



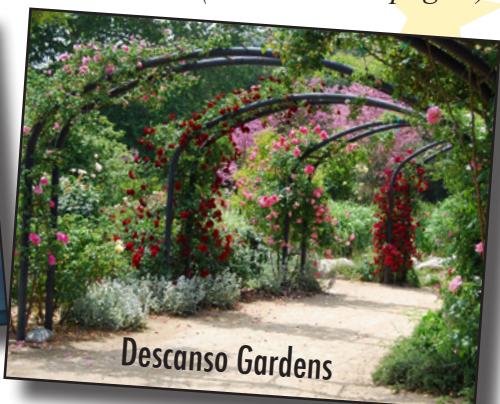
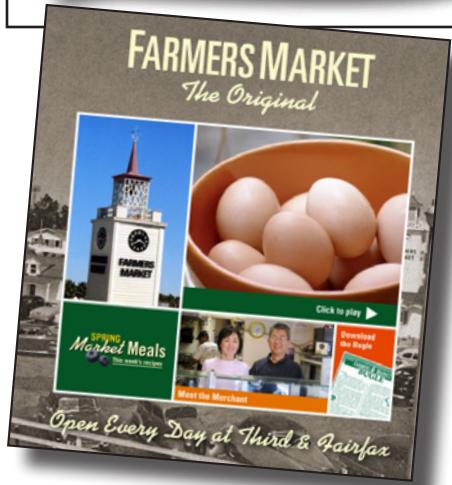
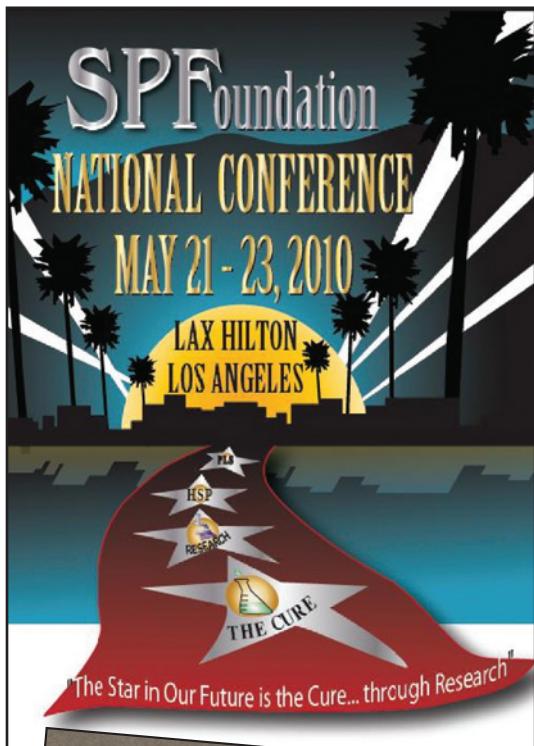
Volume 13, Issue 2

Spring, 2010

Newsletter of the Spastic Paraplegia Foundation

THE 2010 NATIONAL CONFERENCE

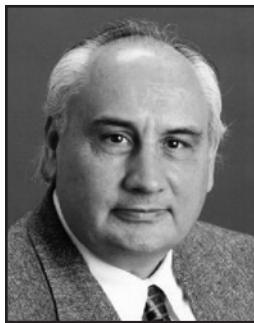
Los Angeles, California - May 21-23



(Continued on page 4)

Editor's Note: I welcome all Letters to the Editor, please e-mail me synapsePLS@comcast.net. I will always print your letters in the next issue.

A Letter from the SPF President



One of the Foundation's main goals is to fund research. We post the research guidelines on the web site in the spring and we award research grants in the fall. It is quite literally an opportunity to reap what we sow. We are building some exciting new relationships with teaching hospitals. They are committed to finding cures for motor neuron diseases and see quite a few HSP and PLS patients. Accelerating research is very important to identifying causes and finding treatments. These treatments could ultimately slow down progression. Some day they will be able to prevent the damage. We will be announcing some of these relationships as soon as we can.

Many of you received an invitation to complete the on-line community survey. The survey is anonymous and takes less than a minute to complete. In this issue you will find what we've learned thus far. You will also find the link to the survey.

Many of you added quite a few suggestions and personal comments on the survey. Some of you shared your ideas by sending them to ideas@sp-foundation.org. One idea was for stretching and yoga videos. Another is to have a place where HSP patients can monitor their progress. We are paying attention and will work on making these available in some form. The National Conference 2010 will be held at the LAX (Airport) Hilton in Los Angeles, May 21-23, 2010. The conference will feature many ways to stretch and stay fit. You'll have an opportunity to learn from other members, you will hear about genetics testing and, of course Dr Fink. I look forward to meeting each and every one of you there. I hope to have a chance to meet you in Los Angeles personally.

Mike Podanoffsky
SPF President

For links mentioned in this publication, go to
<http://sp-foundation.org/synapse/0410>.

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Synapse

Volume 13, Issue 2 - Spring 2010

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).

Synapse Editors

Thurza CampbellSenior Editor
Betsy BaquetMedical Research
Sarah Roberts-WittEvents

Published three times a year for the HSP/PLS community Available online at www.sp-foundation.org

Corporate Address:

Spastic Paraplegia Foundation
212 Farm Rd.
Sherborn, MA 01770-1622

Please Send Donations to:

Spastic Paraplegia Foundation
P.O. Box 1208
Fortson, GA 31808

Please direct correspondence to:

Spastic Paraplegia Foundation
7700 Leesburg Pike, Ste 123
Falls Church, VA 22043
(877) 773-4483
information@sp-foundation.org
www.sp-foundation.org

SPF Board of Directors:

Mike Podanoffsky, President
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Members: Corey Braastad, PhD, Kris Brocchini, Jim Campbell, Jean Chambers, RN, Malin Dollinger, M.D., Ashton Hecker, Annette Lockwood, Jim Sheorn, Mark Weber, Esq.,

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

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National Conference Roundup

FRIDAY, MAY 21, 2010

3:00-5:00pm – Registration

5:00-5:45pm – Welcome cocktail reception cash bar and simple hors d'oeuvres

5:45-6:00pm – Welcome and Introductions by *Craig Gentner*

6:00-7:00pm – Dinner, including dessert

7:00-7:15pm – Break

7:15-8:15pm – “Welcome to the Convention You'd Rather Not Qualify For” by *Malin Dollinger and Craig Gentner*

8:15pm – Instructions for Saturday

8:15pm – Meeting rooms available for meet/greet/social time. You may take this opportunity to meet new people, to discuss challenges and victories, and to get help with day-to-day living

SATURDAY, MAY 22, 2010

8:00-9:00am – Registration Desk Open

9:00am – Welcome by *Craig Gentner*

9:15-10:15am – Keynote Address by *Gary Karp “Life on Wheels”*

10:15-11:00am – “Genetics and HSP/PLS Testing” talk by *Dr. Corey Braastad, Athena Diagnostics and SPF Board Member*

11:00-11:15am – Break

11:15am-12:00pm – *John Fink, M.D. “Current research and understanding of HSP and PLS--What's New and Exciting”*

12:00-1:30pm – Lunch Exhibitors meet and greet

1:30-2:30pm – “Get Fit While You Sit” Talk and Demonstration of Chair Yoga.

