance Screening Items (TIMPSI) in infants with type I Spinal Muscular Atrophy (SMA). Items on the TIMPSI test cover a span of motor function. These items capture the range of skills that infants with SMA Type I typically achieve and allow for change in either direction. Due to the short nature of the test the authors predicted that it would be well tolerated and feasible to administer to infants with SMA Type I.

After training, twelve evaluators scored videos of infants with SMA Type I to assess reliability between evaluators. Reliability of individual evaluators was determined to be excellent by having 9 evaluators test a total of 38 infants twice. Reliability upon retest between the same evaluator was also high. Importantly, TIMPSI scores were shown to correlate to the ability to reach, an important functional skill in children with SMA Type I. The ability to reach appears to vary across the spectrum of SMA Type I and seems indicative of underlying strength, making it a clinically meaningful measurement. Overall this study suggests the TIMPSI is feasible to administer and can be used to reliably assess motor function in infants with SMA Type I in multi-site clinical trials.

“As new therapies emerge for infants with Spinal Muscular Atrophy Type I, it will be critical to have a motor outcome measure that is easily administered, valid and reliable. This study demonstrates that with appropriate training the TIMPSI is a reliable measure, and with minor modifications is both safe and feasible to use in the clinic and research setting. In addition, initial steps toward validation of the TIMPSI for this population are described. The next step is to look at how the test performs over time. For example, is the TIMPSI responsive to change and can it detect meaningful change over time? This work culminated from a larger Project Cure multi-site Trial of valproic-acid and carnitine in infants with SMA, and would not have been possible without the efforts of the entire Project Cure Team and support from Families of SMA.” said lead author Kristin Krosschell of Northwestern University Feinberg School of Medicine.

TIMPSI is being further evaluated in the NINDS NeuroNext SMA Biomarker Trial, along with other trial endpoints and biomarkers. NeuroNEXT is an abbreviation for The Network for Excellence in Neuroscience Clinical Trials, abbreviated NeuroNext, which is a twenty-five site national clinical trial network created by NINDS of the NIH to test promising new therapies for both pediatric and adult patients with neurological diseases. The SMA study is being conducted by the NeuroNEXT Network at 15 sites around the United States. Stephen Kolb, MD, PhD of Ohio State University is the Protocol Principal Investigator for the study. Up to 54 volunteers will take part in this research study. Information will be collected from two groups: infants diagnosed with SMA and infants without a neurological disease (we will call this group the control group). All infants will be between 0-6 months of age at the time of enrollment.

Families of SMA is helping fund patient travel for the study.
SHARING PHOTOS

Maia Shockley

Kyan Kulas

Austin and Jeff Olander

Alli Williams with big sister Amaya

Madi, Molly and Morgan Ramirez

Madi Ramirez

The Ramirez Sisters

Lizzy Hallam

The Williams’s Kids

Alexander Davis

Makenna Rose DeGrush

Lily Kelly
Spinal Muscular Atrophy Clinical Trial Data Presented at the 2013 American Academy of Neurology (AAN) Meeting

The 65th Academy of Neurology (AAN) Meeting was held the week of March 18 in San Diego, California. Data from several SMA projects, including two clinical trials, will be presented to 10,000 clinicians and researchers in attendance. These include presentations by Isis Pharmaceuticals on their Single Dose Phase I Clinical Trial on ISIS-SMNRx and by Project Cure SMA on their Carnival Type I Clinical Trial.

The first clinical trial update at AAN was from Isis Pharmaceuticals. They reported on their Phase I study of ISIS-SMNRx. This trial as single-dose, dose-escalation study designed to assess the safety, tolerability and pharmacokinetic profile of the drug in children with SMA between the ages of 2-14 who were medically stable. ISIS-SMNRx was administered intrathecally as a single injection directly into the spinal fluid. The results shown at AAN indicate that ISIS-SMNRx is well tolerated when given as a single dose at the dose levels evaluated in this study and no safety concerns have been identified. Results from the study support continued development and further examination of ISIS-SMNRx in a longer multiple-dose clinical study in SMA patients.

In October 2012, Isis Pharmaceuticals began their second SMA clinical trial, which is ongoing. It is an open-label, dose escalation study to assess the safety, tolerability, and dose-range finding of multiple doses of the drug delivered intrathecally to 24 patients with SMA. See www.clinicaltrials.gov for more information on this study.

