Synapse

Volume 12, Issue 4 Autumn, 2009

Newsletter of the Spastic Paraplegia Foundation

TeamWalks in Pictures

Connie Duran and family at the Utah TeamWalk

The men are on their mark for The Magnificent Mile

Cece Russell and the Rocheleaus at the finish line in Raleigh, North Carolina

New England TeamWalk participants gather at the entrance to Fenway Park by Ted Williams' statue

Nashville TeamWalk participants with Chairman, Jim Sheorn at the far right

Ann Lakin leads the TeamWalk in New York

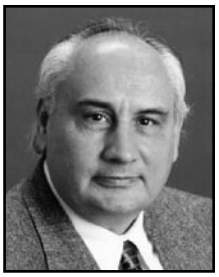
TeamWalkers in Pleasanton, California

TeamWalk summaries begin on Page 5

The SPF table at Mt Kisco, New York Sale Days

SP Foundation News

President's Letter



Mike Podanoffsky
SPF President

As I write this we are in the middle of TeamWalk events. We hold TeamWalk events in the late summer and fall. If you have never attended a TeamWalk or a connection, you definitely should. It is an opportunity for you and your caregiver to meet other people who have the same condition and concerns. I attended the TeamWalk in New England. This year it was a tour of the famed Fenway Park, home of the Boston Red Sox with its famous "Green Monster" wall. Once the ball is over the wall, it is a definite home run. TeamWalks don't necessarily involve walking. You walk or roll as far as you want. Whichever you do, you will never have another experience like it. It's more about connecting with people than the physical activity. It is about getting out of the house and away from the usual routine.

TeamWalks are one of the ways we raise funds for research, which is the main purpose of the Spastic Paraplegia Foundation. That research is making progress, the kind of progress that might just some day result in therapies to slow down, stop, or repair the upper motor neuron damage that causes HSP and PLS. The research is very cutting edge. ***We could double our research funds if each of you raised just $200 each, just by asking your friends and family to donate a small amount.*** That could raise as much as $200,000 we could use to accelerate the progress of research, fund more research, and publish this newsletter more often. All because of you. All for you. I bet all of you could do it. Try it at http://sp-foundation.org/fundraise-challenge.htm.

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

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Medical Research. We have prepared for you a complete chart of all of the research grants SPF has funded since 2003. The chart briefly summarizes the results of each grant to date. You'll find it in the center of the issue. We also announce in another article, (page 4) the 2009 grant recipients, each of whom will receive a $120,000 grant from SPF.

The 2010 National Conference will be held Los Angeles, CA on May 21 – 23, 2010. We are still working out the speakers and agenda. We are focusing on you and time when you can meet others. We also want to provide an update on research and medical progress. You can have your voice heard by sending your suggestions to topics@sp-foundation.org. We love hearing from you.

Mike Podanoffsky,
SPF President

"The Star in Our Future Is the Cure... Through Research"

Contributed by Malin Dollinger, Chair.

The SPF 2010 National Conference will be held in Los Angeles on May 21-23. It will begin with dinner Friday evening, with sessions all day Saturday, and close with a session on Sunday from 9 to 11 am. It will be at the Los Angeles airport [LAX] Hilton Hotel, which is 5 minutes from the airport. A free shuttle is available from the airport to the hotel. The room rate is $95 a night, special for our meeting. We are excited about the upcoming program and look forward to having people from all over the United States, as well as Canada and other countries.

The hotel is well-equipped for our meeting, with splendid meeting rooms close to the elevators and rest rooms, and three types of handicap-accessible rooms, 34 such rooms in all.

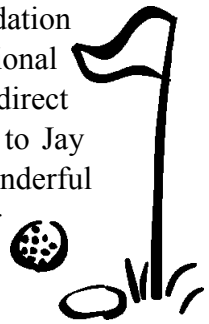
For those who wish to come to Los Angeles earlier, and/or stay later, there are many attractions. These include the Getty Center [art/paintings/sculpture], Knott's Berry Farm, Disneyland, Universal Studios, Hollywood, the beach cities with their attractions, world-class museums, concert halls [e.g. Disney hall], theaters, studio wardrobe outlets [take home a souvenir], many shopping centers, and many other attractions, to be described in our formal announcement and in materials you will be given when you arrive.

Los Angeles has 329 days of sunshine per year -- need we say more!! We look forward to rolling out the red carpet, meeting you, and sharing our journey together. We are not able to make hotel reservations yet, and the official announcement and reservation forms will be available soon, and will contain all the information you need. Please do not call the hotel for reservations yet, since you may not get the special room rate or the correct handicap room. There are three different types of handicap rooms, and this will be explained in detail in the official announcement. If you have other types of questions send an email to malind@cox.net or if you do not have internet access, call Malin at (310) 378 4059.

