

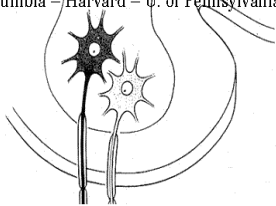


# Spinal Muscular Atrophy

## Inside this issue:

Meet the SMA Team	2
Meet the Extended SMA Team	2
Kid's Corner Answers	2
2009 FSMA Conference Summary	2
BioMarker Study	3
PNCr Study	3
EIM Study	3
Upcoming Events	3
Just for Kids	4

**PNCr - Network for SMA**  
 North-Eastern Clinical Trials Consortium  
 Columbia - Harvard - U. of Pennsylvania



## From the Desk of Dr. Darras:

The team here at CHB has been busy imagining and dreaming for as William Arthur Ward once said, "If you can imagine it, you can achieve it; if you can dream it, you can become it". We are working towards the goal that one day soon we will no longer have to dream for a cure because we will have one, we will no longer have to imagine life without SMA because it will be a reality.

To the families whose children have been living with SMA for many years now, this day may still seem far away and you may lose hope. To the families whose children have been newly diagnosed, you may feel a cure is too far away.

While it is easy to get caught up in feeling hopeless, I encourage you to continue to dream and imagine because we are doing the same. SMA was listed by NIH as the genetic disease mostly likely to find a cure. This is not a coincidence. Research on SMA is moving forward quickly because families like yours are continuously pushing, dreaming, imagining and working towards a cure.

I applaud the efforts you have exerted thus far; your support and willingness to be a part of clinical trials has made my job easier and I am confident that we are moving in the right direction.

## SMA Treatment Acceleration Act of 2009

Legislation to enhance federal funding supporting SMA research is currently collecting support from members of Congress and Senators.

A few of the areas legislation H.R. 2149/S. 1158, the "SMA Treatment Acceleration Act of 2009" would authorize federal funding for include:

- Unify clinical trials sites & establish a national clinical trials network for SMA
- Establish a Data Coordinating Center to provide expert assistance/advice to SMA clinical trials sites
- Expand/intensify federally supported research programs with respect to pre-clinical translational research related to SMA
- Promote collaborative research at the NIH

Currently 17 members of the House of Representatives and 4 Senators have signed onto the legislation. FSMA have generously provided a letter template for families to send to their representatives, which can be found at:

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

Please navigate to Support > Legislative > FSMA Legislative Agenda > SMA Treatment Acceleration Act.

## SMA Registry:

There is a lot of research happening throughout the world on SMA. However, no single study is capable of learning from every individual in the world with SMA.

As many of you know there is a registry for SMA, housed at

Indiana University.

According to their website: "The purpose of the International Spinal Muscular Atrophy Patient Registry is to provide a resource by which individuals and families affected by SMA

and researchers interested in studying SMA can come into contact with one another."

For more information about the registry or to participate please visit their website at: [smaregistry.iu.edu/](http://smaregistry.iu.edu/)

## Meet the SMA Team:

### Neurology:

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

Dr. Peter Kang  
Attending Neurologist,  
EMG Specialist

Hailyly Butler  
Clinical Coordinator

Jayson Caracciolo  
Clinical Coordinator

Lindsay Hansen  
Admin. Assistant

### Orthopedics:

Dr. Frederic Shapiro  
Attending Physician

### Physical Therapy:

Janet Quigley  
Clinical Evaluator  
Sue Riley  
Clinical Evaluator

### Pulmonary:

Lauren Perlman,  
Respiratory Care Continu-  
ing Care Coordinator

Dr. Robert Graham  
Intensivist / Home Care

### Nutrition:

Stacey Tarrant,  
Clinical Nutrition Specialist

## Meet the extended SMA Team:

While our Neuromuscular Clinic is multidisciplinary and includes many amazing people, we realized that SMA care extends into other disciplines. To alleviate confusion and enhance communication we have established an amazing team of people in various specialties that work closely with many families with SMA and are willing to work together with us to provide the best care as possible to patients. While these individuals are not directly part of our multi-disciplinary clinic, we hope that may change in the future to minimize the number of medical visits families have to attend.

We understand that some families affected by SMA have found health care providers, not on the list below, very helpful. We encourage families to stay with those providers as this list is not exhaustive. In addition to helping families directly, these individuals have offered amazing support to their colleagues working with patients with SMA.

For purposes of saving space, full titles and contact information is not available here. However, if you would like to get in touch with one of these specialists about making an appointment, please contact Hailyly Butler at Hailyly.Butler@childrens.harvard.edu and she can provide you with that information accordingly.

### Pulmonary:

Dr. Virginia Kharasch

### Social Work:

Shuei Kozu, LICSW

### Complex Care Service:

Dr. Laurie Glader

### Gastroenterology:

Dr. Laurie Fishman

### Anesthesiology:

Dr. Nevil Sethna  
Dr. Chuck Berde

### Surgery (non-orthopedic):

Dr. Craig Lillehei

### PACT Team:

Dr. Joanne Wolfe  
Dr. Tamara Vesel  
Dr. Christy Ullrich

Janet Duncan,  
Nurse Practitioner

Marsha Joselow,  
Social Worker

Kids' Corner Answers  
(questions on page 4):

- 1) A Stamp
- 2) "Wholesome"
- 3) 1, 2, 3

## 2009 FSMA Conference Summary

Several members of the Children's Hospital Boston SMA team were able to attend the 2009 FSMA conference and we were happy to see several families from the Boston area present.