Isis Pharmaceuticals exclusively licensed intellectual property from the University of Massachusetts to develop this new drug for Spinal Muscular Atrophy. Families of SMA provided over $500,000 in funding support for the University of Massachusetts’ research program responsible for creating this intellectual property.

The meeting abstract for the Isis single dose Phase I Study is below:

Results of an Open-Label, Escalating Dose Study To Assess the Safety, Tolerability, and Dose Range Finding of a Single Intrathecal Dose of ISIS-SMNRx in Patients with Spinal Muscular Atrophy, Claudia Chiriboga, New York, NY, Kathryn Swoboda, Salt Lake City, UT, Basil Darras, Boston, MA, Susan Iannaccone, Dallas, TX, Jacqueline Montes, New York, NY, Heather Allen, Salt Lake City, UT, Rebecca Parad, Boston, MA, Shanda Johnson, Dallas, TX, Darryl De Vivo, New York, NY, Daniel Norris, Carlsbad, CA, Katie Alexander, Carlsbad, CA, Frank Bennett, Carlsbad, CA, Kathie Bishop, Carlsbad. OBJECTIVE: This first-in-human study was conducted to evaluate the safety, tolerability, and pharmacokinetics of escalating single intrathecal doses of ISIS-SMNRx (ISIS 396443) in patients with SMA. BACKGROUND: ISIS-SMNRx is an antisense oligonucleotide (ASO) molecule designed to alter splicing of SMN2 mRNA to increase the amount of functional SMN protein produced. Results from experiments in SMA mouse models showed that ISIS-SMNRx effectively altered SMN2 mRNA splicing and increased SMN protein in the spinal cord and had a significant effect on functional and histological measures of neuromuscular health, including survival, when delivered to the CNS. DESIGN/METHODS: A single dose of ISIS-SMNRx was delivered by intrathecal injection to medically stable SMA patients 2-14 years of age. Four escalating dose levels (1, 3, 6, 9 mg) were examined in cohorts of 6-10 subjects (n=28). Subjects were followed for 29 days (cohorts 1, 2) or 85 days (cohorts 3, 4) post-dosing and monitored for drug safety and tolerability. Plasma drug levels were measured over the first 24 hours and CSF drug levels were assessed at 7 days post-dose. RESULTS: No serious adverse events or dose-limiting toxicities were reported. All adverse events were mild or moderate in severity and none were related to dose level of ISIS-SMNRx. No drug-related changes in neurological exams, laboratory assessments (including CSF safety and cytokines), or systemic evaluations were reported. Intrathecal injections were well tolerated in SMA children. Analysis of drug pharmacokinetics in plasma and CSF indicate that drug levels in SMA patients were dose-dependent and consistent with levels predicted from pre-clinical animal studies. CONCLUSIONS: ISIS-SMNRx is well tolerated when given as a single dose at the dose levels evaluated in this study; no safety concerns have been identified. Results from this study support continued development and further examination of ISIS-SMNRx in a longer multiple-dose clinical study in SMA patients. Supported by: Isis Pharmaceuticals, Inc. Carlsbad, CA.

The second study reported on at AAN was the CARNIVAL Type I Clinical Trial,
which was funded by Families of SMA.
This project demonstrated that recruitment and retention of sufficient numbers of type I infants for a multicenter clinical trial is feasible. This study will pave the way for future multi-center drug studies conducted in infants with SMA, including those by Isis Pharmaceuticals on Isis-SMNRx. This study failed to support efficacy for VPA/L-carnitine in type I infants when compared to cohort of age and gender matched natural history subjects.

