

Synapse

Autumn 2006

Serving the Primary Lateral Sclerosis Community since 1997
Welcoming the SP Foundation since 2003

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There is lots of SP material for sale at <http://www.sp-foundation.org/store.htm> PLS and HSP sweatshirts, tote bags and ornaments available. Just scroll down to Cafe Press.

Now that the holidays are quickly approaching, don't forget about the SPF Shopping Mall. It doesn't cost you anything extra to use it and SPF receives a commission. Please remember to tell your friends and family about it too!

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

SP FOUNDATION

Shopping with SP

SP magnets for your car (\$5) and the '06 TeamWalk T-shirts (\$13 including shipping) are also available by sending your check made payable to SPF to: Linda Gentner, 1605 Goularte Place, Fremont, CA 94539.



Magnet



T-Shirt

The SP notecards are only \$5 a box if you also order a t-shirt.

President's Message – Annette Lockwood

With TeamWalks being held across the country and our National Conference in Long Island, it has been a very busy time of year. In August, Carolyn Sartain-Anderson held a SPF Benefit Dinner in Southampton, NY to celebrate turning 60. Thanks to Carolyn for raising over \$5,500 at this event.

SPF SPASTIC PARAPLEGIA FOUNDATION, INC.  Research Education Support

Lead Sponsor of the Synapse Newsletter
The Spastic Paraplegia Foundation is dedicated to finding the cures for Primary Lateral Sclerosis and Hereditary Spastic Paraplegia through research funding, information and support programs.

<http://www.sp-foundation.org> ~ e-mail: info@sp-foundation.org
Phone (703) 495-9261

Another exciting event was the hiring of Cathy Wilkes as the SPF Executive Director effective September 1, 2006. Cathy worked for the American Heart Association for 15 years so brings a wealth of fundraising experience to help SPF move to the next level. I also had the opportunity in September to represent SPF at two meetings in the Washington, DC area. In mid-October, the first New Moves Retreat for women with HSP and PLS is being held in Tampa. It will provide an opportunity to make new friends, have fun and explore concerns that matter to women with a walking disability. After the Retreat a TeamWalk Connection will be held.

Through September 2006, we have raised close to \$300,000, the breakdown by major initiative is:

\$ 25,500 – **Year End Appeal Letter**
\$ 18,700 - **Memorials**
\$ 19,400 - **Annual Report/Membership Drive**
\$180,400 - **TeamWalks**
\$ 12,000 – **RGM Golf Classic Memorial**
\$ 9,200 – **Pennies**
\$ 12,244 – **ExxonMobil Workplace Giving**

TeamWalk donations are still coming in and two TeamWalk Connections are yet to take place – Tampa (mentioned above) and Raleigh, NC.

National Conference/TeamWalk

My husband and I went to Long Island for the National Conference and TeamWalk. Betsy Baquet did a fantastic job organizing both the Conference and the TeamWalk. We were honored with speeches from Dr. Fink, Dr. Gudesblatt and Dr. Lauring in the morning. The group was then presented with a physical rehabilitation segment by Dr. Craig Rosenberg, Director of Physical Rehabilitation, and James Megna, P.T. and Vestibular/Balance Specialist. Helpful information, technology and case studies on physical therapy options, spasticity management, Botox therapy, and balance therapy were presented. Angela Scicutella, Neuro-psychiatrist, began the next segment, which focused on the psycho-social issues of living and coping with HSP and PLS. Breakout groups were then formed for patients and caregivers/family members. The sessions were led by Drs. Deb Benson and Jean Elbaum, both renowned psychologists in the field of neuro-rehabilitation. The turnout for these sessions was amazing, and the feedback received was wonderful. Efforts are underway to post key slides from the presentations on the website. A special treat at the conference was hearing the original song

entitled, “We Can Move,” written and performed by Mary Prevost, a voice instructor who has HSP. Unfortunately Mary was unable to join us in Long Island so she went to a studio and recorded it for us. The song was combined with a slide show of past SPF events and pictures of those in our patient community who have passed on. Those who attended the TeamWalk held in Eugene, OR were privileged to hear Mary sing her song live. Mary resides in Corvallis, OR.

The National TeamWalk was held at Wantagh Park on a delightfully sunny day. Betsy, with help from her family and friends, hosted over 100 attendees. The children were entertained with face-painting, tattoos and crafts, while adults surveyed the raffle items.

TeamWalk t-shirts (\$10 plus \$3 for shipping) and SPF magnets (\$5 each - similar to the Support Our Troops magnets) will be available for purchase on the website or contact me at 703-495-9261. Help raise awareness with these SPF items.

Life in Motion – Movement Disorders Patient Summit – September 14, 2006

I attended the first Movement Disorders Patient Summit held in Washington, DC on September 14, 2006 along with more than 50 patients, advocates, organizations, health care professionals, media and congressional representatives. The purpose of the meeting was to discuss ways of improving care for the more than 40 million Americans affected by movement disorders – that is more than twice the number of people with diabetes and more than four times the number of those surviving cancer. An opportunity that We Move presents is the prospect of collaborating with other non-profits on a common cause.

A Letter Writing Campaign was initiated by We Move to broaden awareness about movement disorders and set the stage for improved patient care among Congressional representatives and increase media interest. More than 4,000 Americans from every state reached out to their Congressional representatives and educated them about movement disorders via the *Life in Motion* Letter-writing Campaign. Congressman Danny Davis (D-IL), the keynote speaker at the Summit and sponsor of legislation to designate October as national Movement Disorders Awareness Month, spoke about the struggles people with movement disorders experience while seeking a diagnosis.

NIH – Showcasing Success in Neuroscience Research – September 28, 2006

The National Institute of Neurological Disorders and Stroke (NINDS) held this forum for Non-profit Organizations. The day was filled with presentations ranging from Clinical Research Update to How can we work together? To address some of the issues affecting the efficacy of Clinical Research Trials, the Clinical Research Collaboration (CRC) was created. The goal is to increase the number of certified clinical research physicians and available patients. The CRC website (www.nindscrc.com) will serve as a communications hub, where patients, doctors, and the public can search for trials in progress, trials being planned, and ongoing research. Additional information regarding participation can be found on the website or by phone 1-800-305-7811.

As the NINDS budget will basically remain at the 2005 level through 2007, non-profit organizations were encouraged to identify clinical needs, find common themes across diseases, provide seed money, attract new investigators and exchange information. Working together will help to avoid redundancy and assist in the development of the NINDS plan.

Executive Director Hired for SP Foundation

From the SP Board of Directors

Please welcome Cathy Wilkes to the Spastic Paraplegia Foundation. Effective September 1, 2006, Cathy will join SPF as our Executive Director. Cathy worked for the American Heart Association for 15 years so brings a wealth of fundraising experience with her. As the Senior Vice President of the AHA Southeast Affiliate, Cathy was responsible for operations in Tennessee and Alabama with an annual fundraising goal of nine million dollars. Cathy is located in Nashville, TN. She will attend the National Conference/ TeamWalk in Long Island. Cathy was impressed with the results achieved thus far by the SPF community as an all volunteer organization. She is anxious to work with us to raise awareness of the SPF mission and significantly increase our revenue. Please join the Board of Directors in welcoming Cathy to SPF.

Comments from Cathy Wilkes, our Executive Director

I've been employed with the Spastic Paraplegia Foundation for just six weeks and I continue to be amazed at the work being done, not only by the scientists I have met, but by the volunteers behind the scenes.

TeamWalks are being held in so many cities which translate into lots of volunteer hours. Golf tournaments are being played and a Women Conference is being held. All inspired by volunteers. The scientists I have met are working with worms and fruitflies to conduct their experiments. They are excited about each and every process they undertake. With all of this taking place I can truly say how proud I am to be a part of such an amazing organization. The passion instilled by each volunteer and scientist is the reason for the many successes happening now and for all to come in the near future.

The SP Foundation Announces the Recipients of its 2006 Research Grants.