2:30-3:00pm – “SPF Research Grants, Funding, and Our Scientific Review Process ~ How Contributions Will Become the Cure” by *Mark Weber, Esq. SPF Board Member*

3:00-3:15pm – Break

3:15-4:00pm – “Adaptation to Disability” by *Gary Karp*

4:00-4:45pm – Simultaneous Breakout Sessions

- ◆ Stress Relief by Acupressure by *Dr. Lenore Dollinger*
- ◆ Caregivers' Time by *Jim Campbell*
- ◆ Maximizing Your Abilities



5:00pm – Entire group gathers for closing remarks by *Craig Gentner*

Dinner on your own – (There are 3 restaurants in the hotel and others nearby.)

7:00pm – Meeting rooms available for meet/greet/social time.

SUNDAY, MAY 23, 2010

7:30am – State Ambassadors breakfast meeting led by *Linda Gentner*

9:00am – Greeting by *Craig* and outline of today's schedule

9:15-9:45am – “Genetics and HSP/PLS Testing” by *Dr. Corey Braastad, Athena Diagnostics and SPF Board Member*

9:45-10:30am – Breakout Sessions

- ◆ Informal time with Dr. Braastad, for further discussions
- ◆ (Additional Speaker/Title pending)

10:30-11:15am –(additional talk to be announced)

11:15am – Closing Remarks by *Craig Gentner*

11:00-11:30am – Check Out

11:00am-12:30pm – Lunch on your own

1:00pm – Afternoon free for tours that you arrange

(Continued from page 1)

We look forward to rolling out the red carpet at the LAX Hilton, meeting you, and sharing our journey together. All of the details on the hotel's various handicapped room options, as well as registration details can be found easily in two places. You can either go on-line to <http://www.sp-foundation.org/2010nc.htm> or mail in the registration form which was in your Winter Synapse. If you have any questions or need help or advice about attending, send an email to nc@sp-foundation.org.

Regarding Renting a Scooter

We do not yet know if one of the scooter companies will be providing scooters for use during the meeting, without charge. Pending that information, you may wish to rent a scooter yourself. There are two companies that can do this:

South Bay Home Health Care: Weekly rental only @ \$150/week They will deliver to the hotel.
www.myhealthcenter.com or call (310) 618 9555

Scootaround Mobility Solution: 4-7 days at \$235 (less than 250 pound person) or \$250 for over 250 pound person www.scootaround.com

This is provided for your information only, and we cannot vouch for or guarantee any service provided by them.

Things to consider and determine when arranging a tour:

- Pick up time and ending time
- Pick up and ending at LAX Hilton
- Availability of restroom both on board and at planned stops
- Ability to transport mobility aid: wheelchair/scooter/other
- How far do you have to walk (*or roll!*) during tour stops
- Cost of tour and method of payment
- Ability to obtain smaller van or bus, if appropriate
- Ability to securely reserve tour by mail or email or phone
- Ability to stop for refreshments or bathrooms, as needed
- Air conditioning of bus or van
- Ability to handle your special needs, in addition to above

SP Foundation News

Community Outreach Task Force (COTF)

Report #2

Jim Campbell

Reaching out to the SPF community to find out how the SP Foundation could better serve our constituents continues to be the top priority for the SPF Board. The task force launched in October, consist of - Jean Chambers, Marlene Doolen, Ashton Hecker, Judy Johnson, Mari White and me. Our Survey of State Ambassadors completed in January shows they are dedicated and interested in having more visibility and better assisting the SPF national organization. While we wait for the results of the every member Community Survey we are addressing several areas where the State Ambassadors have told us our national SPF product could be improved:

1. A more complete welcoming package for new State Ambassadors including a list of references and more "how to" instructions
2. Resource Book for Connections – how to organize both small, casual Connections as well as larger, more formal seminar-oriented Connections.
3. Add SA bios and photos to the SPF Website
4. Update to the Treatment section of the SPF Website
5. Establish list of HSP and PLS doctors and facilities

We will continue to work these areas and report our progress in future editions of *Synapse*. In the meantime if you have any ideas for us to consider as to what services SPF should provide, more effective ways of reaching our current membership, or ways to extend our reach into the HSP and PLS communities feel free to email the entire committee at cotf@sp-foundation.org.

SPF Community Survey Answers...

Mike Podanoffsky

In early 2010 we sent out emails to all SPF community members inviting them to complete an on-line survey. With about 300 responses so far, we want to share with you a sampling of what we have learned thus far:

What do you want in future Connections? Connections are a way for members of our community to meet and learn. About 1/3 of the respondents have attended a Connection. Several of you noted that Connections were too far to travel or were too expensive. Most of you want Connections to include speakers on research and therapies.

Have you used HSP ListServ or PLS Friends? These are on-line forums where you can get answers from community members to your questions. About 1/3 have used these on-line forums. You can find out more about them on our Web site.

How important to you is our newsletter *Synapse*? About 2/3 of you regularly read each issue of *Synapse* cover to cover. Almost everyone reads something in each issue. So far those responding are evenly divided as to whether *Synapse* should be delivered by mail, by email, or both.

We'd love to add your answers to the survey. If you did not receive an email invitation to complete the survey, it may be that we don't have your correct email address. To tell us where to send your email, go to <http://sp-foundation.org/my-email-is>. To complete the survey (if you haven't already) go to <http://sp-foundation.org/community-survey>. We want to hear what you have to say!

Become a State Ambassadors for SPF

By Linda Gentner VP - SPF

Ambassadors are needed for a few states: MS, ND, OR, SD, UT, WA ... large states need more than one ambassador -- consider being a co-ambassador. You need not live in the state to welcome new members into our community so they do not feel alone. Check out the "Job Description" at http://www.sp-foundation.org/State_Ambassadors.htm.

Don't let the job description scare you away if you are willing to help new people with PLS or HSP. Remember how you felt when you were first diagnosed and thought you were the only one. Contact Linda Gentner with any questions. lkgentner@aol.com

Join us at the 2010 National Conference in Los Angeles for an Appreciation Breakfast for our State Ambassadors!! We want everyone to know that we appreciate our ambassadors. It will be a good opportunity for those who are able to attend to get together and share and ask questions of other ambassadors.

2.912 Mile Fundraiser for SPF

Contributed by Mark Weber



My brother, Michael, has decided to pedal across America to raise \$\$\$ for the SPF. The ride is fully supported by an organization called "America by Bicycle." He will be riding their "America South Fast" course, and is paying for the expenses himself. Mike has been training for months and is in the best shape that he has been in for many years. A friend of his did it in 2008 but was in worse shape and still made it. Mike said "I WILL DO THIS." (He tends to be somewhat determined.)