The Drive "Fore" SPF Receives a $100,000. Donation from The Travelers Foundation

Contributed by James F. Brewi

On a beautiful early fall day in Jamesburg, NJ, The Travelers Insurance Company hosted The Drive "Fore" SPF honoring me, the Regional Director. 72 golfers, mostly my business associates, some I had worked with for thirty years teed it up for the cause. I was diagnosed with PLS in early 2008. With the love and support of my family as well as support from The Travelers I have continued to work and remain focused on the positive. Chairman & CEO of The Travelers, Jay S. Fishman was on hand for the day's events including the reception that followed golf which was joined by an additional 25 family and friends. The day was highlighted when I presented Mr. Fishman with a framed letter from SPF President Mike Podanoffsky thanking The Travelers for hosting this event. Mr. Fishman spoke passionately of the search for a cure and announced that The Travelers Foundation would donate $100,000 to SPF! Additional funds were raised through auctions and direct donations. We are extremely grateful to Jay Fishman and The Travelers for the wonderful support they have provided me and our family.



CHRISTOPHER & DANA REEVE FOUNDATION
TODAY'S CARE. TOMORROW'S CURE.

The Spastic Paraplegia Foundation is proud to announce that it has received a $2,500 Quality of Life grant from the Christopher & Dana Reeve Foundation. The award was one of 86 grants totaling almost $500,000 awarded by the Reeve Foundation to nonprofit organizations nationwide that help people living with paralysis and their families become more integrated members of society. Since inception of the program in 1999, nearly 1,600 grants have been awarded, totaling almost $13 million.

"The Quality of Life program recognizes and supports organizations that assist individuals living with paralysis, their families and caregivers in ways that more immediately provide them with increased independence, well being, and improved access," said Peter T. Wilderotter, president and CEO of the Christopher & Dana Reeve Foundation. "Throughout the past ten years, we have had the privilege of being able to impact the lives of thousands of people living with paralysis through these deserving organizations. As Dana Reeve used to say, 'our Quality of Life program is about freedom' and we are pleased to do our small part to assist the Spastic Paraplegia Foundation in fulfilling its mission."

About the Reeve Foundation

The Christopher and Dana Reeve Foundation is dedicated to curing spinal cord injury by funding innovative research, and improving the quality of life for people living with paralysis through grants, information and advocacy. For more information, and to review the entire list of Quality of Life grant recipients, please visit our website at www.ChristopherReeve.org or call 800-225-0292.

The Spastic Paraplegia Foundation is grateful for the support that the Christopher & Dana Reeve Foundation has given. We look forward in helping to provide information to those affected with either HSP or PLS.

Spastic Paraplegia Foundation announces 2009 Research Grant Awards

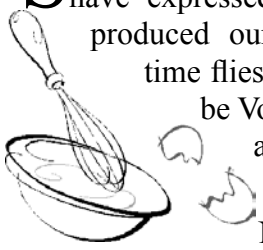
The Spastic Paraplegia Foundation (SPF) announces that it has awarded $240,000 in research grants to two researchers -- $120,000 to each researcher. The SPF's Scientific Advisory Board rated their proposals as outstanding opportunities to advance research and help find the cures for Primary Lateral Sclerosis (PLS) & Hereditary Spastic Paraplegia (HSP). The grant recipients, and their proposals, are as follows:

Hiroshi Mitsumoto, MD, D.Sc., Wesley J. Howe Professor of Neurology, Director, Eleanor & Lou Gehrig MDA/ALS Center, The Neurological Institute, Columbia University College of Physicians and Surgeons, New York, **NY "Multicenter Prospective PLS Natural History Study"** Dr. Mitsumoto, along with 14 collaborators throughout North America will undertake a longitudinal natural history study of PLS and establish a BioBank of biological samples that may be essential for future investigation of PLS pathogenesis. Such a longitudinal study will provide knowledge concerning early signs that predict UMN-onset ALS or PLS as well as the biology of PLS. Patients will be strongly urged to participate in the study once it begins.

Elena Irene Rugarli, M.D., University of Cologne, Germany. **"Exploring alternative functions of paraplegin, a protein involved in autosomal recessive and sporadic HSP."**

Cookin' for a Cure, Volume II

Submitted by Linda Gentner



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 no more than 5 of 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.

TeamWalk Reports

Sarah Roberts-Witt, editor

Salt Lake City, Utah - August 29th

Connie Duran and Melanie McIntosh, Chairs

August 29th turned out to be a pretty special day. Organizing a TeamWalk was a first for me, and as the date got closer, I didn't think I could do it. My friend Melanie gave me a pep talk and my spirits lifted. The park we chose was very nice with a playground for children. It was also well-equipped for walkers, joggers, cyclists and so forth. There are many trees, shrubs, and ponds as well. Basically, a great place to get together.

We had nice warm weather. My husband, Melanie, and I were at the park by 8:00 a.m. setting tables and putting out pastries, fruit, coffee, juice, and bagels. I couldn't find a guest speaker to attend our walk, but that turned out to be just fine. I gave a small speech, thanked everyone for coming, and introduced Linda Gentner to the group. She received a well-deserved round of applause. People started arriving around 9:30 and immediately started digging into the pastries. I had hoped there would be people with HSP attending, besides my children, but either they didn't know about it or couldn't make it. I was so glad Linda Gentner was able to attend. She brought at least 20 people to our walk. There were about 55 people altogether. My family and friends came through for me and were there to support us.