John K. Fink, M.D. Director of the Neurogenetic Disorders Clinic and Professor of Neurology at the University of Michigan was awarded a grant for his proposal entitled "Developing treatment for childhood onset hereditary spastic paraplegia (SPG3A HSP)". Dr. Fink is recognized as the world's leading expert on HSP. He has twenty published medical journal articles on HSP that describe the advances that he has made in understanding the causes of HSP. He was also a major force in organizing two international conferences—one on HSP and one on PLS. Dr. Fink's grant is a two year grant that will total \$120,000.

Nina Tang Sherwood, Ph.D., Assistant Research Professor at Duke University was awarded a grant for her proposal "Understanding the ameliorative effects of temperature in fruit fly models of AD-HSP". Dr. Sherwood has developed a fruit fly model of HSP caused by mutations in its Spastin gene, and discovered much about the role of the Spastin gene product in fruit flies. She first published research about this work in 2004.

Dr. Sherwood was also awarded a two-year grant that totals \$120,000.

Jeffrey D. Macklis, M.D., D.HST, and Paola Arlotta, Ph.D. were awarded a one year grant totaling \$125,000 for their proposal entitled "Molecular Controls over the Development, Connections, and Survival of Upper Motor Neurons". Their 2006

proposal enlarges the scope of their 2005 proposal that the SPF funded last year.

Dr. Jeffrey Macklis is an Associate Professor of Neurosurgery, and the Director of the Harvard Stem Cell Institute, as well as the Massachusetts General Hospital – Harvard Medical School Center for Nervous System Repair. He has written approximately 85 articles regarding his work in major scientific journals. He and his team have made numerous major discoveries in the use of stem cells to grow upper motor neurons. In fact, some of their work has shown that the scientific assumption that upper motor neurons could never be regenerated was wrong!

Paola Arlotta, Ph.D. is an Assistant Professor at the Harvard Stem Cell Institute and the Massachusetts General Hospital Center for Regenerative Medicine. Dr. Arlotta has published papers in major scientific journals describing the advances that she has made working with Dr. Macklis and others regenerating upper motor neurons.

SP Ambassadors for Expanding Horizons

Contributed by Kathi Geisler kathigeisler1@aol.com

Ambassadors have spent time working in their state list of contacts to get organized and let their group know we have a network in their state. We are hoping we can build a Medical Resource List if patients share with us the name of their neurologist to help others. If you do not have internet access, please call the SP President @ 703-495-9261.

AZ, Erika Annis, ArizonaMommyOf3@aol.com

CA, Linda Gentner, lkgentner@aol.com

CT, Mark Weber, markw732@yahoo.com

FL, Flora and Doug Brand, Belgium46@aol.com

GA, Michelle Mobley, kabi2and10@yahoo.com

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PA, Liz Nussear, lizout@aol.com

SC, Cece Russell, cecesc@yahoo.com

SD, Jerry Berge, jebe12@peoplepc.com

TN, Jim Sheorn, jmsheorn@comcast.net

TX, Marlene Doolen, MDoolen512@aol.com

VA, Annette Lockwood,

annette.lockwood@verizon.net

WA, Stephanie Mudd, StephanieMudd@msn.com

Canada, Jean Chambers, jeanchambers@shaw.ca

EVENT REPORTS

August 26 - Tennessee TeamWalk in Nashville

organized by Jim Sheorn, SPF Board Member

The Lunch Connection was held at the Corner Pub and the TeamWalk was held at the Centennial Park

"The Nashville TeamWalk was small but so nice.

Enjoyed lunch with everybody! Chelsea led the walk, with Eddie coming last on his scooter (he says so he could pick up any stragglers--but Jim said it was for another reason--which shall remain within the group)!!!"--Marcia

"The get-together was good. The fellowship of those who suffer with this stuff is so inspiring to me personally. One new attendee was 14 year old Chelsea." --Eddie

September 8-9 - Ohio TeamWalk in Toledo

Contributed and organized by Moira Franchetti

The Connection and TeamWalk were held at the Wildwood Metropark

The TeamWalk Connection went really well! We had 15 people at our Friday Arrival Dinner and twenty at our TeamWalk the next day.

A warm evening in early September of 2006 saw a unique gathering of individuals converging on Bravo Cucina Italian in Toledo, OH. In attendance at our SP 2006 Arrival Dinner were: Moira Franchetti and her husband Michael and children Brynn and Timothy, Kinda and Brad McBride with their children Anna and Ian, Jerry Whipple and Charlene, Dusty Rothman and her son Peter, Tim and Matt Kolhoff, as well as Karen Johnson from our SPF Board of Directors! We all enjoyed an evening of great Italian fare and all the stories of sharing.

The following day we were joined at Wildwood Metropark by Trudy and Caitlin Desmond, Tim's wife and son and daughter, Christine Paet and a friend, and

another Kolhoff brother. We all enjoyed a social hour or so with bagels and fruit courtesy of Meijer, water and Tim Horton's coffee. After socializing a bit we set out on our Walk along Wildwood's multipurpose trail, a nice smooth walk for us 'funny walkers'! All in all I hope I can say that truly, "a good time was had by all!"

September 9 – Georgia TeamWalk in Thomasville

Contributed and organized by Jane Anne King

The Lunch Connection was held at the Plaza Restaurant and the TeamWalk was held at the YMCA. Let me begin by saying - no we did not make a million dollars - but what we did make will help for a cure. We had 33 people at the luncheon and 27 that walked. It is so good to meet new people and some had never met anyone with our diseases. Money is still coming in but we raised several thousand dollars towards Research Awards!

We really had a good time. The raffle, auction and music were fantastic. The place we walked was beautiful we had trees, paved walking trail, etc. My friend Glenda and her husband and daughter who has MS went out to the park and set everything up for us. I do not like to be on TV but had to. Kate Dyer from Channel 6 interviewed me, Ruth and Ben. You always hear that TV puts 10 pounds on you. Well, I looked like the jolly white snow women! We were on the 6:00 news and again on the 11:00 news. They went out of their way to help because they had never heard of this disease. Next year they are going to contribute to the cure.

Our guest speakers were great. Candy Miller, PT, talked about physical therapy and gave us papers on exercise plus many other papers on our disease. Michael Hanna, PT, did a great job on hippotherapy. Everyone wants to do this now! Lt. Rachel Denmark spoke on handicap parking and helped with a lot of questions.

We had a lot of help. The restaurant gave us a room that cost \$150.00 for free and the YMCA charges \$50.00 for clean up and we got that free. CVS Pharmacy gave us drinks free. Blue Bell Ice Cream gave us 100 pops for free. Two of the restaurants gave us coupons for chicken nuggets and ham sandwiches, hamburgers free. Channel 6 Free, Channel 27 Free, Channel 10 Free, CNS Scroll Free. Newspaper Free, Radio Free. I sent out thank you notes and called many of them and they told me how much they appreciate that I called. Most people do not thank them or tell them they did a good job. Next year we

are going to have a concert by Jerry Simmons, Tim and James. Probably on a Friday night and have the TeamWalk on a Saturday.

This is the most gratifying job that you could do. I am 67 years old and if I can do this then the younger ones and older ones can do it. Your aches, pains, depression and being overwhelmed by husband, wife and children demands will disappear and you only have time for this. You will finally feel like a NORMAL human again.

We need to have each and everyone of you to have a TeamWalk. We had 5 people help on this so get your people together. We need this in EVERY state and then we need it in every town. We cannot get a cure if we do not work for it. So don't sit back and say well maybe next year I will do this. I just know how this has made my group feel and we are on a HIGH. So, please help us find our cures!

September 10 - Annapolis, MD

Organized and contributed by Shannon Gregory

On 10 September 2006, SPF held it's first-ever Annapolis, MD TeamWalk. The walk was a great success, drawing in approximately 30 participants from throughout the East Coast, Buffalo, Maryland, Virginia, West Virginia and South Carolina, and raised close to \$6,500.00. The event was kicked off with a registration and opening remarks by Annette Lockwood. The group then took a casual walk, some accompanied by their furry friends, while Matt Gregory and Jacquie Thompson stayed back to cook up a great meal. Julie Thompson spoke on the importance of physical activity and demonstrated some physical therapy exercises while everyone ate. Our silent auction went great as well. I would like to personally thank each and every person who attended the event and those who helped from a distance. It could not have happened without you all. I would also like to thank all of those who gave so generously to support our cause, through financial gifts, as well as items donated. I would especially like to thank Quiet Waters Park for allowing us to host our event at the park, and Harris Teeter Grocery Stores for donating gift cards to purchase the food for the event. It was absolutely wonderful to meet everyone and hear everyone share their stories. I hope to see all of you again soon!

