

# Spinal Muscular Atrophy Clinical Research Center

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COLUMBIA  
UNIVERSITY SMA  
CENTER

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## From the Desk of the Director: Darryl De Vivo, MD

The academic year 2008-2009 has been challenging; and that is an understatement!!! The economic downturn has forced all of us to tighten our belts and to count every penny. Extramural funding has been more difficult to secure and philanthropy has suffered. Fortunately, the SMA Foundation has remained true to its mission and continued to support the activities of the SMA Clinical Research Center. And, the ultimate goal of the Center remains the same; namely, finding a cure for SMA and other neuromuscular diseases that threaten our children. Our SMA team at Columbia remains dedicated to this vision; and you, the patients and their families, remain committed as key partners in this noble endeavor.

Admittedly, there have been some gains and some losses, but the overall momentum continues in a forward direction. The NINDS ultimately succeeded in recruiting Dr. Petra Kaufmann. Certainly our institutional loss is the NINDS' gain. But, she will remain engaged in the goals of the SMA Community from her new position at the National Institutes of Health. And, she will continue to collaborate with us at Columbia as we complete many ongoing clinical research projects. And, we also are fortunate to have others who are ready and able to step in and fill Dr. Kaufmann's responsibilities. Dr. Douglas Sproule will assume a greater leadership role and he will pursue several very important topics such as proper nutrition for patients with neuromuscular diseases, a better understanding of body composition of such patients, and the role of exercise in the management of patients with SMA. Dr. Romana Kulikova also will continue her leadership role in our multi-disciplinary clinic and the rest of the SMA team remains intact and committed.

This year we hope to complete an analysis of the 12-month and the 24-month natural history studies of SMA. This study is essential as a critical bench mark for the Clinical Trials Study that will surely follow in the coming months and years. We, also, are evaluating the 6-minute walk test, (6 MWT) as a critical measure of fatigue that complicates the life of patients with SMA; and we are pursuing some therapeutic interventions that may mitigate this complicating and compromising

*(continued on page 2...)*

## IN THIS ISSUE

From the Desk of Dr. De Vivo	1	News about SMA and Nutrition	3	Patients Artwork Exhibited	5
Dr. Kaufmann departing CUMC	2	Hope for expanded clinical trials	4	Upcoming Events	6
About the SMA Treatment Acceleration Act	3	An update on SMA Day 2009	4		

*(Director...continued from page 1)*

symptom. Our preliminary studies with the 6-MWT have shown us that ambulatory patients with SMA fatigue during this outcome measure. Their performance in the sixth-minute is 17% less than their performance in the first minute, a highly significant difference that highlights the presence of fatigue as a complicating factor in denervating diseases like SMA. Now that we have this effective outcome

measure to assess fatigue, we can explore pharmacological agents that might counteract this symptom and increase the patient's stamina and endurance for important activities of daily living.

We truly hope and expect that the coming year 2009-2010 will be even more exciting and productive as we continue to pursue our ultimate goal. Our Second Annual SMA Day on July 25, 2009 was a great success. The presentations by

Drs. Monani, Krainer, Vitale and Sproule were truly exciting. These facts made it abundantly clear that today's clinical management of patients with SMA is much better than yesterday's management and tomorrow's treatment will even be better. Thank you again for your ongoing support of the many activities being conducted in the SMA Clinical Research Center at the Columbia University Medical Center.

Darryl C. De Vivo, M.D.

## Goodbye from Petra Kaufmann

By Petra Kaufmann, MD

As most of you know, I will be taking a leave of absence from Columbia University Medical Center starting in September 2009 to assume a new position as Director of Clinical Research at the National Institute for Neurological Disorders and Stroke, which is part of the National Institutes of Health in Bethesda, Maryland.

Deciding to take this new position was very difficult for me, as the move means not only that I will be leaving one of the leading academic medical in-

stitutions in the country, but also the place where I "grew up" professionally. However, I believe that my new role offers a unique opportunity to have a greater national impact on neurological clinical research, including trials of new therapeutic agents.

Leaving my patients in the multidisciplinary Pediatric Neuromuscular Clinical Research Center is probably the hardest aspect of my decision to leave Columbia, even though I know that the clinical care and re-

search program will continue to be exemplary under the leadership of Dr. Darryl De Vivo with the assistance of his colleagues Dr. Romana Kulikova and Dr. Douglas Sproule. Furthermore, in order to assure the smoothest possible transition, I will continue to advise ongoing clinical research projects.

It has been a privilege participating in your care and working with you, and I thank you for the opportunity.

## NYC Half Marathon Team Raises Money for SMA Research



The New York City Chapter of Families of SMA hosted their 2nd Annual Half Marathon

Fundraiser at the New York City Half Marathon this past July. The team, named in honor of Greyson Erwin, raised approximately \$70,000 in support of SMA Research, to be split between the Families of SMA and Columbia SMA Clinical Research Center. Corporate support was provided by

several corporations including Phillips-Van Heusen. All proceeds were raised in loving memory of Max Rubenstein. Congratulations!

