

# Synapse

Volume 13, Issue 4 Autumn, 2010

## Newsletter of the Spastic Paraplegia Foundation

### Loop for Life *(see article on page 7)*



Harvey and the hosts of Steel Horse,  
an Indiana television show



Despite the rain, Indianans came out in  
force for the Loop for Life



Bikers line up for the Loop

### President's Letter:



Mike Podanoffsky  
SPF President

I used to work at an odd place. For some reason they used to run out of paper coffee cups, so I started buying them in bulk. I would buy them, and everyone else would use them. One day, I was on vacation and the cups ran out. When I returned I found a note on my desk with the following notation: "you owe me \$8.50 plus gas for paper cups."

We have a Scientific Advisory Board (SAB) that rates the research proposals we receive. This year we received 21 proposals. Of those, an incredible 15 were ranked as worthy of funding. The problem we face is that we can only fund 4 of them.

The foundation plays a critical role in the research of motor neuron diseases. We fund innovative proposals and establish a track record for researchers. They are then able to use this success record to acquire further research funds from sources like the NIH (National Institutes of Health).

SPF research funds therefore "prime the pump." When the SAB writes their recommendations, it often evaluates the chances that this project has in acquiring continued funding. We are the seed funding researchers need to push through new ideas.

Just like paper cups, research is not free. Someone has been paying the tab. I am pretty sure many of you can afford \$10/month, and just that small amount would have funded these highly regarded research proposals. Please donate on our web site.

Earlier this summer we asked all of you to complete a research survey. If you don't recall getting an invite, or didn't complete the survey, you can always do so now (all links for this column are at the end of this article). Among the questions we ask are when you first noticed your symptoms, how many neurologists you saw before you received your diagnosis and are you happy with your current neurologist.

Didn't get a survey invite? [survey@sp-foundation.org](mailto:survey@sp-foundation.org)  
Survey form is at <http://sp-foundation.org/hsp-pls-symptomsurvey.html>

Donate <http://sp-foundation.org/donate.html>

Take care,

Mike Podanoffsky,  
SPF President

## 2010 SPF Research Grant Awards Are Announced

*Contributed by Mark Weber, SPF Board member and Chair of the Research Grant Advisory Committee*

On August 31st, the SPF Board voted to award funding for the following research grants.

**DONG-HUI CHEN, M.D., Ph.D.**, Research Assistant Professor, Department of Neurology, University of Washington, Seattle, WA, “Genome-wide detection of mutations in all genes to identify the cause of a new familial spastic paraplegia.” Two year grant is for \$73,194.

**SUSAN K. McCONNELL Ph.D.**, Susan B. Ford Professor of Humanities and Sciences, Department of Biological Sciences, Stanford University, Stanford, CA “Optimal transplantation strategies for the reconstruction of corticospinal circuitry in HSP and PLS.” Two year grant is for \$120,000.

**ROBERT D. NICHOLLS, D.Phil.**, Director, Birth Defects Laboratories, and Division of Genetics, Department of Pediatrics, Children’s Hospital of Pittsburgh and University of Pittsburgh Medical Center, Pittsburgh, PA, “A Proposal for a Roadmap that Links Different Spastic Paraplegias.” Two year grant is for \$119,779.

**TEEPU SIDDIQUE, M.D.**, Les Turner ALS Foundation/ Herbert C. Wenske Professor, Davee Department of Neurology and Clinical Neurosciences and Department of Cell and Molecular Biology, Director, Division of Neuromuscular Medicine, Feinberg School of Medicine, Northwestern University, Chicago, IL, “NTE-induced Upper Motor Neuron Degeneration in Primary Lateral Sclerosis.” Two year grant is for \$119,979.

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*The Spastic Paraplegia Foundation Inc. (SPF) is a national, not-for-profit, voluntary organization. It is the only organization in the Americas dedicated to Primary Lateral Sclerosis (PLS) and Hereditary Spastic Paraplegia (HSP).*

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 Jim Campbell..... Caring

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#### SPF Medical Advisor:

John K. Fink, M.D., University of Michigan

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# SPF News from the Board

Announcing the 2011 SPF Annual Conference:

## “ROCKETING TOWARD THE CURE THROUGH RESEARCH”

Houston, TX

DATES: JUNE 10-12, 2011



The hotel for our conference will be the Hilton Americas - Houston [www.americashouston.hilton.com](http://www.americashouston.hilton.com)  
1600 Lamar, Houston, Texas, 77010  
Ashton Hecker, Chair: 832-453-0394

**S**ave the dates now, and start thinking about making your travel plans. Plan to renew old friendships and make new ones. Learn, Laugh, Share, and Enjoy. Program information will be available by the first of the year both on the SPF website and in Winter *Synapse*.

Nearby attractions to enrich and extend your trip:

**1) Space Center Houston:** There are behind-the-scenes tours during weekdays at NASA'S Johnson Space Center. The Center has restaurants, theaters, and exhibit halls. Space Center Houston is a place where people can experience space -- from its dramatic history and exciting present to its compelling future. Space Center Houston is the only place on Earth that gives guests an out-of-this-world journey through human adventures into space. Space Center Houston's unparalleled exhibits, attractions, special presentations and hands-on activities tell the story of NASA's manned space

flight program. SCH is the only place in the world where visitors can see astronauts train for missions, touch a real moon rock, land a shuttle, and take a behind-the-scenes tour of NASA. At Space Center Houston, guests can understand the past, experience the present, and be a part of the future of the space Program. [www.spacecenter.org](http://www.spacecenter.org)

**2) Downtown Aquarium:** Part restaurant, part exhibit, the aquarium features tanks filled with exotic marine life. [www.downtownaquarium.com](http://www.downtownaquarium.com)

**3) Houston Zoo:** The 55 acre zoo has more than 1.6 million visitors every year to the animal filled attraction. [www.houstonzoo.org](http://www.houstonzoo.org)

**4) Moody Gardens:** The Aquarium, Rainforest, Discovery Pyramids of Moody Gardens on Galveston Island (45 minute drive) provide eye-opening experiences with science and nature.

**5) San Jacinto Battleground Historical Complex:** The San Jacinto Battleground marks the spot where Texas won its independence from Mexico. The park houses the towering San Jacinto Monument and Museum, with a 489 foot elevator ride to the top for a panoramic view. [www.sanjacinto-museum.org](http://www.sanjacinto-museum.org)

**6) Discover Houston Tours:** Explore downtown Houston during a weekday-led, leisurely paced walking tour. [www.discoverhoustontours.com](http://www.discoverhoustontours.com)

**7) Kemah Boardwalk:** Visit the 35 acre Kemah Boardwalk and enjoy dinner overlooking Galveston Bay and amusement rides. Watch sailboats from your table. [www.kemahboardwalk.com](http://www.kemahboardwalk.com)

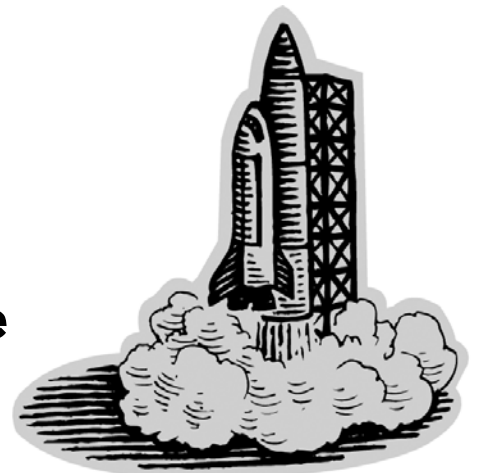


## 2011 SPF CONFERENCE

JUNE 10-12, 2011

HOUSTON, TX

Let your hopes and dreams for a cure take-off with us. We'll have a blast!