September 15-17 - National Conference and TeamWalk Long Island, NY

The complete article is in the SP Foundation section of this issue.

September 15-16 - California TeamWalk and Race for a Cure in Pleasanton

Organized and contributed by Linda Gentner

The CA TW was a success with absolutely the most perfect weather anyone could have ordered. Friday evening, a few of us met for dinner at the Hilton Hotel where our out-of-town guests were staying. People at dinner were Ken & Julie Auer, a couple who are friends of the Auers, Jean Chambers and Joanne Reyerse from Canada and Craig & Linda Gentner. Larry & Brenda Asbury had a motor home mishap so they missed dinner with us.

Bright and early on Saturday morning, about 30 people showed up to participate in the *Race for a Cure* that our church was sponsoring. Many volunteers were registering runners, others were at the water stations along the race route and others were timing the runners on their return. They even had police escorts. The Race committee had hoped for a larger turnout but now they are saying “next year”. It was quite a learning curve. I was at the front to yell “go” and the church bells rang, and the runners were off and running a 10K race. It was repeated 15 minutes later for 5K people to start their walk/run.

Around 10:00 a.m., the SPF community, along with family and friends began to gather. We were about 55 people strong. We were greeted with pastries again this year donated by Sunrise Bakery, juice and coffee to ready us for our arduous walk. Brocchini Farms donated Apple Bites which were a big hit especially with the health-conscious runners.

After registration and socializing we headed out on our walk or stroll or roll... Linda led the way this year in her Jazzy power chair. Kris Brocchini was treated to ride in a golf cart. We casually took in the quaint shops and Farmer’s Market in downtown Pleasanton. It’s nearing election time so the Mayor of Pleasanton was out and she wanted to know what we were all about. She was interested in who all these people were in their brightly-colored spiffy TeamWalk shirts walking down “her” sidewalks. On our return, we had a very nice catered lunch waiting for us, gourmet sandwiches and even miniature veggies flown in from Ohio—quite a treat!

After lunch Dr. Cathy Lomen-Hoerth, Director of the ALS Center at UCSF spoke briefly. She stressed how important it is for us to participate in research studies. She also praised SPF and said how our research grants

are very sought after. Dr. Cathy is my neurologist and I have participated in several studies with her. The Clinic Nurse, Dallas Forshew, was also in attendance. Several of our people with PLS and HSP talked to Dr. Cathy and Dallas individually. Ken Auer, Bob Bain and Kris Brocchini, who were in attendance, are also patients of Dr. Cathy.

We then held our raffle. I had many people shoving money at me and I kept putting it in my bag. When I counted, just the cash, there was \$823 for the raffle. We had many happy winners. We had one very happy person taking the money! The *Race for a Cure* may not have had many runners but they bought lots of raffle tickets!

September 15- 16 – Oklahoma TeamWalk

Contributed by Marleen Doolen

The SPF 2006 Oklahoma TeamWalk was held Saturday, September 16 in Norman, Oklahoma with 25 attending the event. In its second year, Mark Dvorak did an excellent job in planning and hosting the event. A delicious meal at Johnny Corinos Italian Restaurant was the meeting place Friday evening to begin the weekend. Saturday morning 25 participants gathered at Andrews Park for the TeamWalk with the majority participating in the walk. Freedomtown Scooter Company generously donated some electric scooters to be used for the walk. When the walk was completed, a delicious lunch, prepared by Abby Road Catering, was delivered to us at the park. The lunch was donated by one of Mark’s neighbors, the Browns, and it was great. Our guest speaker, Mike Bumgarner, closed the TeamWalk with interaction in the group asking questions about what we were concerned about dealing with HSP or PLS. Saturday evening dinner and visiting was enjoyed at Santa Fe Cattle Co. and then the Holiday Inn reception area.

From Mike Church:

We really enjoyed the walk. It was my first time meeting and visiting with others who are dealing with HSP. We will be back again next year.

From Mark Dvorak, organizer:

I am so pleased I was able to get together with old acquaintances and to make new ones also. Although it was windy and warm we all had a great time. The kids really enjoyed the fans and squirt bottle fans that Betsy sent us. There were 25 people that attended and they came from Missouri, Texas, and Oklahoma. I am very thankful for all of the goodwill that was spread. The weekend was great and I hope to do it again next

year. Thanks to everyone that helped raise funds for medical research!

September 17 - Colorado Connections Meeting

Organized and contributed by Greg

Dennis Morales provided our meeting location - St. James Meeting Hall.

We had 9 participants, and our guest speaker was from Youcantoocan Inc.

(www.youcantoocan.com). Dennis goes by their store almost daily in his work activities, and went in to check it out. Youcantoocan is a company that provides items to assist with a more independent life dealing with aging and disability issues. They have their store, as well as a catalog and online opportunities. She brought along some of the items that are sold that might be useful in our situation. She had gone to the SPF Foundation website to first learn about PLS and HSP.

Beyond this we had our general roundtable type of discussion, how's everyone doing, what's new, have we found anything to help with life's little gifts.... Also, we shared some pizza....

September 23 – Oregon TeamWalk in Eugene

Hosted by Karen Long

Our first TeamWalk walkathon to raise funds for research for HSP/PLS was quite successful. The event was held at Alton Baker Park in Eugene, Oregon on a beautiful sun filled day. Approximately 40 people were in attendance and were able to choose a 3 mile or a 6 mile walk along the Willamette River. After the walk we had snacks, gave away some great gifts and sang our theme song, " We Can Move" with the writer Mary Prevost. It was wonderful to make new friends and bring more awareness to HSP/PLS and our effort to find a cure.

Special thanks to local businesses for their support: A Beneficial Massage, Emerald Produce, Fiddler's Green Golf, River Ranch Restaurant, and Signs Now. With everyone's help \$3209 was raised.

October 5-6 Autumn in Carolina

Conceived, organized and contributed by Don Wilson

The first participants checked into the motel and joined Bettie Jo (PLS) and me for a meal at the Mayflower. Martin (PLS) and Mary Ann Beckner, Gary (PLS) and Robin Duke, Barbara (PLS) and Arthur Neely, along with Lynn Petch (PLS) and friend Debbie Rollins. Our daughter, Tracy, drove in from

Atlanta and joined us. Bettie Jo's care provider Traci Stanley also was present for a full and fun meal.

Rain was still lightly falling as everyone gathered at Kingswood United Methodist Church fellowship hall.

The Friday arrival group was joined by Jean Mills (PLS), her sister, Doris White and caregiver, Mabel Lee; Cece Russell (HSP); Judith West (PLS) with spouse John Skau; Sarah Roberts-Witt (PLS) and friends Kim Black and Kim Harmon; Dana Brower (PLS); and Bettie Jo's sisters, Mary, Janna and June.

The count was nine PLSers, one HSPer, and fifteen caregivers, spouses, friends, and relatives. Old friendships were renewed and new friendships forged, especially by Dana, Lynn, and Sarah, each of whom had never met another PLSer.

Krispy Kreme Doughnuts and fresh-from-the-oven Orange Glazed Sticky Buns sometimes make talking a challenge, but soon everyone felt at home, and conversations filled much of the morning. Tracy Wilson, a trained massage therapist conducted a workshop on hand and feet massage. Bettie Jo took home the best feeling hands as she was the "subject" for the demonstration.

The catered box lunches from Mrs. Pumpkin's hit the spot, and after everyone had finished, Don led a "Thorns and Roses" workshop. Each participant and their caregiver were given a red card and a blue card.