The Project Cure AAN meeting abstract is below:
A Multicenter Phase II Open-Label Trial of L-Carnitine and Valproic Acid in Infants with Spinal Muscular Atrophy Type I. Kathryn Swoboda, Salt Lake City, UT, Kristin Krosschell, Chicago, IL, Thomas Crawford, Baltimore, MD, John Kissel, Columbus, OH, Charles Scott, Fort Washington, PA, Mary Schroth, Madison, WI, Gyula Acsadi, Hartford, CT, Priya Kishnani, Durham, NC, Jurgen-Chrystoph von Kleist-Retzow, Cologne, Germany, Guy D’Anjou, Montreal, QC, Canada, Edward Smith, Durham, NC, Bakri Elsheikh, Columbus, OH, Louise Simard, Winnipeg, MB, Canada, Thomas Prior, Columbus, OH, Allen Tran, Salt Lake City, UT, Bernard LaSalle, Salt Lake City, UT, Sandra Reyna, Salt Lake City, UT. OBJECTIVE: Primary- to explore feasibility of recruitment/retention of type I infants, and assess safety and adverse events in infants receiving VPA and L-carnitine. Secondary- to evaluate exploratory endpoints for efficacy trials. BACKGROUND: Valproic acid (VPA) has been demonstrated in vitro in SMA patient-derived cell lines to increase expression of full-length SMN and to prolong survival and improve neuromuscular phenotype in SMA animal models. DESIGN/METHODS: Open label phase II multicenter trial design included 4 visits over 6 months, then 6-month telephone follow-up. Inclusion criteria: type I infants ages 2 weeks to 12 months with SMN deletion. Exclusion criteria: ventilator use > 12 hours, SMN enhancing medications within 30 days or illness within 2 weeks of entry. Exploratory outcomes: time to death and/or > 16 hours ventilator support (primary efficacy endpoint), Test of Infant Motor Performance Screening Inventory (TIMPSI), Type I Caregiver Questionnaire, maximum Ulnar Compound Muscle Action Potential Amplitude (CMAP), and SMN mRNA levels. Simple frequency and summary statistics used for most variables. Age/gender matching with subjects in the Utah database for primary efficacy outcome. RESULTS: Thirty-eight infants were enrolled across 8 sites in the US, Canada and Germany. Mean age 5.5 months, gender 47% female. A significant proportion already had a feeding tube (11/38) or respiratory impairment (19/38). CMAP (0.68 mV) and TIMPSI (30.45) scores indicated significant baseline denervation/weakness. 261 adverse events included 83 SAEs (77% severe/life-threatening) and 13/38 subjects (34.2%) died during the study. Time to death/ventilator dependence occurred in 37%. CONCLUSIONS: Recruitment/retention of sufficient numbers of type I infants for a multicenter clinical trial is feasible. A promising endpoint for measuring disease progression is time to death and/or more than 16 hours/day ventilator support per day. This study failed to support efficacy for VPA/L-carnitine in type I infants when compared to cohort of age and gender matched natural history subjects. Supported by: Families of SMA (Elk Grove Village, IL, USA). University of Utah Center for Clinical and Translational Sciences CTSA UL1RR025764 and site CTSA at University of Utah, Johns Hopkins, Ohio State, University of Wisconsin, Wayne State University and Duke Medical Center.

Families of SMA has invested $6 million to the development of clinical trial infrastructure, including testing protocols and clinical trial site readiness through Project Cure SMA. Project Cure SMA conducted five clinical trials in different SMA patient populations, spanning from infants to adults. This work helped paved the way for the current major investment in SMA clinical trials by NIH and companies testing novel drugs, such as Isis Pharmaceuticals. For instance, the NINDS has created The Network for Excellence in Neuroscience Clinical Trials, abbreviated NeuroNext. NeuroNEXT is twenty-five site national clinical trial network created by NINDS of the NIH to test promising new therapies for both pediatric and adult patients with neurological diseases. Their first study is a SMA Biomarker Study in infants with SMA Type I to be conducted at 15 NeuroNEXT Network sites around the United States. Stephen Kolb, MD, PhD of Ohio State University is the Protocol Principal Investigator for the study.
BAYADA Pediatrics is proud to support

Families of Spinal Muscular Atrophy

With a broad range of services and a team of pediatric professionals who are committed to keeping children with SMA safe at home, BAYADA provides:

- Experienced nurses with specialized training in caring for children with SMA
- Clinical support 24 hours, 7 days
- Ongoing communication with each child’s parents, physicians, and school
- A care team which includes a registered nurse clinical manager to oversee all levels of care
- Specialized, one-on-one care at school, including field trips, and during transportation
- A variety of payment options, including Medicaid and most insurances

To find a BAYADA Pediatrics office near you, call 800-305-3000 | www.bayada.com


Disclaimer: The acceptance of Vendors and Sponsors does not constitute or imply endorsement by Families of SMA of any company, product or service. FSMA accepts no responsibility for any claims made by any outside party.
BAYADA Pediatrics is proud to renew its partnership with FSMA, as they support our dedication to helping families affected by SMA and to fund research leading to a treatment, and eventually, a cure.

Through this initiative, BAYADA Pediatrics is looking forward to continuing to work with FSMA to strengthen their clinical expertise in caring for children with SMA.