## SPF/NEALS Collaboration is already producing results

Contributed by Jim Campbell

In the Summer, 2010 issue of *Synapse* the SPF Board of Directors announced the formation of a collaborative effort between SPF and NEALS (Northeast ALS Consortium) to focus on HSP and PLS disorders. Dr. Nazem Atassi of Massachusetts General Hospital and Dr. Jinsy Andrews of the Hospital for Special Care were appointed co-chairs of a collaboration working group and have been meeting regularly by teleconference with SPF representatives Mike Podanoffsky, Mark Weber, Dr. John Fink and Jim Campbell. We are pleased to report the following accomplishments so far this year:

1. Creation of a list of NEALS institutions that welcome PLS and HSP patients – In answering a SPF community questionnaire earlier this year one quarter of you said you would like help in finding a physician. In response the NEALS consortium has polled their 92 member clinics for us to see who currently treats HSP and PLS patients and would welcome more. See the initial listing of 50 NEALS clinics on page 5. The clinics listed are spread across North America in twenty seven states and three Canadian provinces. The list identifies alphabetically by state, the facility, the city and whether they will see patients with PLS, HSP or both. Of the 50 clinics welcoming new PLS patients 36 clinics are also welcoming HSP patients. The list also includes the name of a physician who has agreed to be a point of contact and a phone number for making an appointment at the clinic. We expect this initial list to grow over the coming months with more NEALS facilities being listed as well as non-NEALS institutions. If you are not already being seen by a neurologist, we encourage you to contact the listed institution nearest you and arrange for an initial appointment.
2. Survey of HSP and PLS patients for symptoms and diagnosis history – To expand awareness and information on treating HSP and PLS at the member clinics, the NEALS physicians proposed and then authored a questionnaire for the SPF community. Over 200 of you responded anonymously to the survey questionnaire formatted and distributed by SPF email broadcast in June, 2010. NEALS physicians will analyze the results of this survey

looking for commonality and trends separately for PLS and HSP disorders. The objective of the survey is to determine the relative frequency and severity of symptoms of all patients then recommend treatment priorities. If you have not already responded we encourage you to do so on line at: <http://sp-foundation.org/hsp-pls-symptomsurvey.html>. As with the original survey your response will be anonymous.

3. Proposed establishing a NEALS/SPF registry of PLS and HSP patients. The largest existing registry at Northwestern University for PLS patients that contains blood and CSF samples has fewer than 100 participants. Over 300 PLS patients have signed up, track and compare progress and treatments with the Patients-Like-Me Website. In contrast, there is no national registry available for HSP. A registry is useful for the patient and their physician to have a record of their disorder and see their progress relative to others. Registries are more valuable to researchers if the data is entered by physicians and accompanied by data from blood and CSF samples. A NEALS/SPF registry would have two features that are especially appealing to SPF:

1. Data collection by physicians could be easily done nationwide at a number of facilities
2. NEALS has a strong policy of collaboration or sharing of samples among their member institutions for a wide range of studies relative to PLS and/or HSP. Early in our exploration of a NEALS/SPF registry for PLS and HSP patients we will be investigating what other registries already exist and how we can collaborate with them and avoid duplicating their efforts.

4. Established a date and agenda for the first face-to-face meeting of the collaboration working group. We will be meeting November 11<sup>th</sup> in Boston. We expect up to 12 working group members from NEALS facilities across the country as well as the SPF representatives to attend. After reviewing the science available for HSP and PLS, the meeting will focus on guidelines and ground rules for a patient registry as well as how to promote more proposals for the SPF Research Grant awards.

These first few months of collaboration with NEALS have already demonstrated some of the advantages to the SPF community of partnering with a nationwide medical network. We plan to report more progress in the coming issues of *Synapse*.

## SYNAPSE APPEAL

*Synapse* costs lots of money to print and mail, and we need your help to keep it going for another year. Please use the enclosed response envelope to make a donation. Every little bit helps.

## NEALS Hospitals & Clinics Treating PLS and HSP

<b>University of Arkansas</b> - Stacy Rudnicki Little Rock, AR 72205 501-686-5838 .....	PLS / HSP
<b>Barrow Neurological Institute</b> - Shafeeq Ladha Phoenix, AZ 85013 602-406-6262 .....	PLS / HSP
<b>Phoenix Neurological Associates</b> - David Saperstein Phoenix, AZ 85018 602-258-3354 .....	PLS / HSP
<b>Phoenix Neurological Associates</b> - Todd Levine Phoenix, AZ 85018 602-258-3354 .....	PLS / HSP
<b>University of British Columbia</b> - Hannah Briemberg Vancouver, BC, CAN V5Z 1M9 604-822-7929 .....	PLS
<b>University of California, Irvine</b> - Tahseen Mozaffar Orange, CA 92868 714-456-2332 .....	PLS / HSP
<b>Univ. of California, Davis, Medical Center</b> - Bjorn Oskarsson Sacramento, CA 95817 916-734-3588 .....	PLS / HSP
<b>Univ. of California, San Francisco</b> - Catherine Lomen-Hoerth, MD, PhD San Francisco, CA 94143 415-353-2122 .....	PLS
<b>University of California, Fresno</b> - Jeffrey Rosenfeld Fresno, CA 03721 559-264-9100 .....	PLS / HSP
<b>Hospital for Special Care</b> - Jinsy Andrews New Britain, CT 06053 860-827-4924 .....	PLS / HSP
<b>Mayo Clinic, Florida</b> - Kevin Boylan Jacksonville, FL 32224 904-953-6915 .....	PLS / HSP
<b>Emory</b> - Michael Benatar Atlanta, GA 30322 404-778-3444 .....	PLS / HSP
<b>University of Iowa Hospitals and Clinics</b> - Andrea Swenson Iowa City, IA 52242 319-356-4505 .....	PLS
<b>University of Chicago</b> - Kourosh Rezaia Chicago, IL 60637 773-702-9200 .....	PLS / HSP
<b>University of Chicago</b> - Raymond P. Roos Chicago, IL 60637 773-702-5659 .....	PLS / HSP
<b>Beth Israel Deaconess Medical Center</b> - Elizabeth Raynor Boston, MA 02215 617-667-8130 .....	PLS
<b>Brigham And Women's Hospital</b> - Anthony Amato Boston, MA 02215 617-732-8046 .....	PLS / HSP
<b>University of Massachusetts Medical Center</b> - Johnny Salameh Worcester, MA 01655 508-334-9040 .....	PLS / HSP
<b>Massachusetts General Hospital</b> - Nazem Atassi Boston, MA 02129 617-726-1728 .....	PLS / HSP
<b>Lahey Clinic</b> - Jim Russell Burlington, MA 01805 781-744-7273 .....	PLS / HSP
<b>Baystate Medical Center</b> - Carmel Armon Springfield, MA 01199 413-794-1150 .....	PLS
<b>Johns Hopkins</b> - Nicholas Maragakis Baltimore, MD 21287 410-614-9874 .....	PLS / HSP
<b>University of Michigan</b> - Kirsten Gruis Ann Arbor, MI 48109 734-936-9020 .....	PLS / HSP
<b>Saint Mary's Healthcare / Michigan State University</b> - Deborah Gelinias Grand Rapids, MI 49503 616-493-9727 .....	PLS / HSP
<b>St. Louis University</b> - Ghazala Hayat St. Louis, MO 63104 314-977-6086 .....	PLS / HSP
<b>Carolinas Medical Center</b> - Benjamin Rix Brooks, MD Charlotte, NC 28203 704-355-2000 .....	PLS / HSP
<b>Wake Forest University School of Medicine</b> - Michael Cartwright Winston-Salem, NC 27157 336-716-4344 .....	PLS / HSP
<b>Duke University</b> - Richard Bedlack Durham, NC 27707 919-668-7600 .....	PLS / HSP
<b>Dartmouth-Hitchcock Medical Center</b> - Elijah Stommel Lebanon, NH 03756 603-650-5104 .....	PLS / HSP
<b>Robert Wood Johnson Medical School</b> - Jerry Belsh New Brunswick, NJ 08901 732-235-7733 .....	PLS
<b>University of Nevada School of Medicine</b> - David L. Ginsburg, M.D. Las Vegas, NV 89102 702-671-5025 .....	PLS / HSP
<b>Upstate Neurology</b> - James Wymer Albany, NY 12205 518-438-0922 .....	PLS
<b>Beth Israel Medical Center Albert Einstein College- NY</b> - Stephen Scelsa New York, NY 10003 212-844-8490 .....	PLS
<b>Columbia University - MDA/ALS Center</b> - Hiroshi Mitsumoto New York, NY 10032 212-305-1319 .....	PLS
<b>SUNY Upstate Medical University</b> - Jeremy Shefner Syracuse, NY 13210 315-464-4243 .....	PLS / HSP
<b>Cleveland Clinic</b> - Rebecca Kuenzler Cleveland, OH 44195 800-223-2273 .....	PLS / HSP
<b>Ohio State University</b> - John T. Kissel, MD Columbus, OH 43210 614-293-4969 .....	PLS
<b>Ohio State University</b> - Yasushi Kisanuki Columbus, OH 43210 614-293-4969 .....	PLS / HSP
<b>London Health Sciences Centre</b> - Christen Shoemith London, Ontario, CAN N6A 5A5 519-663-3597 .....	PLS / HSP
<b>University of Toronto</b> - Lorne Zinman Toronto, Ontario, CAN M5S 1A1 416-480-4213 .....	PLS
<b>Oregon Health &amp; Science University</b> - Jau-Shin Lou Portland, OR 97239 503-494-7772 .....	PLS
<b>Penn State Hershey</b> - Zachary Simmons Hershey, PA 17033 717-531-4191 .....	PLS / HSP
<b>University of Pittsburgh</b> - Sasa Zivkovic Pittsburgh, PA 15213 412-692-4917 .....	PLS / HSP
<b>University of Pittsburgh</b> - David Lacomis Pittsburgh, PA 15213 412-692-4917 .....	PLS
<b>Drexel University College of Medicine</b> - Terry Heiman-Patterson Philadelphia, PA 19103 215-762-6915 .....	PLS / HSP
<b>Rhode Island Hospital, Brown University</b> - George Sachs, MD, PhD Providence, RI 02903 401-444-8795 .....	PLS / HSP
<b>Texas Neurology</b> - Daragh Heitzman, MD Dallas, TX 75214 212-827-3610 .....	PLS / HSP
<b>University of Texas Health Science Center, San Antonio</b> - Carlyne Jackson San Antonio, TX 78229 210-450-9700 .....	PLS
<b>University of Virginia</b> - Lawrence H. Phillips, II MD Charlottesville, VA 22908 434-924-0000 .....	PLS
<b>University of Vermont</b> - Rup Tandan Burlington, VT 05405 802-847-4589 .....	PLS / HSP
<b>University of Washington</b> - Michael Weiss Seattle, WA 98195 206-598-7688 .....	PLS
<b>Dean Health System</b> - Oliver Ni Madison, WI 53719 608-252-8000 .....	PLS / HSP
<b>Medical College of Wisconsin</b> - PE Barkhaus Milwaukee, WI 53226 414-805-5224 .....	PLS
<b>WVU School of Medicine</b> - Laurie Gutman Morgantown, WV 26508 304-598-4800 .....	PLS / HSP