Ed note: Here's more, in Mike's words:

Alpha!

This spring (April 17 to May 14, 2010) I will be riding my bicycle across the U.S. mainland - from Costa Mesa, CA to Savannah, GA - to raise money for the Spastic Paraplegia Foundation (SPF). (O.K., I'm also doing it for the challenge of riding 2912 miles in 27 days!) Why am I raising funds for the SPF? I am raising funds to support my older brother Mark, one of the co-founders of the SPF, who suffers from Spastic Paraplegia.

PLEASE DONATE WHAT YOU CAN - A LUMP SUM,
OR A CERTAIN AMOUNT PER MILE YOUR ENTIRE
DONATION WILL GO TO THE SPF.

To donate online: www.sp-foundation.org/donate.htm

If you prefer to send a check, please send it to:

The Spastic P.

P.O. Box 1208

Fortson, GA 3

Mahalo (thanks)!

Mike Weber weberm@byuh.edu

<http://rideforspf.blogspot.com>

From the Editor: Here's Mike's itinerary. If he'll be riding near where you live, please plan to cheer as he goes by:

Tour Itinerary		
Day	Location	Day's Mileage
Day 0	Costa Mesa, CA	0
Day 1	Palm Springs, CA	115
Day 2	Blythe, CA	130
Day 3	Wickenburg, AZ	115
Day 4	Cottonwood, AZ	103
Day 5	Winslow, AZ	107
Day 6	Gallup, NM	134
Day 7	Albuquerque, NM	135
Day 8	Albuquerque (REST DAY)	0
Day 9	Las Vegas, NM	135
Day 10	Tucumcari, NM	110
Day 11	Dalhart, TX	96
Day 12	Pampa, TX	112
Day 13	Elk City, OK	97
Day 14	Chickasha, OK	125
Day 15	McAlester, OK	145
Day 16	Ft. Smith, AR	108
Day 17	Ft. Smith, AR (REST DAY)	0
Day 18	Conway, AR	122
Day 19	Brinkley, AR	101
Day 20	Senatobia, MS	113
Day 21	Aberdeen, MS	138
Day 22	Tuscaloosa, AL	110
Day 23	Prattville, AL	117
Day 24	Columbus, GA	115
Day 25	Perry, GA	97
Day 26	Vidalia, GA	104
Day 27	Savannah, GA	128
TOTAL MILEAGE		2912

Events

Sarah Roberts-Witt, Editor

Kansas Connection March 27

Mark Dvorak, Oklahoma Ambassador

I want to say thanks to Mari and Alex White, and Dewey Warkentin-- Mari and Dewey share the role of Kansas ambassadors--we connected in Overland Park, Kansas this weekend (suburb of Kansas City)-- I really enjoyed meeting with them--we visited about many various things and we even discussed getting together again in Wichita, Kansas in the future. Mari's son, (age 7) Alex, is a super sweet little boy. I gave him a ride on my scooter. We zoomed around the hotel--he honked the horn a time or two and when we finished he told his mom-- "It will be past my bedtime by the time we get home, so we better go"--special kid!--all of us could learn from his terrific attitude--you would not even know he is handicapped by the way he acted. We'll keep everybody posted about another Kansas connection!

Upcoming Events

Spring Fling #10 in 2010

May 7-8, 2010

Berkeley Springs, West Virginia

Ronnie Grove, frogrove@verizon.net

Make plans to attend a very special WV Connection with lots of specialists—namely, you. No one knows PLS and HSP better than we do. The rooms at Best Western-Berkeley Springs Inn are filled, however, many other motels are nearby. So mark the date for Berkeley Springs and remember, this is the place where all the women are gorgeous and the men are lucky.

Holes for Wally

May 10, 2010

Wally Chase, wwchase@hotmail.com

Moon Valley Country Club, Phoenix, Arizona

SPF National Conference

May 21-23

Los Angeles, California

All details are to be found on the cover, as well as pages 3 and 4 of this newsletter.

New England TeamWalk

September 11, 2010

Jim Campbell, jimthurza@comcast.net

Location to be decided

Drive "Fore" Spastic Paraplegia Golf Outing

September 20, 2010

Forestage Country Club

Monroe Township, New Jersey

James F. Brew, JBREWI@travelers.com

The Travelers Insurance Company will hold its second Drive "Fore" Spastic Paraplegia Golf Outing.

North Carolina Quest for the Cure and The Magnificent Mile

September 18-19, 2010

Hillsborough St, Raleigh, North Carolina

Sarah Witt, switt@yahoo.com

The NC Quest for the Cure is still being formulated. The 5th Annual Magnificent Mile will be held on Sunday afternoon. 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.

California TeamWalk

for our Cures & Connection Weekend

October 2, 2010

Pleasanton, California

Linda Gentner, lkgentner@aol.com (510) 651-5676, A PLS & HSPers Welcoming Dinner will be held Friday night at the Hilton Pleasanton -- 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.

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.

Living with HSP or PLS

Just a Typical PLS Morning!

Contributed by Arlene in Texas



6am...pull up holding to the rail, waddle down the length of the bed pipe Gene installed, sit in powerchair.

RIDE to the bathroom, pull up holding to the grab bars, do my thing, stand back up holding to grab bars.

RIDE to the dressing table and basin. Wash, fix hair, teeth, etc.

RIDE back to the bed, waddle back down the pipe, sit down and put clothes on, pull back up, waddle back down pipe to powerchair.

RIDE to the kitchen for cup of hot chocolate, check the box of chocolate...empty.

RIDE to the utility room for my "gofer";

RIDE back to kitchen, pull myself up and use the "gofer" on the box that is on the very top shelf, set chocolate down; RIDE back to utility and put the "gofer" back.

RIDE to kitchen, retrieve the chocolate and RIDE to the cabinet and get cup and water;

RIDE to microwave and nuke my drink;

RIDE to the computer and set chocolate down, turn computer on, pick up chocolate and take a sip.

A LOUD sneeze from the den and chocolate is all over shirt and jeans, RIDE to the den and tell Gene who had gotten up early, "follow me, strip these wet clothes off me, and it's all your fault."

Bewildered, he follows me as I RIDE back to the bedroom, get wet clothes off, waddle back down the pipe to the bed and change clothes;

RIDE back to the kitchen, pass Gene sitting in his recliner and tell him to bring me a cup of hot chocolate to the computer! Was it all worth it! Yeah.

Augmentative and Alternative Communication (AAC) Devices and Downloads

Devices:

From Thurza: I use Words + on a Panasonic Toughbook.

http://www.words-plus.com/website/products/systems/f2kliteP_series.htm

The voice I have is "Lauren" Go through your neuro and PCP. Get an Rx for an AAC, then work through a rehab facility where the paperwork will be handled. If you are 65, Medicare covers a lot; then your secondary; finally MDA

will pick up a portion of the remainder. If you're younger, your insurance should also cover it if the Rx is well written. The process can take up to four months, but it's worth it.