Throughout the past year, in the true spirit of providing community service where they live and work, BAYADA Pediatrics offices came together to raise funds and awareness for FSMA. BAYADA staff and their families, donned in their signature BAYADA red, were honored to be able to have a physical presence at multiple FSMA Walk-n-Roll events held in California, Colorado, Delaware, New England, Minnesota, New Jersey, New York, North Carolina, and Pennsylvania.

The partnership allowed BAYADA clinicians to participate in the FSMA national conference. In addition, BAYADA Pediatrics proudly played host to an FSMA delegation who travelled from Illinois to the Philadelphia area to learn more about their exceptional home health care services. The delegation had the opportunity to tour a Simulation Lab, which uses computer-generated, realistic mannequins to train home health care nurses. They also had the opportunity to witness home health care nurses providing care during home visits with pediatric clients.

This year, BAYADA President and Founder Mark Baiada has again agreed to generously support the Walk-n-Roll program as a national sponsor.

“This year, we hope to expand our participation and have even more BAYADA offices become involved in local FSMA events,” shared BAYADA Pediatric Practice Leader Karen Buttler. “It’s a great opportunity to demonstrate The BAYADA Way in action, to network with other families affected by SMA, and to educate families and health care professionals about the high quality home health care services BAYADA Pediatrics can provide for children with SMA.”

Dr. Al Freedman, psychologist, member of the FSMA Medical Advisory Board, and father of Jack, an 18-year old client with SMA, expressed his appreciation to everyone who has helped support FSMA and who has made a difference in the lives of children and families affected by SMA.

“Everybody who cares for families like ours needs to remember that we’re scared,” said Al. “We’re scared every day. And we need help. Every time someone from BAYADA helps us, they are making us feel supported, and less scared, and more hopeful that we can take care of Jack responsibly and that we can give him a life that is meaningful.”

To learn more about BAYADA Pediatrics, please contact BAYADA Community Liaison Amy Welde awelde@bayada.com or call 609-747-8617.

For the latest news visit the FSMA website: www.curesma.org
Results from the Phase II Valproic Acid in Ambulant Adults With Spinal Muscular Atrophy (VALIANT SMA) clinical trial led by Dr. John Kissel at The Ohio State University have been published in the journal Muscle & Nerve. This study was funded by Families of SMA. The study was a randomized, prospective, placebo-controlled clinical trial designed to test the efficacy and safety of the combined treatment of valproic acid in ambulatory adults with SMA Type III. Overall results showed that VPA was not effective in improving strength or function in this population. The trial did demonstrate that the outcomes used in this study were reliable and could be employed in future trials in adults with SMA. The trial was registered at clinicaltrials.gov with identifier: NCT00481013.

Much basic and clinical evidence has suggested that valproic acid (VPA) might benefit patients with Spinal Muscular Atrophy (SMA), including an open label trial of VPA in ambulatory SMA adults, showing VPA markedly improved strength in this population. The clinical trial network Project Cure SMA conducted the 12-month, double blind clinical trial of VPA in ambulatory adults with SMA to test this idea.

The study involved 33 genetically proven adult SMA Type III subjects age 20-55 years. Subjects underwent baseline assessments and then were randomized to receive VPA or placebo. At six months, patients were switched to the other group in a cross over design. The primary outcome was the six-month change in muscle strength. There were multiple secondary outcomes, including changes in hand held dynamometry, ulnar compound muscle action potential amplitudes (CMAP) assessing motor neuron function, timed function tests of stair-climbing and the six-minute walk test, pulmonary function, muscle mass, a quality of life scale, and an SMA modified Functional Rating Scale.

Thirty subjects completed the study and were analyzed. VPA was well tolerated and compliance was good. There was no significant change in any outcome at six or 12 months. Thus, VPA was not effective in improving strength or function in ambulatory SMA adults, which is consistent with the results from the two CARNIVAL Clinical Trials assessing efficacy of valproic acid in SMA Type II/III and Type I patients. The outcomes used in this study were shown to be feasible and reliable, and can be employed in future trials in adults with SMA.

The trial has afforded investigators the opportunity not only to assess the efficacy and safety of this drug treatment, but also to test a number of new clinical trial outcome measures which are essential to conducting successful SMA trials, particularly in adults with SMA. As Dr. Kissel noted, “This is one of only a handful of clinical trials in SMA that has focused exclusively on the adult SMA population, a group that has received relatively little attention in the past. We expect, however, that the data we generate will also prove useful in devising and testing therapies for patients with other types of SMA. We are deeply appreciative of our patients who took the time to participate in the study, as we had patients from all over the country.”