# Fundraising

## Combined Federal Campaign

Jim Sheorn – [jmsheorn@comcast.com](mailto:jmsheorn@comcast.com)

The Combined Federal Campaign or CFC is a fundraising event in which large sums of money are generated for charities that are approved to participate. There are very strict guidelines to participate. The Spastic Paraplegia Foundation (SPF) once again has been approved to participate. **What does that mean for us?** We need to communicate our message to as many Federal employees that we can and generate much needed research dollars. Each Fall Federal employees from across the USA decide which charities will get contributions from their own pay. Employees get to decide how much they will donate and to which charity or charities their donation will go to. The CFC locations are grouped into different regions. Last year, one region generated over 6 million dollars for their charities.

The SPF has received some money from the CFC each year we have participated, but the amount is very small. That's why we need your help. **Please contact any Federal employee you know and help them understand our fight to find cures for Hereditary Spastic Paraplegia and Primary Lateral Sclerosis.** There are many types of Federal employees including postal workers, military personnel, prison workers, CIA, FBI, VA Hospital workers, IRS, police, fire protection and many others.

If you know of any federal employees, please reach out and let them know that the SPF is approved with the Combined Federal Campaign and our number is 12554. If they would like to learn more, please have them log onto our website at [www.sp-foundation.org](http://www.sp-foundation.org) or they can log onto to the CFC video website. There is a two minute video about the SP Foundation located there.

Thank you in advance for your help. Most employees make their decisions during the Fall CFC Fairs, so please contact those that you know soon. If you have any questions about the CFC, please send me an email and I will do my best to help.

*Ed. Note: The following two articles describe two fundraisers for SPF. One's in NC, the other's in ND. Please share with others, through Synapse, your own fundraising successes. Next issue we'll tell about others.*

## The Magnificent Mile

<http://www.magmilerace.com/>

The Magnificent Mile was first run in November 2006. It is the inspiration of Sarah Witt who has Primary Lateral Sclerosis, a motor neuron disease related to ALS or Lou Gehrig's disease; and Scott and Amy Corsmeier, siblings who have Hereditary Spastic Paraplegia, a hereditary motor neuron disease.



Sarah was an avid runner and marathoner until her illness in 2004. Now, she is confined to a wheelchair and unable to speak. Scott and Amy were diagnosed when they were young children. In the four years between 2006-2009, over \$140,000 has been raised for SPF.

## Airplane Rides Fundraiser

*Submitted and organized by Brenda (Mastrud) Rehder*



In 2006, I was diagnosed with HSP. I am a single mom and live in North Dakota. May 15th and 16th we gathered at Vic's Aircraft Sales.

Coffee and donuts were served in the morning, then lunch, airplane rides, with a silent auction each day. For \$20.00, six pilots gave rides around the Fargo-Moorehead area. State Bank and Trust is a proud sponsor of the SPF through its Pay it Forward program. I got the donations for the silent auction from businesses in town. SPF received a check for \$8085 as a result of this fundraising event.



## Events

**Ames, IA**

**June 12, 2010**

*Submitted by Jackie Wellman*

Nine people came out for this connection and had a great lunch. It was pouring rain but did eventually clear up, and no one got lost. It was good to see sweetie, Laurel Yost again. Laurel brought her son, who was recently diagnosed with HSP and his fiancé. We also had David Gibson and his wife, Linda. We brainstormed on the way home and are going to try to figure out a way to have an online auction for fund-raising. Laurel had a big auction last year for her father who had an accident and needed some help with medical bills.

**Austin, TX**

**July 10, 2010**

*Submitted by Marlene Doolen*

The 2010 Austin Patient Connection was held at the Brick Oven. Ten people enjoyed lunch there in a private room, which allowed for good conversation and for getting to know more about each other. The majority of attendees traveled from their home towns three to six hours from Austin. Marlene Doolen talked about the founding and the history of the SPF, handed out copies of *Synapse*, and handed out copies of the SPF web site's home page to show folks where to find information. We had a great time going around the table and talking about a variety of things, which is an important component of each connection.



**Indianapolis, IN**

**August 21, 2010**

*Submitted by Harvey Mover*

This year marked the 15th anniversary of the Loop for Life. Although the weather and skies were a little gloomy, the event was a success. The Indianapolis Metropolitan Police Department provided a safe escort for the several thousand motorcyclists who rode on two and three wheels on I-465. That is the "Loop", which goes about 60 miles around Indianapolis. Preliminary numbers are still being finalized and will eventually be posted on the website at [www.loopforlife.org](http://www.loopforlife.org). The home for the Loop for Life is the Marion County Fair. In addition to

the ride, the event included breakfast and lunch, ride in bike show, a vendor midway, and live music. Each of the riders received a 2010 t-shirt and could purchase other Loop for Life memorabilia, which is available at from Harley-Davidson of Indianapolis, a primary sponsor. This is the sixth year a portion of the money raised will be donated to SPF, which started when the founder of the Loop for Life [Harvey Mover] was diagnosed with HSP. Other funds will be donated to local charities in Indianapolis. The sponsors and volunteers for the Loop for Life continue to make the difference and allow the event to be the best in Indiana. Check the website for updates and more information at [www.loopforlife.org](http://www.loopforlife.org).