The were asked to write the ONE thing that bugged them the most about the other, and the ONE thing that really made them see red. Names were not included on the cards. Each card was read to the group and discussed. As the session progressed, the general comment about the caregivers followed the lines of they do too much, over protect and sometimes are impatient. On the good side, caregivers were considered always there, cheerful, and inspirational. Caregivers considered their "charges" (PLSers and HSPers) of worry too much, sometimes become frustrated when trying to communicate, and sometimes impatient. The charges were most impressed by the dedication of their caregivers who are always there, demonstrate great patience, and make them laugh. The session seemed to be interesting but a bit long.

During the last session, SAWCAR (Scooter and Wheel Chair Association of Racing) founder and promoter of the scheduled afternoon races had been watching the weather. Rain and ceased but the wind was blowing fiercely. After discussion with competitors and caregivers, the decision was made to hold the races quickly and then return to the fellowship hall for presentations. Each racer, three

scooters and five power chairs, was presented with shirt and cap of a current NEXTEL Cup driver. Numbers associated with that driver was added to the machines. As the racers moved out of the fellowship hall, it looked like NEXTEL drivers moving from the garage area to the starting line.

The scooters were first to race. Starting positions were determined by drawing numbers. Martin "Mad Dog" Beckner drew the pole position, with Jean "Smiley" Mills in the middle in her soon to be retired Rascal 4-wheeler, and "I for Duke" Gary Duke on the outside. Smiley, wearing the 21 outfit of Ricky Rudd, jumped to a quick lead as the green flag fell and was never threatened for the lead. "Mad Dog" driving the #8 Dale Earnhardt Jr. scooter and the #88 UPS/Dale Jarrett scooter driven by "I for Duke" fought for second place for part of the first lap, but "Mad Dog" was just too fast. The order of finish was Smiley, Mad Dog and I for Duke.

The five power chairs were next to race. Bettie Jo "Green Thumb" Wilson drew the pole in her Penske South #12 Ryan Newman Special. Barbara "Stormy" Neely in the #20 Tony Stewart chair started second, first time racer Sarah (Keyboard) Roberts-Witt in the Kasey Kahne #9 third, with Cece "The Shot" Russell, the 2005 Scooter winner, moved up to the power chair division driving the #18 J.J. Yeley ride. Starting fifth was Dana "The Cat" Brower also in her first event, driving the #11 Denny Hamlin chair.

Cece "The Shot" was able to get a good start and left the other racers to fight over second and third positions. Stormy and Keyboard had an early battle for second, with Stormy taking that position. The Cat moved in to challenge Keyboard for third and after a short battle moved ahead. Green Thumb had a typical Ryan Newman day. So cold that her chattering teeth almost drowned out the cheering of the fans, she "froze" at the start and after finally getting the chair going straight, hit the inside wall as she drove out of the fourth turn.. The final order was The Shot, Stormy, The Cat, Keyboard and Green Thumb.

Cece and Jean then raced for the overall Championship. In view of the cold weather, the race was shortened to one lap. Cece, The Shot tried everything, but just did not have the speed to catch Smiley.

Everyone quickly returned to the fellowship hall where North Carolina PLS Ambassador presented Cece and Jean with plaques for winning their divisions and a trophy along with a candy bouquet to Jean "Smiley" Mills as the Grand Champion. Every

participant had a great time, even if a little cold, and were ecstatic when told that they each got to take their NASCAR driver outfits home. Even with the cold, the third annual SAWCAR races were a success.

Cece, Barbara and Arthur joined Don, Bettie Jo and Tracy for a meal to end the Fifth Autumn in Carolina. Bettie Jo and I cannot express the joyous feeling that we have in having everyone visit with us and hope to repeat in 2007. The SAWCAR races generate funds for research. We are waiting for the final report from all sponsors and will send that report later.

LIVING WITH HSP OR PLS

A PLSers Story: Reflections of the National Conference and TeamWalk Chair

Contributed by Betsy Baquet

I'd like to start with a proverb that has become my mantra, "It is better to light a candle than to curse the darkness." This Chinese proverb has been one of my many inspirations for taking action and joining SPF's fight for a cure.

Planning this weekend-long event has been incredibly demanding, both physically and emotionally.

However, the rewards have far outweighed the effort. Taking on a challenge that will have a positive impact on the lives of others, as well as my own, has been an incredible experience. Being part of a team effort that has made steady progress towards treatments and cures has been both empowering and liberating, which "flies in the face" of feeling helpless and trapped by either of these two diseases.

Planning and attending this event has enlightened me to what is possible now and for the future, instead of focusing on what is not possible. I have had the opportunity to meet wonderful, inspiring people who continue to find their way around hurdles they can no longer jump over. I've been amazed by people's accomplishments, despite their affliction, and moved by the outpouring of support so many are willing to give. I have learned that identity has nothing to do with how you walk, talk, move around, or any of the other functions that these diseases can affect. It is what is in your heart and how you cope with the challenges and blessings that may come your way each day.

These are lessons I will not forget.

Seeing first hand the level of dedication on the part of our speakers to make this meeting successful speaks

volumes as to their dedication on a daily basis for finding cures, treatments and to improve the quality of life. I'm thankful and full of hope knowing that brilliant minds are working towards the same goals we are, and that the light at the end of the tunnel gets closer every day.

"It is better to light a candle than to curse the darkness." I encourage everyone to take action and volunteer to "light a candle." It's a life-changing experience you will never forget, with rewards of inspiration, hope and a cure!

Augmentative and Alternative Communication

Ed. Note: Thanks to Lynn Holmes for mentioning AAC-RERC in PLS Friends. I have not located phone numbers for these organizations. Those who use or need an AAC might find help through their neurologist.

The AAC-RERC (Augmentative and Alternative Communication - Rehabilitation Engineering Research Centers) <http://www.aac-rerc.com/> conduct a comprehensive program of research, development, training, and dissemination activities that address the NIDRR priorities and seek to improve technologies for individuals who rely on augmentative and alternative communication (AAC) technologies. The mission of the AAC-RERC is to assist people who rely on augmentative and alternative communication to achieve their goals by advancing and promoting AAC technologies and supporting the individuals who use, manufacture, and recommend them.

Augmentative and Alternative Communication Institute <http://www.aac institute.org/> A worldwide resource for:* People with severe communication disorders or who cannot speak, their families and friends* Professionals and Educators* Researchers, Developers and Manufacturers* Funding and other Parties

Augmentative Communication Inc. <http://www.augcominc.com/> publishes resources that help keep busy professionals and individuals with complex communication needs up-to-date on important developments in AAC. ACI is also a partner in the Rehabilitation Engineering Research Center on Communication Enhancement

...as others see us.

Try going in a wheelchair--your IQ is reduced by at least 50 points!! Several times in recent months, personnel in hospitals have even asked my husband to give my registration information like name, address,

etc. My voice is unaffected, so that's not an issue with me. Usually, I'm patient with these people, but several times I've just glared at them and spoken up. The worst place was the hospital where the supervisor in the radiology department shook her finger at me and informed me that handicapped people couldn't expect the same quality of care as non-handicapped people.

Dolores

Dolores,

Good you are able to speak. In my case as my speech is affected. Wheelchair or no wheelchair my IQ is considered ZERO by most. Almost everyone avoids talking to me, & if they choose to talk to me.... they SHOUT! Initially I was getting angry & annoyed & that made me attempt shout back feebly... making things worse! That was long years... back may be 7/8 years back. Nowadays I think within myself (Bet with myself & WIN most times!) as to how various people will behave seeing me, if they talk to me to what decibel will they raise their voice etc. & I tell you... IT IS FUN! *Jagan*

Aquatic Exercises

<http://www.mayoclinic.com/health/aquatic-exercise/SM00055&slide=1>

Aquatic exercise can be fun for people of all ages, sizes and fitness levels. Classes typically include music and choreography. Like what you've seen? Get your doctor's OK to begin an exercise program, sign up for a class and give it a try.

