



# Spinal Muscular Atrophy

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## From the Desk of Dr. Darras:

The current stance of SMA knowledge and research can be described by two Napoleon Hill quotes:

*"Do not wait; the time will never be 'just right'. Start where you stand, and work with whatever tools you may have at your command, and better tools will be found as you go along."*

This is where we are at with our knowledge of SMA and the research happening in the field. We do not yet have the perfect tools to measure whether a medication works or not. But to wait for those ideal tools will prolong the time before a cure is found. Thus, scientists and researchers are pursuing many different avenues of work in SMA simultaneously. Some of this work is looking at the tools themselves and seeing if we can improve them. Currently studies happening in Boston to this effect are our EIM study and AmSMART study (please see page 3 for more details about each of these studies). Simultaneously, we are also working on medication studies, both through our collaborative efforts with the national NPTUNE project and our own PNCR studies (again please see page 3 for more details). In addition, millions of dollars are invested in basic science research that help us further develop those tools that are needed. All of these studies are equally important to move ahead the field of SMA research so we hope you will embark on this journey with us.

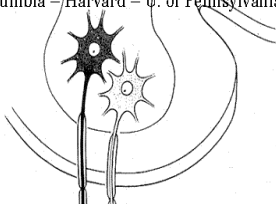
*"Effort only fully releases its reward after a person refuses to quit"*

Over the last 3 years, we have been committed to furthering SMA research as well as improving the clinical care we provide to families. We hope the efforts we have made with our new website, our Boston conference, these newsletters, and our various SMA research studies have been helpful to you and your family.

However, our work in SMA is not done yet. We are committed to working closely with you and your families to provide the best clinical care that we can and further our knowledge of SMA through research. As we hope this is a collaborative effort, please do not hesitate to let us know if there are things that would be helpful, as we would like to explore those opportunities with you.

## PNCR - Network for SMA

North-Eastern Clinical Trials Consortium  
Columbia - Harvard - U. of Pennsylvania



## Meet the SMA Team:

### Neurology:

Dr. Basil T. Darras  
Director, NM Program,  
Principal Investigator

Dr. Peter Kang  
Attending Neurologist,  
EMG Specialist

Erica Sanborn  
Genetic Counselor,  
Clinical Coordinator

Shuei Koza  
Social Worker

Lindsay Hansen  
Administrative Assistant

### Orthopedics:

Dr. Frederic Shapiro  
Attending Physician

Physical Therapy:  
Janet Quigley  
Clinical Evaluator

Sue Riley  
Clinical Evaluator



## Families of SMA 2008 Conference

As many of you may have heard, the 2008 Families of SMA Conference is coming right here to Boston.

This is an amazing 2.5 day conference that provides the most up-to-date information about all aspects of SMA, provides an opportunity to

meet other families living with SMA, and allows children to meet others like them.

The exact conference date at this time still remains open, however it is anticipated to be held near the end of the month of June 2008.

Families of SMA can always use additional hands to help pull together a conference of this level.

If you are interested in helping in any aspect of the conference planning please contact Jim Gaudreau at: [jgaudreau@green-co.com](mailto:jgaudreau@green-co.com)

## Boston SMA Day

We are pleased to announce that our second Boston SMA Day was a great success. The conference was attended by: 26 families, 16 children, and 38 researchers from the Boston area.

Families seemed pleased with our new brighter venue and we hope to be able to hold the event there in future years.

As you read above, the next Families of SMA conference is coming to Boston in 2008. This is really exciting news.

As both conferences take a tremendous amount of work, we are concerned about duplicating talks, efforts, and resources. As such, we are currently planning to delay our next Boston SMA Day until 2009.

In place of Boston SMA day, we hope to offer a social event in the fall of 2008.

As we are always looking for new topics for our Boston SMA Day to make the conference as helpful as possible, please contact Erica Sanborn at 617-355-2752 or [erica.sanborn@childrens.harvard.edu](mailto:erica.sanborn@childrens.harvard.edu) if you have ideas for any talks or discussions that would be of interest for future years.