## Upcoming Events

### SPF Team Challenges - a.k.a. TeamWalks

In response to input from many in our community who can't walk, the Board is suggesting a new umbrella term for all local fundraising efforts—SPF Team Challenges. These events are SPF's main source of fund raising, and we need the patient community—that's you-- to participate. By asking others to sponsor you, you'll be helping raise money for research to cure your disease. Please feel free to either keep using *TeamWalk*, or whatever other name you've used for your event and in your letters to prospective donors. ....or, if you want to take the plunge and organize a Team Challenge, go for it. We hope you'll accept the challenge to help.

### New England Team Challenge

**September 11**

**Anheuser Busch Brewery in Merrimack, NH**

*John Swain, Swain [john.swain@nemoves.com](mailto:john.swain@nemoves.com)*

This year's event will be held at the Anheuser-Busch Brewery, which is fully accessible. The day will begin at 11:30 am in the tent where we will have some social time and lunch. After lunch, we will be given a special opportunity to walk or roll over to the barn to visit with the Clydesdales. Typically, the Clydesdales are only accessible on the first weekend of the month, so we are excited to be given this opportunity! Of course, you are welcome to tour the brewery either before the event starts or after, or not at all.

**Town of Clifton Park Farm Fest  
September 11-12  
The Clifton Park Rotary Event  
September 25-26 Clifton Park, NY**

*Isabel Prescott, [farmer.isabel@me.com](mailto:farmer.isabel@me.com) contact*  
A fund-raiser for SPF will be held during the Town of Clifton Park Farm Fest, and during an event sponsored by The Clifton Park Rotary. Riverview Orchards ([RiverviewOrchards.com](http://RiverviewOrchards.com)) is involved in both of these events. Suzanne Meacham and Jennifer Shea, two cousins who have HSP, will be participating. There will be everything from a corn husking contest to cake decorating. These events are lots of fun for families.

**NC Connection and The Magnificent Mile  
September 18-19, 2010  
Hillsborough St, Raleigh, NC**

*Sarah Witt, [srwitt@yahoo.com](mailto:srwitt@yahoo.com)*  
We will have an afternoon social hour and dinner on Saturday night. The 5th Annual Magnificent Mile will be held on Sunday afternoon. Details and registration information is available at [www.MagMileRace.com](http://www.MagMileRace.com).

If you are traveling to Raleigh for the event, discounted rooms (\$79/night) are available for Friday and Saturday nights at the Clarion Hotel. Call 919-832-0501 and ask for a room in the Magnificent Mile block.

**Drive "Fore" Spastic Paraplegia Golf Outing  
September 20, 2010**

**Forsgate Country Club, Monroe Township, NJ**  
*James F. Brewi, [JBREWI@travelers.com](mailto:JBREWI@travelers.com)*  
The Travelers Insurance Company will hold its second Drive "Fore" Spastic Paraplegia Golf Outing.

**New York TeamWalk  
September 25, 2010 Mount Kisco, NY**

*Ann Lakin, [alakin90@aol.com](mailto:alakin90@aol.com)*  
The New York TeamWalk this year will be held on Saturday, September 25, from 11:00 to 1:30. Though it will not be held in conjunction with Mount Kisco Sale Days this year, meaning we won't have an SPF table at sale days, it will be held on the same day to attract more awareness of and profit for SPF.

**Idaho Connections Support Group Meeting  
October 2, 2010 Boise, ID**

*Pat Bernhard, [patricia.bernhard@gmail.com](mailto:patricia.bernhard@gmail.com),  
208-888-4069*

The Idaho Connection will again be held at the Idaho Elks Rehab Hospital. Dr. Robert Friedman will be there to explain upper motor neuron disorders and answer questions. In addition, lunch will be served and a Medtronic representative will talk about the implanted Baclofen pump.

**California TeamWalk for our Cures &  
Connection Weekend**

**October 1-2, 2010 Pleasanton, CA**

*Linda Gentner, [lkgentner@aol.com](mailto:lkgentner@aol.com), 510-651-5676*

A PLS & HSPers Welcoming Dinner will be held Friday night -- pay for your own dinner. There will be discussion time with dessert and coffee following dinner. Get an early start on the weekend and stay at the hotel Friday night with no need to get up early on Saturday morning. The TeamWalk, lunch, and raffle will be at the Valley Community Church.

**Connecticut Connection**

**October 2, 2010 Hartford, CT**

*Dolores Cannon, [d.carron@sbcglobal.net](mailto:d.carron@sbcglobal.net)*

Our 30th CT Connection will be held at the N.E.A.T. Marketplace in Hartford. One of our speakers will be Dr. Jinsy Andrews, who recently relocated to the Hospital for Special Care in New Britain, CT, from Columbia Presbyterian in New York City. Keith Mullinar, who trains service dogs, will speak and demonstrate one of his company's dogs. In addition, Roland Grundmann, a representative from Ride-Away (provider of vehicle modifications), will speak and demonstrate a modified minivan. In the past, he has distributed discount coupons for vehicle purchase and most likely will do so again. Our meetings are open to both PLS and HSP (*and other interested persons*) and not limited to CT residents. If you need hotel accommodations, please let me know ASAP and I'll arrange discounted rates. We can have Friday night dinner for early arrivals. If you extend your stay to enjoy the beautiful Connecticut foliage, we can also plan a group activity for Sunday.

**Tennessee TeamWalk and Connection**

**October 9, 2010 Nashville, TN**

*Jim Sheorn, [jmsheorn@comcast.net](mailto:jmsheorn@comcast.net), 615-479-7369*

Details are currently being arranged.





## E-Books, Audio Books and Music for your Computer

*Contributed by Thurza Campbell*

I had held off buying a Kindle®, as they are too heavy for me to hold. Not only that, but I like to borrow books from the library, not buy them. Annette Lockwood and I had a conversation at the Annual SPF conference. Thanks to Annette, I launched an exciting exploration into free sources of books on-line.

Many of you are like me, and always have your computer with you. You need no separate dedicated device for e-reading or listening. Your own laptop, iPad or portable audio device can be used to read books, and listen to books or music. You can enlarge the book font to whatever size you need, and insert a bookmark when you are stopping.

Taking Annette's advice, I contacted my local library first. I learned that all libraries must buy permission to loan books. Check to see what permits your town, city, county or state has. I live in Massachusetts. Following instructions from my local librarian, I went on-line and got a library card from the Boston Public Library. I don't live in Boston, but in a small town without the funds for its own permits. Any resident in the state can borrow books, audio books and music. See what local resources are available to you.

There's another good resource available worldwide of books to borrow on which the copyright has expired. These e-books can be borrowed free. Go to Project Gutenberg [http://www.gutenberg.org/wiki/Main\\_Page](http://www.gutenberg.org/wiki/Main_Page)

You can also buy music and books on-line and load them into devices you own. Happy listening and reading!