Water walking with hand webs and water shoes

Water walking is as simple as it sounds. In water that's about waist-high, walk across the pool swinging your arms like you do when walking on land. Avoid walking on your tiptoes, and keep your back straight. Tighten your abdominal muscles to avoid leaning too far forward or to the side. To increase resistance as your hands and arms move through the water, wear hand webs. Water shoes can help you maintain traction on the bottom of the pool.

Deep-water walking with a noodle

With deep-water walking, you start out in water up to your neck. An inexpensive water toy called a noodle can help you stay above the water, and hand webs can increase resistance just as they do in shallower water. Place the noodle between your legs so that it's higher in back than in front. As you walk, move the opposite arm and leg forward. Avoid tilting too far forward, backward or to the side. If you don't know how to swim, wear a flotation vest or float belt instead of a

noodle. For a more intense workout, consider jogging in deep water.

Arm exercise using hand webs

Hand webs can help you strengthen your biceps and triceps. Stand in waist-high water with your arms down, your palms up and your elbows close to your body. Raise your forearms to the level of the water, keeping your elbows close to your body. Then switch direction and push your hands down until your arms are straight again.

Leg exercises using a noodle

Here's another way to use a noodle. Tie the noodle into a knot around your water shoe. Stand with your back to the side of the pool in waist-high water, placing your arms on the pool ledge for stability. Straighten your leg in front of you. Then flex your knee to about a 90-degree position. Return to the starting position and repeat.

Arm exercise using water weights

Water weights are foam barbells that create resistance as you move them through the water. Start with your arms at your sides. Grip the bar of the water weight with your palms facing up. Raise your forearms to the level of the water, keeping your elbows close to your body. Then turn the barbell over so that the palms of your hands face the bottom of the pool. Push your hands down until your arms are straight again.

Resistance exercise using a kickboard

Kickboards provide another type of resistance. Standing up straight with your legs comfortably apart, tighten your abdominal muscles. Extend your arm to the side, and hold the kickboard on each end. Keeping your elbow close to your body, move the kickboard toward the center of your body. Return to the starting position and repeat.

Anger and Bitterness

Contributed by Cathy West.

I was having a really hard time with anger and depression. I was already on 2 antidepressants, don't know how bad off I would be if I wasn't on them. My more athletic friends who I used to hang out with, quit asking me to do things with them. I was working in ICU and when I started falling a lot at work I was told that I had to change to another area of nursing and now I am in Case Management. I didn't know my startle reflex had gotten bad until a couple of weeks ago when someone honked a horn in the parking deck and I almost shot through the roof and then felt my legs turn to jello, but thank goodness I didn't fall. I have been feeling that I am getting a little worse lately

and I think this has started to move into my left hand just a little bit. So, suffice it to say...I was starting to get very bitter and feeling how unfair it was that my life has to be so hard compared to everyone else around me.

I started to become more involved in my church and went to a meeting where women who seemed to have the perfect life stood up and told about all these horrible things they had gone through or are going through now and how their faith is helping them through it. I saw that I wasn't alone in fighting a tough battle. Somehow it made me feel a little stronger. I am trying to find words of encouragement now from scripture and from their stories.

The first person I met with PLS (met on the internet) told me to get a counselor right away. I asked my neuro doc to tell me who I could see for counseling who is experienced in helping someone with chronic diseases. The counselor seemed so understanding and validated my feelings by saying, "Yes, you have a lot of stress going on in your life right now".

Ok, so, to sum it all up....I am working on getting through the anger and bitterness over what life has dealt me. I am using 2 antidepressants, becoming more involved in church and trying to have a closer relationship with God and looking to the scriptures for words of encouragement and strength. I am hanging out with new friends that care about me and include me in things. I think my family is a tremendous help to me daily. I going to a counselor and I am actually looking forward to the next visit and feel optimistic about it. I use a cane all the time now. I know that if I don't stretch daily that I will have a much harder time walking and I know that the best medicine for me is going to the pool and swimming and stretching there. The pool is a big antidepressant for me and when I am in the water I can do what everyone else can do and I get to feel normal for a little while! With bitterness and anger we only hurt ourselves more. Each day is going to be a challenge for us, but we must try to find the beauty and joy in each day and focus on the gifts we have, not the ones we are lacking. Life is short...I want to enjoy what I can. Perhaps see if any of the things I have tried can help you and also.

Letter to the Editor

Patricia Lockwood

This letter is about 14 years overdue. I've been meaning to write you all and tell you what a good job you are doing editing and writing the Synapse newsletter. I look forward to every issue to see what

the newest research studies are and where located. I was diagnosed with ALS in 1992 and went to bed waiting to die. In 1998, my neurologist said, "Well, you haven't died yet, so maybe you have PLS instead. You aren't terminal after all." So, I got up and started trying to rebuild my life as best I could on my scooter and being unable to talk. I had a lot of help from my husband and have adjusted very well. I want to tell everybody out there to never give up. They are discovering more treatments and miracles everyday. Thanks for all you have done.

My Scooter

Contributed by Mary Tipp maryktipp@yahoo.com

I never thought I would see the day that I would be so excited to be getting a mobility scooter. I was diagnosed 2 years ago. My spasticity has gotten to the point I almost can't walk. My neuro. feels I should not have any aid. I was aware I needed more. I went to a MDA CLINIC. I was surprised by the caring people of the MDA and the caring female doctor. First thing she did was write a prescription for a powered scooter. I was very shocked to learn the MDA picked up the tab for all testing and my scooter. What a blessing they gave me. I feel like I have wings or new legs. Was anybody else this excited to get some freedom back and not feeling self conscience about my odd walk from strangers who don't understand?

Mouseless Mouse

<http://pcbility.com/mouseHelp.html>

PCBility mouse is a computer mouse substitute. This software enables you to move and click the mouse cursor by moving your head only. It works using a simple web camera. The product will identify your head, track the head movements and the mouse cursor will be moved to the spot on the screen you are looking at. If you would like to send a click, Just stop moving and a pop- up menu will let you choose the wanted click (Left click, Right click, Double click, Double right click, and even drag and drop). The software is very accurate. Just a few moments of training and you will be able to use your PC with no hands at all.

CAREGIVING

Caring for Someone Living Alone

Contributed by Eva

1. Get your loved one online with PLS Friends or SP E-news. Once posting to us she can decide if she would like to meet us. Also check out our pictures from the Connections on the Synapse website and guide your Mom to them. When she sees all our smiles, I'm sure she will feel better.
2. Get your loved one connected with an ALS Support Group. ALSA covers PLS as well.
3. Get your loved one on a digital high speed cable and have a camera system installed. That way you can check in on your loved one throughout the day to assure he/she is OK....he/she might not like the intrusion but at least he/she'll know your concerns. You might also have a camera on your end to assure by your facial expressions that you are handling things emotionally. If you add a computer microphone you are basically live to each other when you are home.
4. If your loved one doesn't have a friend to get him/her to and from all the tests and appointments, Comfort Keepers provides that service plus many more.

Coping with Burnout – For Caregivers

From MA-ALS

Being a caregiver of someone with a progressive, degenerative disease is a very important role. It usually involves a number of tasks that can be very time consuming, and can require a great deal of effort. If we do not learn to recognize that certain tasks and expectations can take their toll on us, regardless of what role(s) we play in life, we may find ourselves headed down the path of burnout.

Common causes of burnout

- Perfectionism:** A perfectionist continually focuses on what needs to be improved, rather than what has been accomplished. When this becomes our focus, we may never feel that we have succeeded at anything.
- Never-ending tasks:** Never-ending tasks describe work that appears to lack both a beginning and end. This can lead us to feel as if we have no closure, and therefore have not completed anything.
- Work overload:** Work overload is when we have more work to do than we can complete in a given amount of time. When we operate this way, we set ourselves up for failure.
- Impossible tasks:** Impossible tasks suggest that we are physically unable to do something we may feel we

should be able to do. If we believe that we should be able to do something that we cannot do, we automatically feel like we have failed. It is a no-win situation.

Multiple roles: Many of us play a number of important roles in our lives, such as wife/husband, mother/father, caregiver and breadwinner. It is easy to feel overwhelmed when we are trying to play multiple roles in our lives.