Families of SMA has invested $6 million for the development of clinical trial infrastructure, including testing protocols and clinical trial site readiness. FSMA funding to Ohio State University has included three clinical trials and the establishment of a care clinic geared towards infants with SMA Type I.

This funding paved the way for the current major investment in SMA clinical trials by NIH. The NINDS has created The Network for Excellence in Neuroscience Clinical Trials, abbreviated NeuroNEXT. NeuroNEXT is a twenty-five site national clinical trial network created by NINDS of the NIH to test promising new therapies for both pediatric and adult patients with neurological diseases. Their first study is a SMA Biomarker Study in infants with SMA to be conducted at 15 NeuroNEXT Network sites around the United States. Stephen Kolb, MD, PhD of Ohio State University is the Protocol Principal Investigator for the study.
Trophos Announces Positive Interim Safety Data in Pivotal Study of Olesoxime in Spinal Muscular Atrophy

The Data Monitoring Committee (DMC) has reviewed the study data at one year and recommended to continue the study as planned through year two based on safety data and pre-defined stopping criteria.

Trophos SA, a clinical stage pharmaceutical company developing innovative therapeutics from discovery to clinical validation for under-served medical needs in neurology and cardiology, announces today the completion of the interim analysis of the pivotal efficacy study of olesoxime in the rare neurodegenerative condition Spinal Muscular Atrophy (SMA).

The independent Data Monitoring Committee (DMC) has reviewed the treatment effect at one year on the primary outcome measure of efficacy, change in motor function using the MFM scale, together with the latest safety report including electrocardiogram traces, periodic laboratory findings, haemostatic parameters and serious adverse events listings for all participants. Based on the trial stopping criteria as defined in the protocol as well as no safety concerns related to olesoxime treatment, their recommendation is to continue the study as planned.

An interim analysis of efficacy, as included in the study protocol, has been conducted after all participants have been treated for one year. Over 160 patients have been recruited into the trial between October 2010 and September 2011. Following the recommendations of the DMC, the study will continue until all participants have been treated for two years with the last patient out scheduled for September 2013. Top line results are expected by the end of 2013.

“The approval from the DMC to continue the trial based on the interim analysis for this innovative treatment of SMA is perfect news at this stage. We confirmed the good safety of olesoxime treatment and we look forward to completing the two year treatment period to report efficacy in these SMA patients,” said Rebecca Pruss, chief scientific officer at Trophos. “SMA is a progressive and disabling neuromuscular disease. Treatments are desperately needed that slow down or prevent the loss of neuromuscular function in SMA patients. This study of olesoxime has been conducted successfully so far due to the enormous commitment of patients and clinicians to find a treatment for SMA. We believe the results due in fourth quarter 2013 could be an historic moment in the development of treatments for SMA.”

**Trial design and end-points:**
The study is a 24-month randomized, parallel group, double-blind, placebo controlled trial comparing olesoxime against placebo in non-ambulant type II and type III SMA patients aged from 3 to 25 years old. Olesoxime is administered at the dose of 10 mg/kg/day using a specially developed liquid formulation; patients were randomized to receive olesoxime in a 2:1 ratio versus placebo. The primary end-point of the study is the change from baseline in the Motor Function Measure (MFM) functional scale. Secondary endpoints include the Hammersmith Functional Motor Scale and electromyography (CMAP - Compound Muscle Action Potential - and MUNE - Motor Unit Number) as well as measure of safety, tolerance and quality of life. The study is being conducted in 22 centers in France, Italy, Germany, UK, Belgium, the Netherlands and Poland by a consortium of prominent European clinical investigators, all of whom have extensive experience conducting and collaborating in SMA clinical trials.

See [www.clinicaltrials.gov](http://www.clinicaltrials.gov) for more information on SMA clinical trials.
Hope is what gets you out of bed on the day of a big game.
It is what pushes you to be different or unique.
The hope that someone will love and appreciate you for who you are.
When you have a crush and you just don’t know what to say hope is your inspiration.
Hope will get you through the days that want to push you down on your knees.
Hope could be the only pill you’ll ever truly need to take.
Hope is a drug of its own.
The good kind.

**Hope can push you to do incredible things.**
With hope you’ll be lifting boulders in no time.
Hope feeds your soul.
Not only do you need a well balanced diet, but so does your soul.
Without it we would be nothing.
I have a friend that once said he felt the weight of the world was on his shoulders.
That life was just handing it to him that month.
I told him hand life something back.

**Hope**

H = Hold
O = On to
P = Positive
E = Encouragement