Dr. Jill Jarecki of FSMA provided a research update and showed an excellent slide detailing the location of several

advances in the clinical trials pipeline. We fully encourage all families unable to attend the conference to look at Dr. Jarecki's presentation as well as others made available at:

<http://www.fsma.org/FSMACommunity/Conference/2009Presentations/>

The SMA team here at Children's Hospital Boston would be more than happy to discuss the latest developments presented at the conference. For more information, please contact:

Hailyly Butler  
hailyly.butler@childrens.  
harvard.edu  
857-218-4677

### Pilot Study of Biomarkers for SMA Study:

Firstly, thank you to all of the families that participated in this groundbreaking study with us. Over 100 individuals were recruited for the Biomarker study across 18 sites and all study visits and data collection were completed in just 4 months.

Currently, the SMA Foundation and the New England Research Institute are analyzing the data and samples collected. As soon as results are made available to the team at CHB, we will disperse that information to the SMA community.

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

**Haily Butler** at:  
857-218-4677

or

Haily.Butler@childrens.harvard.edu

## Research Update:

### PNCR study:

As many of you know, we teamed up with Columbia University NY, Children's Hospital of Philadelphia, and Rochester University a few years ago to form the Pediatric Neuromuscular Clinical Research (PNCR) Network. The goal of this group is to further research on the east coast for SMA.

We have been very successful at building our infrastructure

and gathering important information through our natural history study. The data collected as part of this study has been highly requested by many groups working on SMA and we are thrilled to be able to provide information that moves research forward.

As of July 2009 we have completed our final 36 month study visit. Nearly one hundred and twenty SMA affected indi-

viduals participated in the Natural History across the three sites. Having such a large amount of data has been very helpful across a multitude of research endeavors.

As we come ever closer to starting a therapeutic trial, we are excited to call upon the natural history study data, a unique and increasingly valuable source of information.

### Electrical Impedance Myography study:

In the summer of 2007 we began a study using a technology called 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.

Recruitment for the EIM study has come to a close and we

currently have over 20 controls and over 30 children with SMA participating in the study. Recently, we expanded the study to include children under 4 years of age and have had great success in recruiting younger children, which is vital to SMA research.

We will continue the data

collection for the EIM study for the next year and have already seen very promising results.

The study team is currently analyzing the first year of data and once this information is published, we will make it available to the SMA community.

## Upcoming Events:

FSMA New England Chapter - 12th Annual Golf Tournament  
**Monday, August 3 at 5:30pm**  
Bear Hill Country Club in Stoneham, MA

With plenty of activities for the entire family, the golf tournament includes a putting contest, a raffle, a silent auction, and a BBQ. Proceeds will benefit research and services for SMA. The requested donation to Families of SMA is \$50 per person. Please contact Paula Barrett for more information or tickets at [pmcbarrett@hotmail.com](mailto:pmcbarrett@hotmail.com). We hope to see you all there!

FSMA New England Chapter - Social Gathering / Chapter Meeting  
**Sunday, August 9 from 4-7pm**  
Heidi & Tripp Johnson's Home  
Medfield, MA

The perfect time to meet other local families in the SMA community and show off your best dish at this potluck style meeting. For more information or to RSVP (due by 08/05), please contact: [heidikjohnson@comcast.net](mailto:heidikjohnson@comcast.net).

The New England area SMA Community is very fortunate to have such a visible and available Families of SMA Chapter. The SMA Team at Children's Hospital Boston is in constant contact with the board of the New England Chapter and we would like to reinforce to the families we have contact with what a valuable resource this group is. Not only does the New England chapter do amazing fundraising work, but they are also very passionate about supporting families like their own. If you would like to speak with a member of the New England chapter please contact newengland@fmsa.org or simply mention your interest to Haily or Jayson at CHB, haily.butler@childrens.harvard.edu or

# Just For Kids

Hey kids! How smart are you? Can you figure out these fun brainteasers?

(Answers on page 2)

#1 What goes around the world but stays in a corner?

#2 What is it that, after you take away the whole, some still remains?

#3 What three numbers have the same answer when added together and multiplied together?

Word Fun...

Can you find these summer words?

Beach  
vacation  
waves  
sunshine  
camp  
friends  
sun  
flowers  
party

R	F	S	N	S	Y	U	E	A
R	H	D	O	U	W	W	O	O
F	E	N	I	H	S	N	U	S
L	S	E	T	O	H	C	E	C
O	S	I	A	P	B	V	A	D
W	I	R	C	E	A	M	P	E
E	W	F	A	W	P	R	T	T
R	I	C	V	C	A	E	T	I
S	H	S	A	I	U	A	E	Y

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  
300 Longwood Avenue—FE 1116  
Boston, MA 02115



**Children's  
Hospital  
Boston**