Self-sacrifice: Self-sacrifice occurs when we agree to take on certain tasks despite our true desire to complete them. When we continually agree to things that we would rather not do, we inevitably become resentful.

Unspoken feelings: Any emotion that is unexpressed creates "blocks" in our ability to complete tasks and to function in relationships. If we do not speak about how we are truly feeling in a situation, those emotions surface in other, unproductive ways such as being

chronically late, frequently forgetting things, or reacting in ways that are inappropriate to the situation.

Common symptoms of burnout

It is important to remember that we are not alone in our experiences with burnout. At some point in time, it is likely that most of us will experience one or more of the following symptoms:

"Negative" emotions: "Negative" emotions are the feelings that are often the least comfortable to feel. One symptom of burnout is consistent "negative" feelings such as anger, anxiety, dissatisfaction and guilt.

Interpersonal problems: We might experience conflict with others in the form of emotional outbursts, overreacting, hostility and withdrawal.

Health Problems: Some common health problems associated with burnout are frequent insomnia, fatigue, headaches, backaches, lethargy and high blood pressure.

Poor performance: We may become less productive due to boredom, lack of enthusiasm, feelings of fear or an inability to concentrate.

Substance abuse: Another symptom of burnout is a marked increase in the consumption of alcohol and/or other drugs, cigarette smoking, caffeine and food.

Workaholic: We might be inclined to work more hours due to feelings of inadequacy, believing that the more we work, the better we will feel.

Depression: Depression is the suppression of emotions. We may be depressed if we notice an overall feeling of hopelessness and meaninglessness.

Loss of self-esteem: Simply stated, the loss of self-esteem equals a decrease in self-confidence.

Solutions for dealing with burnout

Once we become more aware of why we experience burnout, and the various ways burnout manifests itself, we can begin to focus on ways to guard against it. It is time to develop a plan of action!

Take care of yourself:

- Regularly feed your body nutritious foods
- Get sufficient rest
- Exercise routinely
- Pay attention to your body's signals of stress

Practice stress-reducing strategies:

- Breathing exercises
- Yoga
- Progressive relaxation techniques
- Massage
- Meditation
- Tai chi

Develop a strong support system:

- Surround yourself with friends/family by whom you feel supported.
- Attend a support group where you can share your concerns and feelings.
- Create a support/discussion group at work where you can share your concerns, while being willing to talk about your part in the problem and the solution.
- Utilize your company's Employee Assistance Program (EAP) to receive support/counseling or referrals for services that can assist you.
- See a counselor or therapist if you are in need of more extensive mental health support.

Create a fulfilling life:

- Make conscious decisions about how you want to spend your time.
- Say "yes" to what you want to say "yes" to, and say "no" to what you want to say "no" to.
- Acknowledge your priorities and actively build your life around them.

The key to avoiding burnout is to continually seek balance in our lives. The more informed we are about our own issues with burnout, the better armed we will be to take care of ourselves. Consequently, the better job we do of taking care of our own needs, the more we can be physically, mentally, and emotionally available to those around us.

MEDICAL UPDATES

UCSF Study Needs Participants

Contributed by Linda Gentner

Dr. Catherine Lomen-Hoerth asked me to publicize this study in our patient community. You will need to pay your own travel expenses. If you're nearby, please sign up to participate.

Brain Structure and Function in ALS and PLS

Purpose of the study:

ALS and PLS are neurological conditions that were traditionally considered to only affect the motor systems in the brain. We have learned that among a subset of ALS and PLS patients, other brain regions may be involved, including those governing our cognitive functions, such as memory, attention, and concentration. It is our goal to understand which regions of the brain are affected by ALS and PLS and how patients differ with respect to symptoms, disease severity, and prognosis. We will use highly sensitive MRI technologies to determine how we can detect these neurological brain changes as early as possible. Early detection of patients who are at greater risk for cognitive symptoms in addition to motor involvement would significantly inform the development of improved treatment and disease management strategies.

This study is funded by the ALS Association. Research is a primary mission at UCSF, and without participation from patients such as yourself, we would be unable to understand these conditions or pursue effective treatments.

Length of the study:

Your participation in this study will last 12-18 months. Your participation will include 2 visits (one visit at the beginning and one at the end). Each visit will be approximately 3 hours.

You will be asked to participate in three activities:

- 1) Neurological evaluation at the UCSF ALS Center
- 2) Cognitive testing: we will administer measures of memory and concentration
- 3) MRI at the UCSF Imaging Center at China Basin.

Where the study is located:

1) UCSF Parnassus Main Campus: You will undergo a neurological evaluation at the UCSF-ALS center.

2) UCSF Imaging Center at China Basin (right across from the AT & T Baseball park/

Caltrain station): This is where you will undergo the cognitive testing and the MRI.

Catherine Lomen-Hoerth, MD, PhD

Director, ALS Treatment and Research Center at UCSF

350 Parnassus Avenue, Suite 500

San Francisco, CA 94117

(415) 514-0490; (415) 514-0491

A Letter about the NWU PLS Registry

Dear PLS Friends,

Coordinating the PLS Registry continues to be an exciting challenge. With the advantage of working with PLS exclusively, I am updating existing records in addition to recruiting new members and facilitating collection of blood samples, medical records, and family health information. We have begun adding spinal fluid samples to our databank as well, and continue to refine the arrangements for coordinating the lumbar punctures and shipment of frozen samples from other locations.

We anticipate some ebb and flow in our PLS collection numbers, as some of the samples awaiting diagnosis verification will inevitably change categories to HSP, ALS, or other. To that end we are requesting registry members to assist us with updating their medical records on a periodic basis.

Patients in our PLS database reside in 40 different states and 8 countries. One estimate of the incidence of PLS in the U.S. is 400-500. Based on that estimate, if 156 of our current 182 (subtracting 8 foreign members and 12 awaiting diagnosis verification) PLS registry members are verifiably diagnosed with PLS, we may have enrolled 31-39% of the total number of PLS patients in the country.

We are very grateful to registry members who continue to provide welcome assistance with recruitment, as they encourage members of their local support groups and websites to participate in the registry.

The following table provides an update as we continue in the data collection phase of the PLS work.

Samples for Disequilibrium studies	Aug-04	Aug-05	Sep-06
Sets of patients and both parents	13	20	20
Sets of patients & unaffected brother or sister	68	84	111
Family sets (patient, both parents & 1 or more sibs)	5	9	12
Sets of patients, 1 parent and 1 or more sibs			17
Samples for Case Model studies			
PLS patients	102	152	182
PLS spousal controls	8	26	30
Samples awaiting verification			
May include changes in diagnosis to SP, ALS, other			12
Spinal fluid samples		4	5

We are very excited that with the newest technology, and the largest number of ALS DNA samples in the country, Dr. Teepu Siddique is beginning a whole genome association study of ALS. We hope to achieve our goal of including virtually every PLS patient in North America as soon as possible so analysis can begin with the PLS samples as well.

For information on joining the PLS Registry I welcome a contact by email, mail, or phone. We unfortunately do not have a toll-free number, but if I receive voicemail or a note in the mail, I will happily return the call. For more information about our program, please visit our website at www.neurogenetics.northwestern.edu

Best regards,

Grace Carlson-Lund, RN, BSN

Clinical Research Nurse

Neuromuscular Disorders Program

Northwestern University Feinberg School of Medicine

303 East Chicago Avenue

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Research Snippets

ALS Linked to Variant of Gene Involved in Pesticide Processing

In a published report by researchers at Northwestern University in Chicago led by Teepu Siddique, M.D.,

gene evidence suggests that differences in the ability to detoxify pesticides and nerve gases might explain why ALS occurs at higher frequency among veterans of the Gulf War, and why certain environmental exposures might increase the risk of the disease. As reported at the meeting of the American Academy of Neurology in San Diego this spring and now published on line in advance of the August issue of Neurology, variants of genes coding for the so called PON enzymes that handle pesticides and other chemicals are linked to a two-fold increased risk to develop ALS. This suggestive finding is not cause and effect but gives a place to start looking for genetic and environmental risk factors for the disease.