## NIH Enrolling Participants in a Study: Oxidative Stress in Motor Neuron Disease

<http://clinicaltrialsfeeds.org:80/clinical-trials/show/NCT01143428>

Background: - Primary lateral sclerosis (PLS) is a disorder in which nerve cells in the brain that control movement degenerate. The cause of PLS is not known, but some research has suggested that environmental factors that produce oxidative stress trigger PLS in people who carry certain genes. Oxidative stress is

caused when the body makes chemicals called "free radicals" faster than its natural systems can break them down. Oxidative stress can be triggered by exposures to chemicals related to the bodily effects of lead, smoking, alcohol consumption, physical activity, and psychological stress. Chemicals produced by the body during oxidative stress can be measured in the blood and urine. Researchers are interested in studying the physical, neurological, and chemical effects of PLS to better understand the effects of oxidative stress on the disorder. Objectives: - To study the relation of oxidative stress to the diagnosis and progression of motor neuron disease. Eligibility: - Individuals 20 years of age or older who have been diagnosed with PLS, and have had symptoms of PLS for at least 5 but not more than 8 years and been previously enrolled in 01-N-0145 Screening: Neurologic Disorders with Muscle Stiffness Design: - Participants will have an initial study visit and three follow-up visits. Each visit will require approximately 3 days of testing at the National Institutes of Health Clinical Center. - As part of this study, participants will have the following tests and procedures: - Neurological examination to test muscle strength, sensation, coordination, and reflexes, as well as clarity of speech - Tests of memory, attention, concentration, and thinking - Surveys on oxidative stress, including questions on life, mood, jobs held, and habit - Electromyography to record the electrical activity of muscles - Transcranial magnetic stimulation to measure electrical activity translated from their brain to the muscles - Blood, urine, and skin biopsy samples for testing and sample collection - After the initial visit, participants will have three more visits, once each in the following 3 years.

## Physical Therapy Booklet Available

*Contributed by Dolores Carron*

Many of you participated in a survey about your experiences with PT. The survey project was conducted by Mary Pease, a graduate student at Quinnipiac University in Hamden, CT. The end product of that research is a book of exercises that are recommended for PLS (but certainly appropriate for HSP as well). Please contact me directly to request your exercise booklet at [d.carron@sbcglobal.net](mailto:d.carron@sbcglobal.net) or call me at 860-666-9862.

## The Startle Response a.k.a. The Startle Reflex

*Contributed by Malin Dollinger*

I did a simple a study to discover what percent of people with PLS and HSP on our web sharing pages have the startle reflex. I have not asked people who do not have a neurologic problem, for example, if they have a startle reflex. There were 37 responses, about ¾ women and ¼ men. Of the 37 responders, 25 have PLS and 11 have HSP. The term refers to a sudden overactive response to a sudden sound, or unexpected movement. Perhaps we should call this a “startle response” instead of a ‘startle reflex,’ since the latter is normally present in newborn babies, and disappears at about 4 to 6 months of age. In adults the so-called startle reflex [not the same as the one in newborns!] is seen in various neurological conditions involving the brain, as are our upper motor neuron conditions, PLS and HSP. Conclusions:

1. The startle response is very common in PLS and HSP. Since the part of the nervous system that is involved is the brain, it is to be expected, since that’s where the bodies of the upper motor neurons are.
2. You can tell your spouse, significant other, friends, and family that you don’t do this on purpose.

## Kakkis EveryLife Foundation to CureTheProcess

*Emil D. Kakkis, M.D. Ph.D., President*

<http://www.kakkis.org/>

When your child gets sick, you hope there will be a treatment available for them.

For many patients with rare disorders, those treatments don’t exist. This situation is made even more tragic when the science exists to develop a treatment, but the cost and complexities of development have slowed or even stopped progress. Despite the advances made in the last 10 years for some rare diseases, there are hundreds of others where the science exists for a treatment, but the next step of development will not happen.

I created the Kakkis EveryLife Foundation to CureTheProcess and focus on fixing the problems associated with developing treatments for rare disorders.

Our goal is to improve the regulatory environment surrounding the approval of effective treatments. We plan to effect change in regulation of rare disease treatments by insightful scientific analysis and dialogue, grass roots support and political sponsorship, complementing but not competing, with the role of patient societies and research foundations.

### *We believe:*

- \* No disease is too rare to deserve treatment.
- \* We already have the science we need to treat more rare disease patients.
- \* We need an improved process with new study designs and disease measures to accelerate the development of new treatments.
- \* We need the right people in both industry and FDA to make these changes effective.
- \* All new drugs for rare diseases should be safe.

The Kakkis EveryLife Foundation will be focused primarily on analyzing the problems and providing solutions that can accelerate the development process for rare disease treatments. We know it will be hard and complex, but the science is here and we have many patients depending on us, to CureTheProcess and treat them.

## Goals for CureTheProcess Campaign

<http://www.curetheprocess.org/>

The CureTheProcess Campaign strives to inspire science-driven public policy that will increase the predictability of the regulatory process for rare disease treatments. Our goal is to give even the rarest diseases access to the accelerated approval process and fulfill more completely the original intentions of the Orphan Drug Act. The following goals need to be reached to put orphan treatments on the fast track:

***Establish a new Office of Drug Evaluation  
for Genetic and Biochemical Diseases,  
consolidating expertise to review treatments  
to ensure they are safe and effective.***

Biochemical and genetic disorders require specialized training and experience to best evaluate new therapies. The establishment of a new Office of Drug Evaluation for rare biochemical and genetic diseases would assure that the appropriate rare disease experts are recruited and integrated with existing expertise at FDA.



***Devise new clinical study design and analysis paradigms for rare diseases that properly account for clinical heterogeneity and disease complexity to properly capture treatment effects.***

While traditional randomized, controlled studies have been used in rare diseases, this design is relatively insensitive to changes in heterogeneous patients and fails to allow the assessment of all types of patients with all types of disease outcomes. A creative effort is needed to develop new paradigms in study design that capture individual benefit in a broad array of patients, utilizing all the clinical data to establish efficacy. The medical science needs to drive the statistical analysis.

***Create a new standard for the surrogate and biomarker endpoints used for rare disorders, to allow treatments for these diseases to have full access to the accelerated approval pathway.***

Due to the rarity of the disorders, the use of direct, relevant surrogate or biomarker endpoints as clinical study endpoints is essentially impossible for some rare disorders because none of these surrogates have been validated or evaluated in clinical studies and are therefore unavailable for development use. However, the data show that biochemical markers relevant to biochemical genetic disorders are far better predictors of disease and treatment effect than many of the approvable surrogate markers currently used for drug approvals.

**Outcomes:**

A **streamlined development path** will shorten timelines and reduce the financial risk associated with development of rare disease therapeutics. The result should be a surge in development activity for even the most rare disorders.

More patients with rare biochemical and genetic disorders will get **earlier access** to specific, effective treatments.

Certain treatments for rare biochemical or genetic disorders that are now unaddressed because of the difficulty in assessing the clinical outcome, will now be targets of drug development as **appropriate surrogate markers are identified**.

**Investment in early stage biotech** companies focused on rare diseases will increase and have a positive impact in local communities and biotechnology jobs.

A new Office with experts trained and knowledgeable in the disease area, will allow for **an improved and more specialized FDA review**.

**July 15, 2010 (Novato, California)** - Just 16 months after the Kakkis EveryLife Foundation kicked off the CureTheProcess Campaign, the Foundation, in association with the National Organization of Rare Diseases (NORD) and numerous other patient and physician organizations, have increased the support and commitment to improving rare disease regulatory policies.

A US Senate Appropriation bill has been submitted including language supporting two of the Campaign's goals. Specifically, the Bill supports the creation of new guidances which could improve the scientifically sound use of surrogate endpoints and new clinical study designs and analysis. The Senate Bill also includes an appropriation for the Food and Drug Administration to hire new staff to fulfill these requirements.

"We are especially grateful to Senator Sam Brownback (R-KS) for his leadership on this issue and to Senator Herb Kohl (D-WI) for his support," said Emil Kakkis, M.D. Ph.D., President of the Kakkis EveryLife Foundation. "We are very pleased to see so much progress made, in such a relatively short time."

The bill includes the first increase for the Orphan Product Development Grant program since FY 2005. The program is increased by \$2,000,000 for a total grant level of \$16,035,000. The Bill also includes specific funding for the Office of the Associate Director for Rare Diseases in the Center for Drug Evaluation and Research (CDER). Funding for this office is increased by \$1,000,000 to hire additional staff with specific expertise in facilitating the development and review of products to treat rare diseases.

***How can you help?***

\*Click on <http://www.kakkis.org/> to learn more.

\*Contact your US Senator to request his/her support for this important work.