Downloads for Your Computer or iPhone:

From Galen: For those of us who are really cheap, there is a free program developed for an ALS type. If you like it you can make a donation, but there is no requirement to do so. It works with several Windows flavors (XP, Vista and 7) and with Linux under WINE, though it is unsupported for anything but Windows. www.etriloquist.com

From Don Wilson: If you have a laptop, a \$99.00 software which includes two AT&T voices is available: <http://www.talkforme.com/products.html>

It works for all Windows series, which means even an older model can be used. And the BEST PART! You can download a 30-day trial. That trial will not include the AT&T voices, but it will give you an opportunity to see it work.

Another free AAC software is called Dasher:
<http://www.inference.phy.cam.ac.uk/dasher/>

From Penny: I have Nextup talker www.nextup.com and I like the voice. I use it on a wee tiny netbook. However, many use an iPhone with the program called Proloquo2go www.proloquo2go.com and love it...small, cheap, easy.



Bank Your Voice

by Alyssa Quintero MDA/ALS Magazine

While there's no easy way to prepare for losing your voice, voice banking enables people with ALS to capture their voices via recordings that can be used on Windows-based computers and alternative augmentative communication (AAC) devices.

To start, you can record your voice on a Windows-based computer using the sound recorder accessory and saving your recordings in waveform (.wav) files. Many people record signature phrases like "I love you," or stories for their children and grandchildren, songs and laughter.

Advances in AAC technology allow some programs to use your prerecorded voice on your speech-generating device. You can import the audio files from your PC, or record your voice directly onto some AAC devices.

Speech-language pathologists (SLPs) suggest thinking about voice banking — in any form — before experiencing any detectable changes in speech.

Please check out www.modeltalker.com

NWU Evaluation of Clinical Data of 233 Patients Reporting Sporadic PLS

By Nailah Siddique

The table below is from an intensive chart review of our "PLS" patients done a year ago. The first issue with any PLS study is to make sure we're looking at only apples, no oranges.

Then the second issue is what combination of relatives do we have to go with these patients. There are currently 3 techniques for looking for genes that increase risk for developing a disease. One is to use patients matched with their parents (both parents). This group is hard to find among PLSSers because of the generally later age of onset of PLS. But we have roughly 25 of them. To use them effectively we would need about 200. Then you can also use a patient matched with an unaffected brother or sister. We've got 127 of those. Again 200-plus is the goal. Lastly you can use stand alone patients compared to controls of the same age, gender and ethnicity. So we've got the best start on that-- if you look at the table. The problem you can clearly see is that you need more patients. With this technique the magic number is about 400. The good part is we have controls gathered from other studies that we could use.

The next issue though, after we get the samples, would be funding to actually do the work. Right now we're gathering these samples using money from a kind of slush fund that we have for general lab expenses. Funding is a very, very big problem in our world right now.

Definite PLS (UMN signs, bulbar involvement, 5 or more years).....	81
Probable PLS (UMN signs, bulbar involvement, 3-5 years).....	48
Suspected PLS (UMN signs, bulbar involvement, < 3 years).....	61
Was or became classical ALS with marked LMN involvement	4
Was or became slow progressing UMN predominant ALS.....	11
UMN signs, no bulbar involvement after 5 years, no family history – sporadic spastic paraparesis	19
Other diagnoses with signs/symptoms not consistent with PLS	9

To reach Nailah and arrange to send your blood samples, either call or e-mail:

Nailah Siddique RN MSN, Clinical Nurse Specialist
Neuromuscular Medicine Northwestern University,
Feinberg School of Medicine (312) 503-2712;
nsiddique@northwestern.edu

Cookin' for a Cure, Volume II

Submitted by Linda Gentner

REMINDER ... your recipes are still needed ... submit your favorite recipes with your local flair!

Several people, especially our newer members, have expressed an interest in a cookbook. We produced our first SPF cookbook in 2003 -- time flies. So our next SPF fund raiser will be Volume II. HSP and PLS members are welcome to submit your favorite recipes.



Natasha Schaff is chairing this project as her way to help her father who has HSP.

E-mail or mail your recipes to: Natasha Schaff, natashaschaff@yahoo.com

3131 Grove Ct, Mandeville, LA 70448 If you have any questions or are interested in helping with typing, please contact Natasha

Jim Sheorn's PT Stretches



Hip Adductor Stretch: (inner thigh muscles)

Standing – walk feet apart wide, can do a small lunge toward each side to isolate one more than the other

½ Butterfly – on edge of bed or stair landing with one leg off, other leg to be stretched is bent at knee (1/2 butterfly) push down on knee – couldn't find a picture but it looks like the "sitting hamstring stretch" below except the straight leg will be bent!

Hamstring Stretch: (back of thigh)



Standing – bend forward at hips and try to touch your toes, keep knees straight! A modified version is below to isolate one hamstring at a time



Sitting: on edge of bed or stair landing with one leg off, other leg to be stretched is straight at knee, bend forward and try to touch toes



With assistance- you lying on your back with one leg stretched out in front of you, the leg to be stretched hip flexed to 90 degrees, press-up on lower leg to straighten knee (you may use a strap with this one as well)

Calf Stretch: (back of lower leg)

Wrap a belt or strap around forefoot and pull back on foot ("toes toward your nose")

Standing – place forefoot on a small object keeping heel on the ground, lean forward at hips

Stretches should be held for at least 20 seconds and performed 3-5 times (preferably twice a day!) :)



The exercises below (strengthening exercises) are done in repetitions building up to 30 at a time consecutively, when this comes easy you can increase the number of sets and/or use ankle weights to add resistance.

EXERCISES



Ankle:

Toe Raises: While sitting bring toes up (moving at ankle) keeping heel on ground, bring them up and down



Knee:

Knee Extension: Sitting on edge of bed or chair, straighten knee and return to bent position, repeat!



Hip:

Straight Leg Raises: Lying on your back raise your leg up and down keeping knee straight



Hip Abduction:

Lying on your back: keeping knee straight, move leg out and in (do not rotate hip outward, keep knee and toes pointed toward ceiling); or lying on back with knees bent use theraband wrapped around thighs and move knees out and in



Lying on your side: lift top leg up and down keeping knee forward

Genetic Alliance Celebrates Announcement of Genetic Tests Registry

Voluntary Registry First Step Toward Transparency and Collaboration in Genetic Testing

WASHINGTON DC- Genetic Alliance applauds the announcement today by Francis Collins, M.D., Ph.D., Director of the National Institutes of Health (NIH), of a voluntary registry for genetic tests offered by labs nationwide. The registry will be hosted at NIH, which will complete a broad consultation with prospective users and submitters before the registry is established.