Kids' Corner Answers  
(questions on page 4):

- 1) A carpet
- 2) Footsteps
- 3) A glove

## Information Cards

The most common question we are asked when providing families with a new diagnosis is "why have I never heard of this?"

Unfortunately SMA is not a rare condition in the world of inherited conditions and thus one of our major goals is to help raise awareness about SMA in the community.

In an effort to do this, we have created basic information cards (postcard size) about SMA. These cards can be customized easily with your/your child's picture.

Our hope is that these cards will provide parents an easy way to explain to others they interact with about SMA. Hopefully this will not only raise awareness but decrease some

ignorance we have heard that many families encounter.

If you are interested in having some of these information cards, please contact [erica.sanborn@childrens.harvard.edu](mailto:erica.sanborn@childrens.harvard.edu). If you have a particular picture you would like included for you/your child, please enclose a picture with your email.

## SMA Type I Guidebook

We are in the process of creating a guidebook specific to SMA type I, to help families navigate all of the difficult decisions that they encounter.

If you are a parent of a child with SMA type I, and have

thoughts about what would have been helpful for you to have in an educational tool at the time of diagnosis, please contact Erica Sanborn with those ideas.

Also we would like to make the guidebook as personal

as possible so if you have pictures of your child that we could include, or quotes or messages you have for other families new to your situation, please also contact Erica at 617-355-2752 or [erica.sanborn@childrens.harvard.edu](mailto:erica.sanborn@childrens.harvard.edu)

### **NPTUNE study:**

We are pleased to announce that we have finally gotten all the necessary approvals to run the NPTUNE study.

This study is intended to evaluate the best dose of sodium phenylbutyrate for patients with SMA. Here in Boston we are only studying

patients with type I SMA who are between the ages of 2-24 months, weighs at least 7kg, and does not use ventilation support for more than 16 hours per day.

For this Phase I study, only 30 patients are needed nationwide. We hope to move

through this phase of the research quickly which is great for SMA research in general, but less optimal for families that are interested in the research and can not participate. We will make every effort to accommodate interested participants.

### **PNCR study:**

In 2004, our hospital teamed up with Columbia University NY and Children's Hospital of Philadelphia to form the Pediatric Neuromuscular Clinical Research (PNCR) Network to further research on the east cost for SMA. We have been very successful at building our infrastructure and gathering important information through our natural history study.

We have been fortunate that 21 of our local families have donated their time to participate in this important study. While we originally planned to close enrollment this summer, we have decided to continue to enroll interested families in an abbreviated protocol to continue to learn about outcome measures important to SMA research.

As many of you know, we applied for federal funding in 10/06. We are saddened to report that we did not receive funding. However, we are addressing the concerns of the NIH and plan to reapply for funding this coming October. As always we will keep you posted on the status of these efforts.

### **Electrical Impedance Myography study:**

In addition to our various medication trials in various stages of development, we are also embarking on a new observational study using electrical impedance myography (EIM). EIM is a new method that we hope will prove effective at measuring nerve changes in children and adults with SMA in a reliable way.

If this hypothesis proves true, we plan to incorporate EIM as an outcome measure in future SMA clinical trials. This method is more appealing than many outcome measures currently used as it is non-invasive, painless, and yet reliable in other motor neuron conditions, like ALS.

This study involves evaluations at the hospital every six months but can be combined with other clinical or research visits. At this time we are now open for enrollment into this study. We are seeking 30 patients with SMA, all types, over age 4 and 30 individuals without SMA, also over age 4.

### **AmSMART study:**

We are pleased to announce that we have also begun a new Quality of Life study for children with SMA. Currently the tools used for measuring quality of life, do not provide great information for children with neuromuscular conditions.