## Reactions/Attitudes

*Contributed by Annette Lockwood*

After reading the story that follows, I was reminded of a phrase that I read several years ago, "It is not the action - it is your reaction that counts."

This phrase really hit home with me. I even used it in a speech to high school graduates. You can't change the "action or event" that upsets or haunts you but you can change how you respond or react to it.

John is the kind of guy you love to hate. He is always in a good mood and always has something positive to say. When someone would ask him how he was doing, he would reply, 'If I were any better, I would be twins! He was a natural motivator. If an employee was having a bad day, John was there telling the employee how to look on the positive side of the situation. Seeing this style really made me curious, so one day I went up and asked him, "I don't get it! You can't be a positive person all of the time. How do you do it?" He replied, "Each morning I wake up and say to myself, 'you have two choices today. You can choose to be in a good mood or you can choose to be in a bad mood.' I choose to be in a good mood. Each time something bad happens, I can choose to be a victim or I can choose to learn from it. I choose to learn from it. Every time someone comes to me complaining, I can choose to accept their complaining or I can point out the positive side of life. I choose the positive side of life. "Yeah, right, it's not that easy," I protested. "Yes, it is," he said. "Life is all about choices. When you cut away all the junk, every situation is a choice. You choose how you react to situations. You choose how people affect your mood. You choose to be in a good mood or bad mood. The bottom line: It's your choice how you live your life.

I reflected on what he said. Soon hereafter, I left the Tower Industry to start my own business. We lost touch, but I often thought about him when I made a choice about life instead of reacting to it. Several years later, I heard that he was involved in a serious accident, falling some 60 feet from a communications tower. After 18 hours of surgery and weeks of intensive care, he was released from the hospital with rods placed in his back. I saw him about six months after the accident. When I asked him how he was, he replied, "If I were any better, I'd be twins...Wanna see my scars?"

I declined to see his wounds, but I did ask him what had gone through his mind as the accident took place. "The first thing that went through my mind was the well-being of my soon-to-be born daughter," he replied. "Then, as I lay on the ground, I remembered that I had two choices: I could choose to live or I could choose to die. I chose to live." "Weren't you scared? Did you lose consciousness?" I asked. He continued, "...the paramedics were great. They kept telling me I was going to be fine. But when they wheeled me into the ER and I saw the expressions on the faces of the doctors and nurses, I got really scared. In their eyes, I read 'he's a dead man' I knew I needed to take action."

"What did you do?" I asked. "Well, there was a big burly nurse shouting questions at me," said John. "She asked if I was allergic to anything. 'Yes,' I replied." The doctors and nurses stopped working as they waited for my reply. I took a deep breath and yelled, "Gravity!" Over their laughter, I told them, "I am choosing to live. Operate on me as if I am alive, not dead." He lived, thanks to the skill of his doctors, but also because of his amazing attitude. I learned from him that every day we have the choice to live fully. Attitude, after all, is everything.

### ***Strong in Spirit***

*By Liz Lepper*

*I am strong in spirit,  
But weak in body  
Days to weeks,  
Weeks to months,  
Months to years,*

*I feel the years fly by  
As I face the future  
Baring my face to the sun  
I let the warmth spread over me  
Shattering the chains of darkness,*

*As time begins to turn once more,  
Darkness arise to circle around me  
Dragging me down into its depths,  
I crumble beneath this heavy weight  
Shattered by the sorrow of losing a love*

## EXERCISE AND YOU: THE IMPORTANCE OF POSTURE

*Contributed by Liz Wroblewski, MPT*

The last issue I wrote about the efficient technique of diaphragmatic breathing. Its uses include oxygenation of the muscles, relaxation, core muscle activation and mobilization of the spine. This deep breathing goes hand in glove with optimal posture. Good posture allows the rib cage to fully expand for breaths and positions the shoulder girdle for good upper extremity function. Your ribs literally sit on your pelvis! The pelvis is “rolled back” in slouch sitting compromising core support by the lower trunk muscles.

The Slouch-OverCorrect exercise shown below assists in finding and practicing an optimal sitting posture. Start in the slouch position as in A. Then arch the low back to the extreme as in B and then release that overcorrection by “10%” as in C. Practice this throughout the day. Catch your slumps and correct to an optimal position. You don’t want either extreme so remember to “release by 10%”; that is, release just a little. Note how your pelvis positions itself on the “sit bones” to bear body weight. Now practice your deep breathing in this position.

Posture and breathing are building blocks for an exercise program. Incorporate these into your daily activities and exercise. Practice this sitting on a stability ball adding simple balance challenges such as arm lifts and small amplitude alternate hip marches maintaining the 10% position. This is a challenge!

For prolonged sitting choose a chair with a back. Insert a lumbar roll in the small of your back to support the lumbar curve. You can purchase a lumbar roll commercially or go to How to Make a Lumbar Roll on YouTube or Google “how to make a lumbar roll” for instructions.



- A -  
Slouch



- B -  
Over Correct



- C -  
“10%” Release  
Optimal Position

### My Sweet Silence

*By Liz Lepper*

As my silent cries  
Echo through the midnight sky  
An answering cry comes my way  
Long and deep,  
Dark yet sweet

As my silent tears  
Wash away the midnight stars  
An answering sound fills my mind  
Broken and weary,  
Empty yet full

As my silent sorrow  
Fills every crack in my soul  
Driving the darkness deeper in  
Cold and alone,  
Strong yet weak

As my silent burdens  
Drag me down into the ground  
Allowing the darkness back in  
Scared and despairing,  
Hopeful yet bleak

### Synapse Appeal

Please use the enclosed envelope to make a donation. Thanks to everyone who donated in 2010 to help offset the printing and distribution expenses.

### Support SPF with One Step a Month

Consider making a monthly donation to help SPF move a step closer to a cure. Our One Step a Month Program is a win-win! Recurring gifts allow us to plan ahead with confidence, making sure we take the best steps towards finding the cures for HSP and PLS. Plus, recurring donations allow you to give in a convenient, safe and secure way. <http://www.spf-foundation.org/donate.htm>



## Medical Updates

### **UM Researcher to Test Stem Cell Treatment for Alzheimer's Results from ALS Trials Spur Optimism**

May 2, 2010 By Ryan Beene and Tom Henderson

Buoyed by early results of stem cell-based trials on patients with Lou Gehrig's disease, Eva Feldman, M.D., co-director of the A. Alfred Taubman Medical Research Institute at the University of Michigan Medical School, is now taking aim at a far bigger target: Alzheimer's disease. The investigation into an Alzheimer's treatment piggybacks on current Phase I human trials for patients with Lou Gehrig's disease led by Feldman that are under way at Emory University Hospital in Atlanta. The trials test the safety of injecting neural progenitor cells, essentially stem cells that have developed beyond the embryonic phase and are predisposed to becoming nerve cells, into the spinal cords of patients with Lou Gehrig's disease. Feldman will continue to serve as principal investigator on that trial - the first FDA-approved trial using stem cells on Lou Gehrig's patients in the U.S. - as she and her team begin work on Alzheimer's trials. Eighteen Lou Gehrig's patients will be tested in all. The disease afflicts as many as 30,000 patients in the U.S. Feldman sped up her timetable for taking on Alzheimer's after seeing promising early results with three Lou Gehrig's patients. The first patient was injected on Jan. 19. The third operation, on April 14, was filmed by CNN. Feldman said she is prohibited from discussing whether patients report such results as increases in strength or sensation. But there have been no ill effects from the three surgeries. Each patient is injected at five spots on the spinal cord, with about 100,000 cells per injection. Feldman said she is excited about expanding stem cell trials to Alzheimer's because of the far larger pool of would-be patients. "Alzheimer's is going to be easier to do than ALS," said Feldman. The transition from Lou Gehrig's to Alzheimer's disease is a natural one because the treatment potentially addresses the same problem. The neural progenitor stem cells work by surrounding specific large nerve cells that are sick and halting further degeneration caused by the disease, Feldman said. "In the spinal cord, these nerve cells produce the nerve tissue fibers

that extend through the muscles of our body, and in the brain, the same type of nerve cell facilitates thinking processes," Feldman said. "The kind of stem cells we're using have a particular proclivity to rescue cholinergic neurons, and it's cholinergic neurons that degenerate and become diseased in Lou Gehrig's disease and Alzheimer's disease." The Phase I Lou Gehrig's disease trials are scheduled to finish by the end of June 2011. If they go as hoped, Phase II trials, which assess efficacy, can begin as early as January 2012. Feldman said Phase II trials could add the UM hospital as a test site in addition to Emory. Feldman said she recently took on a new, young ALS patient, to whom she could, for the first time in her 20 years of treating patients at UM, offer some encouraging words about future treatments. "For 20 years, there has been little hope I could offer patients. Now there is truly tangible hope. We are truly beginning to try a therapy that can allow us to help halt the progress of this dangerous disease," she said. "Patients ask me 'what will the future hold?' I told my new patient, things are extremely hopeful now. The future is very bright. And not just with ALS or Alzheimer's, but with Parkinson's and Huntington's, too."