Genetic Alliance, along with Genetics and Public Policy Center, Coalition for 21st Century Medicine and others, has called for a mandatory genetic testing registry for several years. "This is a wonderful step forward for genetic testing and the integration of genetics into medicine," said Sharon Terry, president and chief executive officer of Genetic Alliance. "We are glad NIH plans to learn from the experiences and expertise of the individuals and institutions that will be using and registering information. We also look forward to the registry becoming mandatory so that we are all apprised of the quality and availability of genetic testing across the nation."

A mandatory registry would enable a forward-looking oversight system that is flexible and nuanced. It would undergird a transformed system that is open, transparent, and coordinated with all stakeholders and agencies, and that balances safety, innovation, ethical and social issues, viability, and the risks and benefits of improved health. Dr. Collins' announcement today is a major advance toward transparency, interagency collaboration and 21st century healthcare.

About Genetic Alliance: Genetic Alliance transforms health through genetics, promoting an environment of openness centered on the health of individuals, families, and communities. Genetic Alliance brings together diverse stakeholders that create novel partnerships in advocacy; integrates individual, family, and community perspectives to improve health systems; and revolutionizes access to information to enable translation of research into services and individualized decision making. For more information about Genetic Alliance, visit <http://listserv.galists.org/t/461205/52768/15/0/>

For more information: Vaughn Edelson, Programs Manager, (202) 966.5557 ext. 213, vedelson@geneticalliance.org

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. Go to www.spf-foundation.org/donate.htm

A SPF Ambassador Was a 2010 Paralympic Domestic Dignitary

Contributed by: Jean Chambers, SPF Ambassador for Canada



The flame has been snuffed and the world has gone home. All of Vancouver is recovering from the best party EVER. Although I had entered the draw to be a volunteer with the 'official' Vancouver group, my name was not drawn so I did my volunteering at our local Community Centre in West Vancouver as we were a venue site with Cypress Mountain. You know the mountain – the one

with scant snow! Our Advisory Committee on disability issues hosted an evening at the ice rink with a sledge hockey demonstration where some of the local sledge hockey players demonstrated their skills on the sledges. They had brought some extra sledges and invited the guests to try them out. There was a one hour wait for a turn! No – I did not take a run.

During the Paralympics, we hosted a large display of some of the pieces of equipment used by the athletes plus accessible equipment available for use in our mountains and on the water for people with various disabilities. These included sit skis, 'no tip' kayaks, and all terrain wheelchairs.

As a volunteer I was appointed to the position of "2010 Paralympic Domestic Dignitary" which gave me a pass to all of the Paralympic events, with the exception of the Opening and Closing Ceremonies and the Gold Medal Sledge Hockey game. Alas, I missed your Gold medal sledge hockey game. But, I did see 2 other games including the Canadian one where Norway beat us for the Bronze medal by scoring the winning goal with 3.6 seconds left in the game. The next day we watched the Gold, Silver and Bronze medal curling where I was helped considerably by one of the wonderful 'blue jacket' volunteers. A quote from her: "my daughter has been in the Special Olympics for years, so this is my pay back time." What a great attitude.



The Paralympic games served as a great opportunity to raise awareness on issues of accessibility and will leave a tremendous legacy for all who want to experience sports with any of a wide variety of disabilities.

Pat Croom

Written by Daniel Vance

Pat Croom reads this column in the *New Bern (NC) Sun Journal*. Not long ago, like so many others, she responded to my request asking readers to send in their personal stories about disability. Due to her disability, Pat was unable to speak for a telephone interview. So with her permission, I have included here an edited version of her email.

She wrote: "I just read your column on disabilities and wanted to thank you for allowing us to share how we live with a disability. I was diagnosed with Primary Lateral Sclerosis (PLS) in June 2000. This is a rare neurological disorder.

My husband and I love to dance. Around 1995, I found myself sitting more than dancing because of cramps in my lower legs. Little did I know this was an early PLS symptom.

In 1998, I began falling due to poor balance and had to use a cane. I started going from doctor to doctor to find out what was wrong. When I received my diagnosis, my sister insisted we move from New Bern to Garner to be near her and her children. She built us an (accessible) home next door to her. After 38 years in New Bern, we moved in 2001.

Now I use a wheelchair. I still have some use of my hands. My voice is gone and I use a [special device] to communicate. I've never asked God why and I'm not bitter. The hardest thing is dealing with how other people treat me. I've lost friends that were once like family. I try to remain positive and thank God every day for my family and church. My devoted husband of almost 50 years makes it possible for me to remain home.

PLS has changed my appearance. My facial expressions have become exaggerated and I've gained weight. But life is good.

I'm part of an online support group, PLS Friends. PLS is often confused with ALS (Lou Gehrig's disease). PLS usually progresses slower than ALS. Mine has progressed fairly fast and affects my legs, arms, speech, and swallowing. I sometimes think I have ALS, but that really doesn't matter. I refuse to go through test after test (and some are painful) to find out. I'd rather enjoy life than dwell on my disability. So I take one day at a time and do what I can. Thanks for letting me share my story."

Getting Work Resources

From the Reeve Foundation's Paralysis Resource Guide

Social Security Administration operates the Ticket to Work and PASS programs. For information call toll-free 1-800-772-1213. Visit the Internet site, www.ssa.gov for details on all SSA programs. Use the Search function on the home page and type in "Ticket" or "PASS" or other topics of interest.

The Rehabilitation Services Administration (RSA) administers grant programs and projects that serve individuals with disabilities in the areas of vocational rehabilitation, supported employment and independent living. RSA, 400 Maryland Avenue, S.W., Washington, D.C. 20202-2551; telephone (202) 245-5482; on the Internet visit <http://www2.ed.gov/about/offices/list/osers/rsa/index.html>

Disability and Business Technical Assistance Centers:
The National Institute on Disability and Rehabilitation Research (NIDRR) has established 10 regional centers to provide information, training and technical assistance to employers, people with disabilities and other entities with responsibilities under the ADA. The centers act as a "one-stop" resource on ADA issues in employment, public services, public accommodations and communications. To locate the center in your region call toll-free 1-800-949-4232; or visit the Internet site www.adata.org

Office of Disability Employment Policy (ODEP) is a federal agency that works to increase job opportunities for adults and youth with disabilities while striving to eliminate barriers to employment. Contact ODEP by way of the U.S. Department of Labor, Frances Perkins Building, 200 Constitution Avenue, NW, Washington, DC 20210; toll-free 1-866-0DEP-DOL, or visit <http://www.dolgov/odep>

The Job Accommodation Network (JAN) is a free consulting service that provides information about job accommodations, the Americans with Disabilities Act (ADA), and the employability of people with disabilities. JAN also staffs the Small Business and Self-Employment Service (SBSES) of the Office of Disability Employment Policy; SBSES provides information, counseling and referrals about self-employment and entrepreneurship for people with disabilities. Call toll-free 1 (800) 526-7234 or visit the Internet site <http://janweb.icdi.wvu.edu>