Because of this a research group in Texas developed a Quality of Life tool specific for children with SMA.

We have been using this new tool in our PNCR natural history study in an effort to establish validity and reliability.

As we need more participants to establish this validity and reliability, we are seeking individuals with SMA ages 2-18 and their parents to participate in a Quality of Life study.

This study involves coming to the hospital 2 times, 4 weeks apart to complete questionnaires.

For children ages 2-4, only a parent will complete the 2 questionnaires.

For children 5-18, both the child and parent will complete the two questionnaires independently.

If you are interested in more information about any of these studies please contact:

**Erica Sanborn** at:

617-355-2752

or

Erica.sanborn@childrens.harvard.edu

## Upcoming Events:

### Walk-N-Roll:

Save the date for this year's Walk-N-Roll in Woburn. We've made plans for *Sunday, September 23rd* to be a beautiful sunny day. Please join us for a 2-mile stroll/roll around lovely Horn Pond and the Family Fun Festival following the walk. We have food, music, entertainment plus a raffle and silent auction tent filled with lots of exciting items- Red Sox Tickets, Weekend Vacations, Jewelry, a LoJack system, an iPod, and lots more.

Registration: 9am Walk 10am

### Golf Tournament:

Join us on *Monday, October 1st, 2007* for the FSMA Charity Golf Tournament in honor of William Johnson. The tournament is being held at the New England Country Club in Bellingham, MA, with an afternoon shotgun start, cart included. The format will be best ball and the day will include lunch prior to the start along with dinner after the tournament. The evening will be topped off with a silent and live auction with special guest and Emcee Susan Wornick from Channel 5 News. For more information and registration forms please contact heidik-johnson@comcast.net .

### Comedy Club:

Join us on *Thursday, November 8th, 2007* for the 4th Annual FSMA Comedy Night at Giggles Comedy Club/Prince Pizza located on route 1 southbound in Saugus, MA. Tickets are \$30 each and include a full night of comedy (3 comedians) and all you can eat pizza. Doors open at 6:30PM with pizza served from 7:00 to 8:00 PM. Comedians start at 8:00 PM. For more information or to buy tickets please contact Joanne Schamberg at joanne@yourpromosolution.com or Christine Boulter at christine.boulter@verizon.net.

*For more information regarding Families of SMA, please contact Jim Gaudreau, president of the New England Chapter. He can be reached at: jgaudreau@green-co.com*

Please keep visiting our website for the most up to date information about research and upcoming events. This newsletter is also posted on the website to make it easier to share with interested friends and family.

[www.childrenshospital.org/sma](http://www.childrenshospital.org/sma)



**Children's  
Hospital  
Boston**

## Just For Kids

Hey kids! How smart are you? Can you figure out these fun brainteasers? (Answers on page 2)

#1

What do you get when you cross an automobile with a household animal?

#2

The more of them you take, the more of them you leave behind. What are they?

#3

What has 4 fingers and 1 thumb but is not alive?

Riddles from:

<http://www.niehs.nih.gov/kids/rd1.htm>

*Word Fun...*

*Can you find these words that describe summer break?*

CAMP  
FRIENDS  
HOT  
NO SCHOOL  
PLAYGROUND  
POPSICLES  
SUMMER  
SUN  
SWIMMING  
VACATION

U S B K S C A F B D F I P Z H

S L S C W T Y R Y Z E P I O U

A V A H O G H I O T A T T B F

Y A S I C I S E L C I S P O P

S C A M P L D N K A F X L K G

B A E B U E S D V S A K D S N

E T W T R Q T S T B T H N Y I

K I S L C R S O Y H I P U A M

Y O A E E K E L F M O L O D M

K N U S N F X M T U N W R I I

D N K I R I R E M Y S P G L W

R R M O N A P O U U L O Y O S

H B Q H D F M P S Y S C A H L

E R L T E N O S C H O O L H M

W P E J K C J Y N H Y I P U S