### **Parkinsonism and Motor Neuron Diseases: Twenty-Seven Patients With Diverse Overlap Syndromes**

Movement Disorders 2010 Jul 28. [Epub ahead of print]  
Gilbert RM, Fahn S, Mitsumoto H, Rowland LP.  
Department of Neurology, NYU Langone Medical Center,  
New York, New York, USA.

It has long been recognized that signs of motor neuron disease (MND) may accompany clinical evidence of parkinsonism in different neurodegenerative conditions. By using the Columbia University Division of Movement Disorders database, we reviewed data from 5,500 cases of parkinsonism and recorded the presence of upper motor neuron (UMN) dysfunction, lower motor neuron (LMN) dysfunction, or both. Among the 27 patients so identified, we counted those with autonomic dysfunction, cerebellar dysfunction, or dementia. Among the 27 cases, seven had UMN signs and LMN signs as well as parkinsonism and were diagnosed with amyotrophic lateral sclerosis (ALS)-parkinsonism (Brait-Fahn

disease). Three of the seven had dementia that was not deemed to be frontotemporal dementia (FTD). Six other patients had no LMN signs but had UMN signs and parkinsonism and were classified as having primary lateral sclerosis (PLS)-parkinsonism. Four patients had both UMN and LMN signs with parkinsonism as well as the characteristic dementia of FTD; they were diagnosed with FTD-parkinsonism-ALS. Seven patients had MND, parkinsonism, and autonomic or cerebellar dysfunction, a combination compatible with multiple system atrophy (MSA). Three patients had syndromes compatible with hereditary spastic paraplegia (HSP). In sum, we found that MND occurs in association with diverse parkinsonian syndromes; some are heritable, others sporadic and causes are uncertain. Having MND may be a risk factor for parkinsonism. A prospective study may elucidate this possibility.

### **A Genome-Scale DNA Repair RNAi Screen Identifies SPG48 as a Novel Gene Associated with HSP.**

PLoS Biol. 2010 Jun 29;8(6):e1000408.

*Sabicki M, Theis M, Krastev DB, Samsonov S, Mundwiler E, Junqueira M, Paszkowski-Rogacz M, Teyra J, Heninger AK, Poser I, Prieur F, Truchetto J, Confavreux C, Marelli C, Durr A, Camdessanche JP, Brice A, Shevchenko A, Pisabarro MT, Stevanin G, Buchholz F. Max Planck Institute for Molecular Cell Biology and Genetics, Dresden, Germany.*

DNA repair is essential to maintain genome integrity, and genes with roles in DNA repair are frequently mutated in a variety of human diseases. Repair via homologous recombination typically restores the original DNA sequence without introducing mutations, and a number of genes that are required for homologous recombination DNA double-strand break repair (HR-DSBR) have been identified. However, a systematic analysis of this important DNA repair pathway in mammalian cells has not been reported. Here, we describe a genome-scale endoribonuclease-prepared short interfering RNA (esiRNA) screen for genes involved in DNA double strand break repair. We report 61 genes that influenced the frequency of HR-DSBR and characterize in detail one of the genes that decreased the frequency of HR-DSBR. We show that the gene KIAA0415 encodes a putative helicase that interacts with SPG11 and SPG15, two proteins mutated in hereditary spastic paraplegia (HSP). We identify mutations in HSP patients, discovering KIAA0415/

SPG48 as a novel HSP-associated gene, and show that a KIAA0415/SPG48 mutant cell line is more sensitive to DNA damaging drugs. We present the first genome-scale survey of HR-DSBR in mammalian cells providing a dataset that should accelerate the discovery of novel genes with roles in DNA repair and associated medical conditions. The discovery that proteins forming a novel protein complex are required for efficient HR-DSBR and are mutated in patients suffering from HSP suggests a link between HSP and DNA repair.

### **PatientsLikeMe: Some Highlights of Its First Four Years**

**Spring 2006:** The ALS community launches!

**July 2007:** Your contributions lead to PatientsLikeMe's first scientific discovery. PatientsLikeMe publishes a paper in *Acta Psychiatrica Scandinavica* showing that some PALS experience uncontrollable bouts of excessive yawning; an unusual symptom twice as common in the bulbar-onset form of the disease compared to those with a limb-onset.

**February 2008:** An Italian study finds that the drug lithium carbonate slows the progression of ALS. Many of our PALS begin taking the drug, and so with your help, PatientsLikeMe embarks on our own observational study of the drug by launching a novel tool on the site.

**May 2008:** Your voice in research is heard loud and clear. PatientsLikeMe publishes its first survey in the *European Journal of Neurology*, showing that while 90% of PALS were warned about physical symptoms of their condition, only 10% were warned about possible psychological consequences, and two-thirds *wanted* to be told.

**August 2009:** Thanks to you, PatientsLikeMe now has the largest dataset of patients with Primary Lateral Sclerosis (PLS) and Progressive Muscular Atrophy (PMA) online than ever recorded before. We shared this data back to the community through an upgrade to our ALS charts.

*Ed note: As of mid August 310 PLS have registered.*

**December 2009:** The power of sharing your health data is highlighted in the PatientsLikeMe presentation at the 20th International ALS/MND Symposium in Berlin where the PatientsLikeMe research team unveiled our next round of analyses on the lithium study. The team also presents preliminary results of

our genetics tool, and a commercial project exploring emotional lability in ALS, undertaken with our partners Avanir Pharmaceuticals (see “how we make money”).

**January 2010:** PatientsLikeMe rolls out new profile charts in the ALS community to help you, the patient, understand your own profile better and tell your story more effectively.

**May 2010:** This month we see the release of a study carried out in collaboration with our partners at the NEALS Consortium investigating some of the barriers to research participation that some of you may experience. Additionally, all your contributions to the lithium study have been collected and analyzed by now, and the PatientsLikeMe research team submits it for peer-review publication to a leading journal. Thanks to you and your peers, this final paper highlights the potential of new ways of collecting and analyzing data in ALS to advance clinical trials and find faster cures.

### **Cognitive Profile in HSP with Thin Corpus Callosum and Mutations (Executive brain function effected) in SPG11**

*L. Siri1, F. M. Battaglia1, A. Tessa2, A. Rossi3, M. Di Rocco4, S. Facchinetti1, M. Mascaretti1, F. M. Santorelli2, E. Veneselli1, R. Biancheri1* Child Neurology and Psychiatry Unit, G. Gaslini Institute and University of Genova, Italy2 Molecular Medicine Unit, Bambino Gesù Pediatric Hospital, Roma, Italy3 Pediatric Neuroradiology, G. Gaslini Institute, Genova, Italy 4 Second Unit of Pediatrics, G. Gaslini Institute, Genova, Italy

Neuropediatrics 2010; 41(1): 35-38 Autosomal recessive HSP with thinning of the anterior corpus callosum (ARHSP-TCC) due to mutations in SPG11 on chromosome 15q (MIM610844) is the single most common cause of ARHSP. It is characterized by slowly progressive paraparesis and peripheral neuropathy. Although cognitive impairment, sometimes diagnosed as mental retardation, is an almost invariable feature, the extent and specific neuropsychological features are not fully understood. We report a comprehensive neuropsychological assessment in two ARHSP-TCC patients harbouring mutations in SPG11. A specific impairment in executive functions occurring even before cognitive decline, may be considered the core of the neuropsychological profile of patients harbouring mutations in SPG11.