The National Business & Disability Council is a resource for employers seeking to integrate people with disabilities into the workplace and also for companies hoping to reach them in the consumer marketplace. The National Business & Disability Council, 201 LV. Willets Road, Albertson, NY 11507; telephone (516) 465-1515; www.nbdc.com

The National Center on Workforce and Disability/Adult is funded through the U.S. Department of Labor's Office of Disability Employment Policy (ODEP) and provides training, technical assistance and information to improve access for all in the workforce. The Institute for Community Inclusion, UMass Boston, 100 Morrissey Boulevard, Boston, MA 02125; toll-free 1 (888) 886-9898; www.onestops.info

The National Collaborative on Workforce and Disability for Youth (NCWD/Youth) works to ensure that youth with disabilities have full access to services in order to maximize their opportunities for employment and independent living. NCWD/Youth, Institute for Educational Leadership, 4455 Connecticut Avenue, NW, Suite 310, Washington, DC 20008; toll-free 1-877-871-0744 or see <http://www.ncwd-youth.info>

PASS Tutorial: Cornell University, The Social Security Administration and the New York State Office of Vocational and Educational Services for Individuals with Disabilities have teamed up to create a useful Internet site to provide assistance for the Plan for Achieving Self-Support (PASS). Includes detailed explanation of the PASS program as well as a helpful tutorial on completing the application. Visit the Internet site www.passonline.org

Proyecto Vision: Disabled Latinos face higher rates of unemployment than other disabled Americans and non-disabled Latinos. This project connects disabled Latinos with employment services and related resources and helps Latino organizations to better serve their disabled community members. Offers bilingual services. For more call Proyecto Vision, toll-free 1 (866) 367-5361; or visit the Internet site www.proyectovision.net

The Abilities Fund is the first financial lending institution devoted exclusively to advancing entrepreneurial opportunities for Americans with disabilities. The Abilities Fund, 101 East Van Buren Street, Centerville, IA 52544; toll-free 1 (888) 222-8942; or visit www.abilitiesfund.org

Caregiving

What Defines You?

Contributed by Geoff Kettling

1) HSP or PLS or any other medical matter is just 1 characteristic it is not the whole of your being. Who are you, what are your roles: friend, parent, child, other relative, worker, someone who is tall, not-so tall, young, not-so-young, ancestral heritage, current identified ethnicity, friend, lover, humorist, smarty pants, anything else?

2) How to explain paraplegia/paraparesis and the hidden disability? The majority of all medical conditions are hidden/invisible. If someone states, "You don't look like you have a disability." I may respond, (*though mine is visible*), wow you got really good eyes if you can see cancer (*or other disability*). Paraplegia/paraparesis I explain as partial paralysis it's not full. I have use of my legs and do use them, but can't use them as well as you, yet more than someone who has total paralysis. Similar to the difference between legally blind (*20/200 and/or 10 degree field of view in both eyes*) and totally blind (*no light perception*).

3) For workers, if you need accommodations on the job site or to do essential functions of the job itself, you can approach your supervisor and/or HR. If there is no official documentation or record of your request you have no protection under ADA or ADAAA (*Amended Act being proposed*). Also if you constantly give 125% and that's what your employer will ask of you, but now need to back down to 100% then it's time for a candid chat with your boss and this may be necessary to take care of yourself, and may allow for new staff position if there's enough work. Jobs can be accommodated in many instances, and you can get creative. This can include flex time, swapping duties with colleagues, breaking up breaks differently, etc. Close parking placards, allows me to choose whether or not to use them, whether I am driving or passenger.

4) Exhausted, worn out from trying to walk, run, stand? Here's the pride pill --swallow it. Here's my story: After 2-3 hour monthly shopping at major store X I'm worn out. Now I can roll 5 miles in my wheelchair. I can zip through the stores, the zoos, theme parks, etc... I have energy reserves to do what is necessary, and I'm not grouchy after 3 hours of shopping. Point being, adding a wheelchair to my repertoire improved my quality of life, it didn't decrease it.

I am a person with HSP/a disability. I am living with HSP/a disability. I am not a paraplegic/disabled person. I am not suffering with HSP/paraplegia/a disability. I am a person not a condition, and I am living.

Husbands as Primary Caregivers

submitted by Jim Campbell



Remember the part of the wedding vows that says: "For better, for worse, in sickness and in health..." Well, I still do because for the first 40 years of our marriage my wife Thurza used to refer to me as "For better, for better." Thurza has always been very competent and self sufficient, so in our early years I was the one that thought I deserved the caregiving and support in our relationship as I faced challenges at work, etc.

This gives you a clue of how unprepared I was 10 years ago for her diagnosis of PLS and my evolving role as primary caregiver for her. I have to admit initially I went through all kinds of denial and avoidance including cringing each time her cane accompanied us to a restaurant or another public place. It probably took me several years to come to the realization this disease wasn't conveniently going to disappear and she really needed my help. I am ashamed to admit it, but thinking of someone else's needs first doesn't come naturally to me. Even after 10 years I still tend to forget things she regularly needs help with. I am in admiration of those caregivers who are always on top of the situation and looking ahead to the patient's upcoming needs. My sexist impression is that most of those sensitive, super thoughtful caregivers are women, not formerly pampered husbands like I am. In the last three or four years I am pleased to say, I have gotten over my concern about appearances and now proudly push her in her manual wheel chair or lead the way through a crowd for her power chair. She hasn't let this disease get the best of her and has continued to function in new ways. This is not to say our relationship now is always one of wedded bliss. Far from it! A marriage counselor once said to us that it's okay for Jim to max out occasionally. I'm sure I have taken liberties with the term "occasionally" and probably confused it with "frequently". On the upside over these 10 years, I have discovered that caregiving is actually more fun when you do it cheerfully and willingly with a smile. Like in so many things a smile is contagious so we can both enjoy the moment when I am getting her ready for bed or helping her with pills. So, fellow husbands, it may not be second nature to us but all of us can rise to the occasion and return all the love and care our spouses have given us for so many years and do it with a smile. And don't forget to offer lots of unsolicited hugs. **Happy Caregiving!**

Medical Reports

Betsy Baquet, editor

50% of HSP Cases Caused by Mutations in One of Three Genes

Mutations at over 40 genetic sites cause HSP. However, according to research conducted by SPF Scientific Advisory Board member Craig Blackstone, over 50% of cases are caused by mutations in SPG3A, SPG31 or SPG4. Blackstone, along with colleagues from NIH in Bethesda, MD, have determined that a single mechanism is likely to underlie HSP caused by these three mutations. Specifically, data suggests that the proteins made by these three proteins are all involved in the shaping and cellular interactions of the endoplasmic reticulum (ER). Defects in these processes underlie 50% of HSP cases caused by mutations in these three genes.