*Left: Picture of the team for Greyson at the New York City Half Marathon. Right: Greyson Erwin*



## An Introduction to the SMA Treatment Acceleration Act

By Mackensie Yore, Research Management Associate, SMA Foundation

### *What is the SMA Treatment Acceleration Act?*

The SMA Treatment Acceleration Act is a bipartisan piece of legislation, which has the goals of providing critical support for SMA clinical trial sites and ensuring that the SMA community is prepared for clinical trials.

### *Why is it needed?*

Comprehensive SMA patient care and clinical research involves a multidisciplinary team, which can include pediatric neurologists, pulmonologists, orthopedists, physical therapists, occupational therapists, nutrition experts, and others. Members of the healthcare team are often not reimbursed by insurance for the counseling and oversight they provide for clinical trials. More importantly, clinical research is expensive, and clinics need help from the major funding sources of medical re-

search, such as the NIH, to build and support clinics that can run SMA clinical trials.

### *What are its provisions?*

The SMA Treatment Acceleration Act will: 1) Provide funding to support SMA clinical and research teams, 2) Enhance the SMA Patient Registry, 3) Provide federal coordination for SMA research and therapeutics development, 4) Improve public and professional education and awareness regarding SMA.

### *What can we do to assist with its passage?*

It's a community effort! You can help get the SMA Treatment Acceleration Act passed by writing letters to, calling, or visiting your government representatives to encourage them to support the bill. The more cosponsors a bill has, the more likely it is to be considered important to the country.

### *Where can I find additional resources?*

You can find electronic versions of sample letters to Congressional representatives at the following website: [www.smafoundation.org](http://www.smafoundation.org), [www.mdaua.org](http://www.mdaua.org) (see their capwiz program), [www.fightsma.org](http://www.fightsma.org), [www.fsma.org](http://www.fsma.org).

In addition, you can contact: Families of SMA Greater New York Chapter  
Rockville Center, NY  
(516) 214-0348  
[greaterny@fsma.org](mailto:greaterny@fsma.org)  
Contact: Debbie Cuevas

Finally, contact our Government Relations experts for help.

Donna Mullins:  
[dmullins@wswdc.com](mailto:dmullins@wswdc.com)  
Laura Lay: [llay@wswdc.com](mailto:llay@wswdc.com)

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## Emerging Research on Nutrition and SMA

By Douglas Sproule, MD

How much should my child weigh? Is he or she getting sufficient calories? Too many? These important questions, frequently vocalized by parents, have, unfortunately, for a long time remained unaddressed by clinical research. Thankfully, this is starting to change.

This past fall, Messina and colleagues published a survey of Italian SMA type 2 patients,

reporting a high incidence of low weight and swallowing dysfunction. The authors speculated that this group of children (and adults) is at high risk for malnutrition. We recently published a study of body composition among children with SMA types 2 and 3. In contrast, we observed a high rate of obesity, with most children having greater than average amounts of fat compared to children without SMA.

How do we make sense of these seemingly contradictory observations? For some children, particularly those with significant swallowing difficulties, maintaining sufficient calories may be a challenge. For many others, however, there is a risk of consuming too many calories, as they are less active, and thus require fewer calories, than children

*(continued on page 6...)*

## Computer Vision to Speed Clinical Trials

By Nancy Ott, Publications Specialist, NREC

In Spinal Muscular Atrophy (SMA), motor neurons gradually lose their ability to carry messages from the spinal cord to the muscles. Although this degenerative disease doesn't yet have a cure, several promising therapies are being developed. However, the high cost of clinical trials is a barrier to their approval by the FDA. Babies in clinical trials for SMA therapies must be admitted to a hospital or monitored at home by a clinician, both of which are expensive.

Enter Columbia University's Petra Kaufmann (of the SMA Clinical Research Center) and NREC's (National Robotics Engineering Center of Carnegie Mellon) Dave LaRose. They are investigating whether intelligent vision systems can be used in clinical trials for SMA. Because it affects movement, SMA is a good candidate for computer vision. Babies would be observed in their homes during clinical trials by a vision system that monitors their breathing and other muscular movements. The data it gath-

ers would show what effect a therapy has on muscle tone, respiration, and other markers for SMA. This home-based monitoring would be much less costly than hospital admissions or clinician visits, speeding the approval of new therapies. It would also be more comfortable and less disruptive for patients and their families. And the unblinking eye of the camera would record every move that a patient made, giving researchers insight into the disease's progression.

The pilot study is looking at whether respiration data gathered by a computer vision system correlates with respiration data recorded by body sensors and clinical observation. Patients in the study wear a sensor vest (or life shirt) that records their respiration. At the same time, they are monitored in their cribs by stereo cameras. The vision system tracks the three-dimensional movement of markers on the life shirt. Additionally, it tracks the movement of the patient's wrists and ankles, which also



*A patient under observation at home with the pilot version of the vision system.*

can indicate the progress of the disease. The cameras use infrared light that's invisible to humans, allowing the babies to be monitored day and night. A laptop computer stores the image data for processing. The system correlates the computer vision estimates of the patient's respiration are with the respiration data from the life shirt. It also correlates limb motion with observations gathered by human clinicians. If the vision system data and life shirt data correlate well, the computer vision system can be further developed and adopted for use in clinical trials.