ALS2 Mutation Targets Upper Motor Neurons in Mice

Two publications in the June Annals of Neurology explain why it has been difficult to make a mouse that shows damage from mutation in the alsin protein implicated in a type of ALS that begins at a young age.

Investigators working with Don Cleveland, Ph.D., at the University of California, San Diego, reported online in June in the Annals of Neurology that mice missing the alsin protein show progressive degeneration of upper motor neurons traveling down the spinal cord but no damage to the lower motor neurons that go from the spinal cord to muscles. The mice moved more slowly than normal mice but did not show muscle weakness. The affected mice do not show the spasticity from upper motor neuron damage that people show. Thus alsin mutation produces a distinct disease that is not exactly the same as ALS, the researchers concluded. Instead it more closely resembles a severe form of hereditary spastic paralysis.

Reporting in the same publication, researchers working with George Haase, M.D., Ph.D., at Institut National de la Sante et de la Recherche Medicale, and AVENIR, France, show that motor neurons with depleted alsin (by a different technique than that used by the Cleveland group) die at a rate of 30 to 40 percent in culture, and surviving cells had limited ability to send out fibers.

Two structural proteins interact in Hereditary Spastic Paraplegia

Two proteins that help nerve fibers stay intact and able to supply the rest of the cell with needed materials apparently can cause HSP. The proteins are spastin

and atlastin, and they may have to interact properly to maintain nerve fiber health, suggested the Columbia University researchers, led by Brett Luring, M.D., Ph.D., in their report in July in the Proceedings of the National Academy of Sciences.

Genetics of Cognitive Change That is Linked with ALS

An international collaboration has discovered a new mutation that appears to cause some cases of ALS. As reported online in *Neurology* June 28, the mutation to a protein called charged multivesicular body protein 2B (CHMP2B) previously associated with cognitive change was found in two unrelated patients with motor neuron disease, as published by Elizabeth Fisher, Ph.D., University College London, and colleagues. These findings add to others that implicate vesicle function in motor neuron damage.

SPF funded scientists report findings

Drs. Vincent Cunliffe and Jonathan Wood received a 2 year SPF research grant in 2004. This is an abstract of their first paper describing their work. Their work sheds more light into the role of the Spastin protein in causing the symptoms with which we are all too familiar.

"The microtubule-severing protein Spastin is essential for axon outgrowth in the zebrafish embryo."

Wood JD, Landers JA, Bingley M, McDermott CJ, Thomas-McArthur V, Gleadall LJ, Shaw PJ, Cunliffe VT.

MRC Centre for Developmental and Biomedical Genetics, Department of Biomedical Science, University of Sheffield, Sheffield S10 2TN, UK; Academic Neurology Unit, University of Sheffield Medical School, Sheffield S10 2RX, UK.

SPG4 mutations are the most common cause of autosomal dominant HSP and spastin (the SPG4 gene product) is a microtubule severing protein that shares homology with katanin, the microtubule severing activity of which promotes axon growth in cultured neurons. Given the sequence and functional similarity between spastin and katanin, we hypothesised that spastin promotes the dynamic disassembly and remodelling of microtubules required for robust, properly directed motor axon outgrowth. To investigate this hypothesis, we cloned the zebrafish . Our results reveal a critical requirement for spastin to promote axonal outgrowth during embryonic development, and they validate the zebrafish embryo

as a novel model system in which to dissect the pathogenetic mechanisms underlying HSP. Taken together with other recent studies, our findings suggest that axon outgrowth defects may be a common feature of childhood SPG3A and SPG4 cases..

The Spastic Paraplegia Rating Scale (SPRS): a reliable and valid measure of disease severity.

Schule R, Holland-Letz T, Klimpe S, Kassubek J, Klopstock T, Mall V, Otto S, Winner B, Schols L. Hertie Institute for Clinical Brain Research and Department of Neurology, Eberhard Karls University Tubingen, Germany.

A 13-item scale was designed to rate functional impairment occurring in pure forms of spastic paraplegia (SP). Additional symptoms constituting a complicated form of SP are recorded in an inventory. Two independent patient cohorts were evaluated in a two-step validation procedure. Results: Application of SPRS requires less than 15 minutes and does not require any special equipment, so it is suitable for an outpatient setting. Conclusion: The Spastic Paraplegia Rating Scale is a reliable and valid measure of disease severity.

New Source of Multipotent Adult Stem Cells Discovered in Human Hair Follicles

Researchers at the University of Pennsylvania School of Medicine have isolated a new source of adult stem cells that appear to have the potential to differentiate into several cell types. If their approach to growing these cells can be scaled up and proves to be safe and effective in animal and human studies, it could one day provide the tissue needed by an individual for treating a host of disorders, including peripheral nerve disease, Parkinson's disease, and spinal cord injury. "We are very excited about this new source of adult stem cells that has the potential for a variety of applications," says senior author Xiaowei (George) Xu, MD, PhD, Assistant Professor of Pathology. "A number of reports have pointed to the fact that adult stem cells may be more flexible in what they become than previously thought, so we decided to look in the hair follicle bulge, a niche for these cells." Xu and colleagues report their findings in the latest issue of the *American Journal of Pathology*. Hair follicles are well known to be a source for adult stem cells. Using human embryonic stem cell culture conditions, the researchers isolated and grew a new type of multipotent adult stem cell from scalp tissue

obtained from the National Institute of Health's Cooperative Human Tissue Network.

The multipotent stem cells grow as masses the investigators call hair spheres. After growing the "raw" cells from the hair spheres in different types of growth factors, the investigators were able to differentiate the stem cells into multiple lineages, including nerve cells, smooth muscle cells, and melanocytes (skin pigment cells).

The differentiated cells acquired lineage-specific markers and demonstrated appropriate functions in tissue culture, according to each cell type. For example, after 14 days, 20% to 40% of the cells in the melanocyte media took on a weblike shape typical of melanocytes. The new cells also expressed biomarkers typical of pigment cells and when placed in an artificial human skin construct, produced melanin and responded to chemical cues from normal epidermis skin cells.

After 14 days, about 10% of the stem cells in the neuronal cell line -- a type of cell not present in skin or hair -- grew dendrites, the long extensions typical of nerve cells and expressed neuronal proteins. The neurotransmitter glutamate was also present in the cells, but the neurotransmitter dopamine was not detected. Thirdly, about 80% of the stem cells grown in the muscle media differentiated into smooth muscle cells. These new muscle cells also contracted when placed in a collagen matrix.

Overall, the researchers showed that human embryonic stem cell media could be used to isolate and expand a novel population of multipotent adult stem cells from human hair follicles. "Although we are just at the start of this research, our findings suggest that human hair follicles may provide an accessible, individualized source of stem cells," says Xu. The researchers are now working on inducing other cell types from the hair sphere cells and testing the cells in animal models.

Study co-authors are Hong Yu, Suresh M. Kumar, and Geza Acs, all from Penn; and Dong Fang, Ling Li, Thieng K. Nguyen, and Meenhard Herlyn, all from the Wistar Institute, Philadelphia.

Juvenile Onset PLS

"The first ALS2 missense mutation associated with JPLS reveals new aspects of alsin biological function." Panzeri C, De Palma C, Martinuzzi A, Daga A, De Polo G, Bresolin N, Miller CC, Tudor EL, Clementi E, Bassi MT. IRCCS E. Medea, Laboratory of Molecular Biology,

Bosisio Parini Lecco, Italy.

The causative gene for JPLS was found to be ALS2, which is also responsible for a recessive form of ALS, for infantile onset ascending hereditary spastic paralysis (IAHSP) and for a form of complicated hereditary spastic paraplegia (cHSP). All ALS2 mutations so far reported generate alsin protein truncation. Here, we describe the first homozygous missense mutation in ALS2, p.G540E. Our results provide the first demonstration that a missense mutation in alsin is cytotoxic.

Brain atrophy in pure and complicated hereditary spastic paraparesis: a quantitative 3D MRI study.