SOURCE: <http://health.groups.yahoo.com/group/PLSers-NEWS/message/4770>

Mutation that Causes HSP with Thin Corpus Callosum Also Causes Juvenile Onset ALS

The mutation of the spasticsin gene is the single most common cause of autosomal recessive HSP with thin corpus callosum. Common features between ALS and HSP prompted a study where 25 families with autosomal recessive juvenile ALS and long-term survival were investigated for mutations in the spasticsin gene. Surprisingly, 12 sequence alterations in the spasticsin gene were identified in 10 unrelated pedigrees with autosomal recessive juvenile ALS and long-term survival. The study indicates that mutations in the spasticsin gene could cause a much wider spectrum of clinical features than previously recognized, including autosomal recessive juvenile amyotrophic lateral sclerosis.

SOURCE: <http://brain.oxfordjournals.org/cgi/content/abstract/133/2/591?maxtoshow=&hits=10&RESULTFORMAT=1&andorexacttitle=and&andorexacttitleabs=and&fulltext=spastic&andorexactfulltext=and&searchid=1&FIRSTINDEX=0&sortspec=relevance&fdate=1/1/2010&resourcetype=HWCI>

SPF Funded Scientists Publishes his Research on the Spastin Protein—Basic Growth Factor Enhances Axonal Branching and Microtubule Severing

The SPF awarded Dr. Peter Baas at the Drexel University College of Medicine a research grant in 2007 in the amount of \$132,000. For two years, Dr. Baas has been studying the role and function of the spastin protein, gaining a better understanding of its impact on axonal branch formation and micro-tubule severing and

transport—processes critical to the communication between upper motor neurons and lower motor neurons. Dr. Baas discovered that by adding a growth factor, microtubule severing increased whether or not the spastin or katanin proteins were present. He also observed that when spastin was over expressed, axonal branching, short microtubule transport, and frequency of microtubule ends were also enhanced.

SOURCE: <http://health.groups.yahoo.com/group/PLSers-NEWS/message/4755>

Key Finding on Cause of a Rare Form of HSP

Patients with a rare HSP called SPG5 have mutations in the gene coding for the oxysterol 7 alpha hydroxylase (CYP7B1). It was demonstrated that four patients with the SPG5 disease have 6-9 fold increased plasma levels of 27-hydroxycholesterol. A much higher increase, 30-50 fold, was found in cerebrospinal fluid. If the accumulation of 27-hydroxycholesterol is an important pathogenetic factor, a reduction of its levels may reduce or prevent the neurological symptoms.

SOURCE: <http://health.groups.yahoo.com/group/PLSers-NEWS/message/4752>

Mutation of FA2H underlies a complicated form of hereditary spastic paraparesis (SPG35)

Using samples from a large Omani family, British scientists mapped a gene for a novel autosomal recessive form of HSP (SPG35) in which the spastic paraparesia was associated with intellectual disability and seizures. Magnetic resonance imaging of the brain of SPG35 patients showed white matter abnormalities suggestive of a leukodystrophy. Mutations in the fatty acid 2-hydroxylase gene (FA2H) were reported in the original family with SPG35, as well as a Pakistani family with similar clinical features. These results demonstrate that mutations in FA2H are associated with SPG35.

SOURCE: <http://health.groups.yahoo.com/group/PLSers-NEWS/message/4751>

First U.S. Stem Cells Transplanted into Spinal Cord

For the first time in the United States, stem cells have been directly injected into the spinal cord of a patient. Doctors injected stem cells from 8-week-old fetal tissue into the spine of a man in his early 60s who has advanced ALS. It was part of a clinical trial designed to determine whether it is safe to inject stem cells into the spinal cord and whether the cells themselves are safe.

Longtime ALS researcher and University of Michigan neurologist Dr. Eva Feldman is overseeing the first human clinical trial of a stem cell treatment in ALS patients. “We are entering a new era of cell therapeutics for ALS, and in my opinion, it is a new era of hope for patients with ALS,” Feldman said.

There will be a total of 12 patients involved in the study, receiving stem cell transplants at Emory University in Atlanta, Georgia. The first of these patients received a transplant into the lumbar area of his spine, which is the area that controls leg function. Unlike the controversial use of embryonic stem cells, which destroys the embryo, these transplants will use neural stem cells from an eight week-old fetus.

These neural stem cells will not cure the disease and will not grow new motor neurons. Instead, they are intended to protect working motor neurons and significantly slow down disease progression.

Lucie Bruijn, Science Director of the ALS Association says that injecting stem cells into the spinal cord -- in the region where the motor neurons are located that affect ALS – is a breakthrough. But she cautions that this is only the first step in the first part of this clinical trial. It's too early to draw any conclusions about the effectiveness of this treatment, especially since the trial has only just begun.

SOURCE: <http://www.cnn.com/2010/HEALTH/01/21/stem.cell.spine/index.html?iref=allsearch>

Covington, Louisiana—Groundbreaking Stem Cell Research Being Done (3/26/10)

Dr. Gabriel Lasala, Medical Director of TCA Cellular Therapy, is leading a team of scientists in stem cell research that could give hope to thousands living with ALS. The research being done could transform Covington, Louisiana, into a global medical magnet.

We are the only company that is utilizing adult stem cells in ALS. The only company in the country,” says Dr. Lasala. World renowned stem cell biologist Dr. Jose Minguell left Chile to join Lasala’s team. The process involves taking a patient’s bone marrow, separating the stem cells, growing them from thousands into millions and then reinjecting them to regenerate damaged tissue.

Because of Lasala’s success rate in other protocols, the FDA has granted expanded access to the ALS clinical trials, meaning more patients will be able to receive treatment. The process is moving quickly. A commercial product using stem cells to treat ALS could be on the market in as little as three years.

SOURCE: <http://www.msnbc.msn.com/id/36044117/>

ISIS-SOD1-Rx Anti-Sense Drug—Human Trial Commences on Patients with Familial ALS

Human testing has begun of ISIS-SOD1-Rx, an experimental “antisense” drug designed to block production of toxic SOD1 protein in people who have developed ALS because of a mutation in the SOD1 gene. Antisense compounds are pieces of genetic information that keep other genetic information from being processed. The study represents the first time a drug designed to block toxic SOD1 has been administered directly into the central nervous system of humans.

Drug developer Isis Pharmaceuticals has begun tests of the compound in 32 people with the SOD1-related form of the disease. Investigators at six U.S. sites will use an external pump to administer 12-hour infusions of ISIS-SOD1-Rx directly into the fluid that surrounds the brain and spinal cord. The delivery method, called “intrathecal injection,” is expected to effectively target the cells that produce the toxic SOD1 protein.

The trial will assess the safety, tolerability and pharmacokinetics (the ways a drug works in the body) of the antisense compound in people with the disease. According to Timothy Miller, investigator at Washington University in St. Louis, “It is evident that certain cases of familial ALS are related to mutant forms of SOD1. Therefore, the selective inhibition of SOD1 production could provide a way to improve the outcomes of these patients with ALS.” C. Frank Bennett, senior vice president of research at Isis Pharmaceuticals says, “This study is the first step in demonstrating the applicability of antisense drugs to treat severe neurodegenerative diseases.”