### **Brain Trauma May Produce Conditions which Mimic ALS**

*Condensed from an article in New York Times by Alan Schwarz*

In the 71 years since the Yankees slugger Lou Gehrig declared himself “the luckiest man on the face of the earth,” despite dying from a disease that would soon bear his name, he has stood as America’s leading icon of athletic valor struck down by random, inexplicable fate.

A peer-reviewed article in a leading journal of neuropathology, suggests that the demise of athletes like Gehrig and soldiers given a diagnosis of ALS might have been catalyzed by injuries only now becoming understood: concussions and other brain trauma.

Doctors at the Veterans Affairs Medical Center in Bedford, Mass., and the Boston University School of Medicine, the primary researchers of brain damage among deceased National Football League players, said that markings in the spinal cords of two players and one boxer who also received a diagnosis of ALS indicated that those men did not have ALS. They had a different fatal disease, doctors said, caused by concussion like trauma, that erodes the central nervous system in similar ways.

The finding could prompt a redirection in the study of motor degeneration in athletes and military veterans being given diagnoses of ALS at rates considerably higher than normal, said several experts in ALS. The Yankees legend had a well-documented history of significant concussions on the baseball field, and perhaps others sustained as a battering-ram football halfback in high school and at Columbia University. Given that, it’s possible that Gehrig’s renowned commitment to playing through injuries like concussions, which resulted in his legendary streak of playing in 2,130 consecutive games over 14 years, could have led to his condition.

### **Crucial Part of Cell Affected by Mutated HSP Related Protein**

*Contributed by Mark Weber*

The principle investigator in the article below, Dr. Craig Blackstone, is on the SPF Scientific Advisory Board. Although it may appear that this article is simply about a part of a cell, it is about THE PART that many believe is being harmed by the defective proteins created by mutated genes that cause HSP.



## "Further Assembly Required: Construction And Dynamics Of The Endoplasmic Reticulum Network"

Seong H. Park and Craig Blackstone

### Abstract

The endoplasmic reticulum (ER) is a continuous/membrane system comprising the nuclear envelope, ribosome-studded peripheral sheets and an interconnected network of smooth tubules extending throughout the cell. Although protein biosynthesis, transport and quality control in the ER have been studied extensively, mechanisms underlying the notably diverse architecture of the ER have only emerged recently; this review highlights these new findings and how they relate to ER functional specializations. Several protein families, including reticulons and DP1/REEPs/Yop1, harbour hydrophobic hairpin domains that shape high-curvature ER tubules and mediate intramembrane protein interactions. Members of the atlastin/RHD3/Sey1 family of dynamin-related GTPases mediate the formation of three-way junctions that characterize the tubular ER network, and additional classes of hydrophobic hairpin-containing ER proteins interact with and remodel the microtubule cytoskeleton. Flat ER sheets have a different complement of proteins implicated in shaping, cisternal stacking and microtubule interactions. Finally, several shaping proteins are mutated in HSP, emphasizing the particular importance of proper ER morphology and distribution for highly polarized cells.

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<http://www.sp-foundation.org/shopping.htm>

GoodShop features a multitude of popular stores for all your needs. The percentage donated varies by store and appears on the website. There is no additional cost and you can still take advantage of sales and coupons!

## Caring\*

### Words for Reflection

*Frederick Buechner*

"Your life and my life flow into each other as wave flows into wave, and unless there is peace and joy and freedom for you, there can be no real peace or joy or freedom for me. To see reality--not as we expect it to be but as it is--is to see that unless we live for each other and in and through each other, we do not really live very satisfactorily; that there can really be life only where there really is, in just this sense, love."

### **RESPIRE CARE**

*From "Inspire" magazine, from Mass. ALSA*

One way to avoid caregiver burnout is for the caregiver to recognize his or her limits.

Many agencies offer respite care. Contact the National Respite Network at 1 (800) 773-8433 or [www.chtop.org/ARCH/National-Respite-Locator.html](http://www.chtop.org/ARCH/National-Respite-Locator.html) for help finding resources in your area.



**[www.carepages.com](http://www.carepages.com)**

Use CarePages to connect family and friends and share news, support and encouragement. Whether you are in the hospital, receiving care at home, or recovering, CarePages provides the forum to share stories and get support.

*\* formerly Caregiving. The name of this section has changed to reflect that caring goes two ways – given and received by both.*

## The Duel

by Eugene Field

The gingham dog and the calico cat  
Side by side on the table sat...

The air was littered, an hour or so,  
With bits of gingham and calico...

While the old Dutch clock in the chimney place  
Up with its hands before its face,  
For it always dreaded a family row...

Next morning, where the two had sat  
They found no trace of dog or cat...

The truth about the cat and pup  
Is this:  
they ate  
each other up.



**G**ather round SPF friends, I'd like to share a thought or two,  
And when I'm done, you can decide if what I say is true.  
My words are rough and trite I know,  
But Friends, I'm worried we're turning into foes!  
So please listen to the thoughts I express, and take no offense,  
My concerns, you see, are making me quite tense...

HSP or PLS? Which disease deserves the best?  
Doctors, treatments, money too,  
Solutions that number more than 'a few'

PLS or HSP? It's My disease - that, all others, supersede.  
HSP is bad, indeed, but most can talk and most can walk.  
HSP is weakness and pain,  
BUT I have PLS, so don't you dare complain!

PLS is bad, it's true,  
There are things we can't do,  
Legs and arms cease to work,  
It can make you go berserk,  
Words are slurred and hard to hear,  
A silenced voice is the fear.

Yes, PLS – it's terrible and it's sad,  
But move on over, move aside,  
HSP deserves the moneyed ride!

But wait - HSP or PLS? My disease deserves the best?  
PLS or HSP? My doctor's research is the one to heed.  
Hmm, when did my disease become the only need?

HSP AND PLS – both cause sadness and distress –  
In mind, body, and spirit, I must confess.  
PLS AND HSP – both cause balance to falter and legs to weaken,  
Our hope for a cure fades like an old waning beacon.  
Motor neuron problems are so unjust,  
Why oh why, did this happen to us!

US! US! Just a moment now, isn't that the key?  
We belong to the SPF group, there should be no you or me.  
Instead of succumbing to anger and despair,  
We need to unite as one; this dedicated group repair!  
There is no PLS OR HSP, instead there needs to be US,  
And WE, not them or me!  
HSP and PLS – all of us want answers, a belief,  
That someday soon, we'll get relief.  
PLS and HSP– we want to move freely amidst laughter and fun,  
And to dance unaided in the sun.  
So let's run this race, in a manner of speaking, together.  
We're not on opposite sides of the fence, but birds of a feather.

HSP or PLS? Which disease deserves the best?  
Both. And that's the real test.

So now friends, my words I'll bring to a close,  
For the night is descending and soon I must doze.  
Let me share one last thought: don't let this subject be for naught,  
PLS And HSP – listen to your heart, I think you'll agree,  
That united as one, our will and our work can not be undone!

– Beth Anne Shultz

*"Success is not measured by what you accomplish, but by the opposition you have encountered, and the courage with which you have maintained the struggle against overwhelming odds."*

– Orison Swett Marden

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**Houston 2011 SPF Annual Conference: "Rocketing Toward The Cure Through Research"**



*A Houston  
Zoo Resident*



*Butterflies at the Museum  
of Natural History*



*Downtown Houston*



*Sam  
Houston  
Monument*



*See NASA's  
future plans  
at the Space  
Center*



*Hilton Americas-Houston*



*Hotel's pool on the 24th floor*



*Hotel's skyline terrace*