## Update on SMA Day 2009

By Jackie Montes

To kick off SMA Awareness month in August, we held our 2<sup>nd</sup> annual SMA family day on 7/25/09. It was a successful event with more than 40 people in attendance. Research lectures included Dr. Adrian Krainer, of Cold Spring Harbor

Laboratory, who spoke about antisense oligonucleotides as a treatment for SMA, and Dr. Monani, of Columbia University's Motor Neuron Center, about preclinical research, as well as clinical talks about scoliosis and nutrition manage-

ment. Cynthia Joyce, of the SMA Foundation, spoke about the Treatment Acceleration Act. While the adults sat in on the lectures the children had a blast with the help of Child Life Services from NYPH and volunteers.

## Patients' Artwork Inspires at International Exhibition

By Petra Kaufmann, MD, MSc

In March 2009, HeartSCAPE: "Creative Arts Pathway to Expression" for Children with Neuromuscular Disorders: An International Exhibition, was held in Athens, Greece with the goal to develop guidelines and a forthcoming consensus statement that will inform policy makers, support caregivers, and improve the lives of people living with pediatric neuromuscular disease. The conference focused on Spinal Muscular Atrophy Type 1 (SMA 1) and Duchenne Muscular Dystrophy (DMD), but much of what was discussed would be beneficial to people with other neuromuscular disorders as well. Three continents and seven different countries were represented, which allowed participants to share their specific native approaches to caring for someone with a neuromuscular disease.

The group worked on developing recommendations on how caregivers and members of society can best support people along their journey with neuromuscular disease, starting at the time of diagnosis. The meeting included a special evening in a cultural center in Athens to raise awareness and ensure that the voice of people with neuromuscular disease is heard. People with neuromuscular disease, conference participants, politicians, ambassadors from several countries, a Greek Bishop, patient advocates, and funding organizations gathered to share their

perspective. The event was enriched by a pianist playing a beautiful piece of music he had composed, as well as a very special international exhibit that displayed artwork made by people with neuromuscular disease and their siblings, including children from New York City.

Contributions to the exhibit from the Columbia University Pediatric Neuromuscular Center included the following: Owen, living with SMA 1, created a story and pictures expressing his love of space and dinosaurs; Mercedes, painted a picture of two trees and butterflies flying from one to the other, which she made thinking of her brother who has DMD; Adelina, living with muscular dystrophy, created a lively explosion of colors; and Noah, living with DMD, painted a vibrant heart.



*Dr. Kaufmann with the artwork from Columbia University Pediatric Neuromuscular Center on display in Greece.*

The pieces that were exhibited in Athens were created at a special event for families that was held at the Children's Hospital of New York on January 30, 2009. Several family members and children attended the event, as well as several volunteers, and two art therapists from the Child Life and Creative Arts Therapy Department at Mount Sinai who assisted with the event. Children were invited to express themselves through the creation of visual images, poetry, and other creative modalities and some of the resulting artwork was sent to the display in Athens and some of it was kept to be put on display at the Pediatric Neuromuscular Center at Columbia University.

The exhibit displaying the artwork was well received and affected, in some way, everyone in attendance. It certainly raised awareness and exemplified that children and adolescents living with neuromuscular disease have the same interests as everyone else their age – dinosaurs, colors, butterflies, and hearts. The artwork will become part of the permanent art collection of the Muscular Dystrophy Association of Greece, MDA Hellas, a charity organization that raises awareness and funds for neuromuscular diseases.

We thank everyone who made this event possible, namely

*(continued on page 6...)*

**(Nutrition...** continued from page 3)

without SMA. This can lead to obesity. This concern appears to be most acute among higher functioning children

with SMA type 2. What these observations highlight is the vital role of dedicated, expert nutritionist as part of routine, multidisciplinary care in SMA, and the intense need for fur-

ther study. We intend to continue our research on nutrition in SMA and expect to report more on this topic very soon!

**(Artwork...** continued from page 5)

The New York Life Foundation Wintergarden Programming Series who sponsored, in part, the art event held at Columbia

University and the Shuler Family who donated funds for framing the artwork. We also acknowledge the MDA, the SMA Foundation, the Niarchos Foundation and the team from John Hopkins who made this

all possible. We especially express heartfelt gratitude to the families and artists for generously contributing their time and work to this special exhibit.

## Upcoming Events

Kiley & Dylan's Sweet Dreams Foundation and Sophia's Cure Foundation will be hosting the 3rd Annual Gala to Cure SMA. The Gala will be held at Bridgewaters at the South Street Seaport on Thursday, November 12, 2009 from 6:30pm - 10:30pm. There will be Silent Auction, Live Auction, 50/50 Raffles and an Awards Ceremony honoring some special people within our SMA community in New York

Tickets are \$150 per person.

RSVP by 10/12/2009 to:  
 Debbie Cuevas  
 516-214-0348  
 Cure SMA NY Gala  
 47 Melrose Avenue  
 East Northport, NY 11731

SMA Clinical Research Center  
 Columbia University  
 180 Ft. Washington Avenue, 5th Floor  
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