SOURCE: <http://quest.mda.org/news/als-sod1-trial-%E2%80%98watershed-moment%E2%80%99>

Gladstone scientists identify role of key protein in ALS and frontotemporal dementia

Scientists at the Gladstone Institute of Neurological Disease (GIND) have identified the reason a key protein plays a major role in two neurodegenerative diseases. In the current edition of the *Journal of Neuroscience*, researchers in the laboratory of GIND Associate Director Steven Finkbeiner, MD, PhD, have found how the protein TDP-43 may cause the neurodegeneration associated with ALS and frontotemporal lobar degeneration with ubiquitin-positive inclusion bodies (FTLDu). TDP-43, is the major component of protein aggregates in patients with these diseases. Mutations in the TDP-43 gene are also associated with familial forms of ALS and FTLDu.

Under normal circumstances, TDP-43 is a common protein that stays mostly in the nucleus and has several beneficial functions. However, in patients with ALS and FTLDu, TDP-43 is redistributed from the nucleus to the cytoplasm and forms insoluble TDP-43 aggregates in the nucleus, cytoplasm, or neuronal processes.

The team has developed a technique to see the location of the TDP-43 in rats to find out how this protein might be involved in neurodegenerative diseases. Using an automated microscope that can examine hundreds of thousands of neurons individually over several days, scientists used sophisticated statistical analyses to follow the fate of each individual neuron and determine its risk of death at any given time.

The researchers found that the mutant TDP-43 was toxic to neurons and that more of it was found in the cytoplasm. The amount of cytoplasmic TDP-43 was a strong and independent predictor of neuronal death. "Our results indicate that the mutant protein is mislocalized to the cytoplasm," Finkbeiner said. "Although we don't know the underlying mechanism, the protein seems to become toxic in the cytoplasm and then causes death of the neuron."

SOURCE: http://www.eurekalert.org/pub_releases/2010-01/gisi011210.php

New ALS drug slips through telling 'Phase II' clinical trials

Scientists report after completing a Phase II clinical trial that a drug in a family of anti-anxiety agents has potential to slow the muscle weakening that comes with ALS. A phase II trial is an early, small-scale test to show if the drug works and continues to be safe.

A report online December 4 in the journal *Amyotrophic Lateral Sclerosis* says the drug talampanel showed some ability to slow the loss of major daily life activities such as speaking, walking and dressing that typically slip away as the disease progresses. The drug is a member of the benzodiazepine family - anti-anxiety and muscle-relaxing agents that work in the brain and spinal cord.

The study, by a scientific team from Johns Hopkins and Indiana University, reveals there's enough benefit from this new use of talampanel to propel it into larger trials that will definitively tell its worth. "The research demonstrates that talampanel appears able to slow the progression of disabling ALS symptoms," says Johns Hopkins neurologist Jeffrey D. Rothstein. "The effect isn't overwhelming at the dosage of medicine used

in this early, very small trial," he adds. "Still, having promising human data is reason enough to keep it in the drug pipeline where we can really find out where it stands for patients."

Several facts about talampanel make it especially attractive to try as a possible therapy, Rothstein says. The drug's talent is its ability to block specific receptors on ALS-vulnerable nerve cells that are docking sites for the neurotransmitter glutamate. Excess glutamate causes excitotoxicity—a process that can kill the motor neurons that enable movement. Earlier studies have confirmed excitotoxicity as a cause of damage in ALS animal or cell models, as well as patients. Levels of glutamate are elevated in as many as 40 percent of patients with ALS whose disease appeared to arise spontaneously.

Currently, a large international trial of talampanel is under way, due to end in 2010.

SOURCE: http://www.eurekalert.org/pub_releases/2010-01/jhmi-nad010410.php

miRagen announces discovery of key microRNA implicated in Lou Gehrig's disease

miRagen, a biopharmaceutical company that develops innovative microRNA-based therapeutics for cardiovascular and muscle disease, announced the publication of data demonstrating that microRNA-206 (miR-206) plays a crucial role in the progression of amyotrophic lateral sclerosis (ALS or Lou Gehrig's disease) and in neuromuscular synaptic regeneration in mice. The findings, published in the December 11 issue of the journal *Science*, reveal miR-206 as a potential target for the development of therapeutic agents for the treatment of neuromuscular disease. The study was conducted by researchers led by Eric N. Olson, Ph.D., at the University of Texas Southwestern Medical Center.

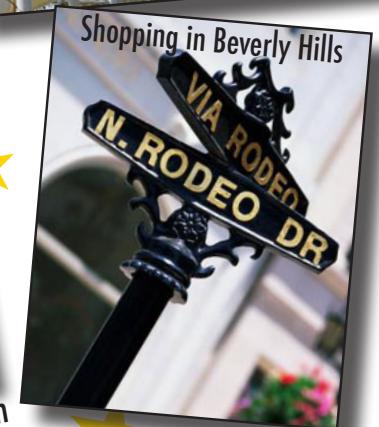
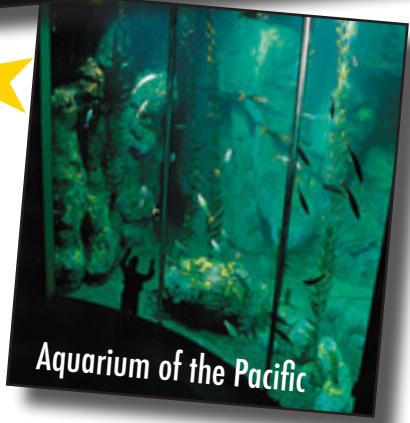
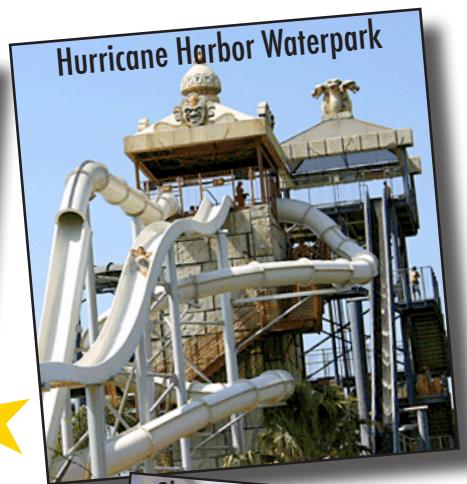
"We are, quite frankly, excited by these findings," said Dr. Olson, Chairman and Professor of Molecular Biology at the University of Texas Southwestern Medical Center and miRagen's Chief Scientific Advisor. "One of the hallmarks of ALS and other similarly degenerative muscle diseases is the inability of the neuromuscular synapse to transmit the impulse that leads to muscle contraction. Our data show that miR-206 plays a central regulatory role in this process. We view this as an extremely promising target for therapeutic intervention."

SOURCE: http://www.eurekalert.org/pub_releases/2009-12/mti-mad120909.php



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