Kassubek J, Sperfeld AD, Baumgartner A, Huppertz HJ, Riecker A, Juengling FD.

Department of Neurology, University of Ulm, Ulm, Germany.

The purpose of this study was to evaluate if brain volumes in HSP were altered compared with a control population. Atrophy was more pronounced in c-HSP, consistent with the more severe phenotype including extramotor involvement. Thus, global brain atrophy, detected by MRI-based brain volume quantification, is a biological marker in HSP subtypes.

Hereditary spastic paraparesis and psychosis

McMonagle P, Hutchinson M, Lawlor B.

Department of Neurology, St Vincent's University Hospital, Elm Park, Dublin, Ireland.

Reports of psychiatric disorders in patients with HSP are limited to mood disturbances rather than to psychosis. We had noted significant psychotic illness in several patients recruited to a survey of HSP in Ireland and therefore set about examining the frequency and nature of psychosis in our patients with HSP. Two groups of HSP patients may have increased risk of developing psychosis: those with Kjellin's syndrome and those with SPG4-HSP.

Merger Of Nanowires And Neurons Could Boost Efforts To Measure And Understand Brain Activity

Opening a whole new interface between nanotechnology and neuroscience, scientists at Harvard University have used slender silicon nanowires to detect, stimulate, and inhibit nerve signals along the axons and dendrites of live mammalian neurons. Harvard chemist Charles M. Lieber and colleagues report on this marriage of nanowires and neurons in the journal *Science*. . "These

extremely local devices can detect, stimulate, and inhibit propagation of neuronal signals with a spatial resolution unmatched by existing techniques." The tiny nanowire transistors developed by Lieber and colleagues gently touch a neuronal projection to form a hybrid synapse, making them noninvasive, and are thousands of times smaller than the electronics now used to measure brain activity. "This work could have a revolutionary impact on science and technology," Lieber says. "It provides a powerful new approach for neuroscience to study and manipulate signal propagation in neuronal networks at a level unmatched by other techniques; it provides a new paradigm for building sophisticated interfaces between the brain and external neural prosthetics; it represents a new, powerful, and flexible approach for real-time cellular assays useful for drug discovery and other applications; and it opens the possibility for hybrid circuits that couple the strengths of digital nanoelectronic and biological computing components."

SPF funded researchers publish paper re: ALS2/Alsin

The SPF funded Drs. Hayden and Leavitt last year. Their paper is about the function of Alsin, a protein that can cause juvenile onset PLS if it is abnormal."ALS2-deficient mice exhibit disturbances in endosome trafficking associated with motor behavioral abnormalities" Mark Devon RS, Orban PC, Gerrow K, Barbieri MA, Schwab C, Cao LP, Helm JR, Bissada N, Cruz-Aguado R, Davidson TL, Witmer J, Metzler M, Lam CK, Tetzlaff W, Simpson EM, McCaffery JM, El-Husseini AE, Leavitt BR, Hayden MR. Centre for Molecular Medicine and Therapeutics, Department of Medical Genetics, University of British Columbia, 980 West 28th Avenue, Vancouver, BC, Canada V5Z 4H4.

ALS2 is an autosomal recessive form of spastic paraparesis (motor neuron disease) with juvenile onset and slow progression caused by loss of function of alsin. To establish an animal model of ALS2 and derive insights into the pathogenesis of this illness, we have generated alsin-null mice. There is a significant decrease in the size of cortical motor neurons, and Als2(-/-) mice are mildly hypoactive. Altered trophic receptor trafficking in neurons of Als2(-/-) mice may underlie the histopathological and behavioral changes

observed and the pathogenesis of ALS2.

Two Approaches to Drug Discovery in SOD1-Mediated ALS.

Broom WJ, Auwarter KE, Ni J, Russel DE, Yeh LA, Maxwell MM, Glicksman M, Kazantsev AG, Brown RH Jr.

Day Neuromuscular Research Laboratory, Mass General Institute for Neurodegenerative Disease, Massachusetts General Hospital, Charlestown, MA. To date, 105 different mutations spanning all 5 exons have been identified in the SOD1 gene. Mutant SOD1-associated ALS is caused by a toxic gain of function of the mutated protein. They have designed 2 cell-based screening assays to identify small, brain-permeant molecules that inactivate expression of the SOD1 gene or increase the degradation of the SOD1 protein. Here they describe the development and optimization of these assays and the results of high-throughput screening using a variety of compound libraries, including a total of more than 116,000 compounds. Ultimately, the authors believe that these 2 cell-based assays will provide powerful strategies to identify novel therapies for the treatment of inherited SOD1-associated forms of ALS.

Neuropsychological changes in patients with primary lateral sclerosis.

Piquard A, Le Forestier N, Baudoin-Madec V, Delgadillo D, Salachas F, Pradat PF, Derouesne C, Meininger V, Lacomblez L. Federation des Maladies du Systeme Nerveux, Hopital de la Pitie-Salpetriere, Paris. Primary lateral sclerosis (PLS) has been defined as rare, and the neuropsychological changes remain poorly defined. We studied 20 patients with a diagnosis of PLS. We carried out an extensive psychometric testing including a general assessment (memory, language, attention, visual-constructional ability and praxis) and a more specific assessment of prefrontal and premotor cortex functions in order to characterize the neuropsychological profile of the patients compared to matched controls, and explore executive functions and premotor cortex functions. None of the PLS patients was demented but they all presented memory deficits reflecting an executive dysfunction. All patients but three had signs of premotor and/or prefrontal cortex deficits. The cognitive impairment in PLS, specifically related to a frontal lobe dysfunction, seems qualitatively similar to ALS. Our results suggest a

patchy distribution of cortical involvement in PLS but it remains difficult to draw any definite conclusion as to the spatio-temporal progression of the disease into the different regions of the frontal lobe.

Dear Synapse Readers,
Brainstorming is underway to figure out how SP Foundation can best serve our patient community. A Communications Committee has been formed. SP Board members Kris Brochini, Linda Gentner and Mark Weber; Kathi Geisler – webmaster of www.sp-foundation.org ; Cathy Wilkes, Executive Director of SP Foundation; and Jim and Thurza Campbell - webmaster and editor of www.synapsepls.org
During the next year we want to eliminate duplication, enhance what is good, assure confidentiality in any list and data base, expand the circulation of the quarterly newsletter, and make as simple as possible finding information.

The goal is to have one SP website which will include Synapse. There will continue being an on-line and paper quarterly newsletter. We hope to unify patient lists into one secure data base.

I'd appreciate any and all suggestions of how we might better serve you. Please either e-mail me at synapsepls@comcast.net or write to me:

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Sherborn MA 01770-1622

Sincerely,
Thurza

Synapse

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Event Photos

<p>National Conference - NY</p> 		
<p>SP President Annette Lockwood welcoming 50 attendees</p>	<p>(l-r) Annette with new SP Foundation Executive Director, Cathy Wilkes</p>	<p>Betsy with husband Ed lead over 100 walkers through the Long Island park.</p>
<p>National Conference - NY</p> 	<p>TeamWalk-California</p> 	<p>TeamWalk-Georgia</p> 
<p>Face painting was a popular item with the many kids that participated in the walk</p>	<p>Over 60 people attended the TeamWalk in Fremont.</p>	<p>A police cruiser provided escort for 27 walkers in Thomasville</p>
<p>TeamWalk-Maryland</p> 	<p>TeamWalk-Massachusetts</p> 	<p>SAWCAR in North Carolina</p> 
<p>30 people enjoyed a home-cooked meal after their walk in Annapolis.</p>	<p>Thirteen people and one dog circled the Lexington common.</p>	<p>Power chair racers get the green flag from event organizer, Don Wilson.</p>
<p>TeamWalk-Ohio</p> 	<p>TeamWalk-Oklahoma</p> 	<p>TeamWalk-Tennessee</p> 
<p>Ohio's TeamWalk this year was held in Toledo.</p>	<p>A covered wagon bridge was part of the course for the Norman group of 25</p>	<p>Eighteen people attended this TeamWalk in Nashville</p>