Raleigh, N. Carolina - September 12th-13th

Sarah Witt, Chair

The second weekend in September was one for the ages here in Raleigh. We started off on Saturday with a Quest for the Cure Seminar. Dr Colin Bishop of the Wake Forest Institute of Regenerative Medicine was our first speaker.

He gave an incredible and hope-filled presentation on the state of stem cell research. Our next speaker was acupuncturist Brian Kramer, who presented the theory behind Chinese medicine and discussed the ways in which acupuncture can be used to manage pain and spasticity. Nina Tang Sherwood of Duke University returned this year, and compared and contrasted the various animal models currently being used to study motor neuron disease.

Saturday night, Cece Russell and the Witt family joined more than 3000 Raleigh-area residents for a fun evening at the soccer field. The Carolina Railhawks, a professional soccer team in the Triangle, played and beat The Puerto Rico Islanders 2-1. The Railhawks organization is donating a portion of its ticket proceeds from that game to the SPF.

Sunday was The Magnificent Mile, which was phenomenal. We had 803 participants this year, who were joined by a couple hundred spectators. Cece Russell, Judy West and family, the Rocheleaus [Jim, Shea, 4-year-old Nathan, and 2-year-old Brady], and the Witts proudly represented the SPF. The men's and women's Competitive Mile races were thrilling as always. Both our defending champs were again victorious. Bobby Mack won in a time of 4:10 and set a new state record in the process. On the women's side, winner Kasia Sujkowski crossed the finish line in 5:03, a mere half-second ahead of her closest competitor. Nearly 400 joggers and walkers came out for the Recreational Mile, and 70 kids ran in our quarter-mile and 100-yard dash competitions. When all is said and done, we significantly raised awareness of PLS and HSP in the greater Raleigh area and secured a nice chunk of change for SPF's research fund.

Mt Kisco, New York - September 13th

Ann Lakin, Chair

The weekend started with SPF having a table at Mt. Kisco Sale Days. The table was manned by Helen, Joe, Chris, Kathy Kienlen, Ann (Marie) Kienlen-Lakin, and their parents Joan and Jack Kienlen. Ann's daughter Laura and husband Len helped out also. On Sunday we had the TeamWalk in beautiful weather, sunny and cool. We had between 10 and 15 more people than last year. The gathering started around 11:00 am. People had a chance to socialize and purchase raffle tickets. We had double the number of raffles we had last year. Despite the poor economy, we received generous raffle-item donations from local businesses. Around noon, we took our walk to the end of the street then through the parking lot back to our original destination. I'm sure we made quite the picture with all our crisp white shirts moving along the street.

After the walk we returned to our original spot and called the raffles. The tables were set up near the Mt. Kisco train station. There was a lot of foot traffic there because as people were going to Mt. Kisco Sale Days, and we got many donations as folks walked by. I'm sure a lot more people became aware of SPF because of that. All in all it was a very successful day.

Nashville, Tennessee - September 19th

Jim Sheorn, Chair

We had 16 people participate, which was a nice increase from last year. To date, $8,080 has been turned in. We are still anticipating about $1,000 to come, which will bring our grand total to more than $9,000. We had a great presentation from two physical therapists,

one of the best I have seen. They educated participants about HSP, PLS, and what can be done to improve mobility. It was like a HSP/PLS 101 class with an emphasis on Physical Therapy. Following that, we had one of our long time Connection participants, Terri McDonough, share her experience with Bioness. She has tried the device with one leg. Next week, she will try two at the same time. It looks like some insurance companies are starting to reimburse for it. Unfortunately, our key note speaker, Dr. Peter Hedera from Vanderbilt could not join us but he did send highlights of the research that he is currently doing with HSP. That information and the physical therapy presentation will be posted at SPF's web site. Overall, it was a big success. Each year, we hope to increase both the number of participants and the amount of money raised. I communicated with close to 100 people via email and regular mail, which motivated some to be Walkers by Proxy. In fact, part of the grand total was sent in by one such person.

Females of all ages prepare to compete at The Magnificent Mile

Boston, Massachusetts September 20th

Jim Campbell, Chair

Everyone had a fantastic day at the New England TeamWalk at Fenway Park. 81 people from the SPF community, friends and families participated. It was a crystal clear day in the 70s. We had box lunches and thanked the Red Sox Foundation and Covidien VP Matt Gattuso (whose sister has PLS) for making this event possible. After lunch, all participated in stretching exercises ably led by SPFer Liz Wrobleski. We then had our TeamWalk, which was a guided tour of Fenway Park (donated by the Red Sox Foundation)... each wearing our new Red Sox hat (donated by Covidien). We learned lots of interesting trivia about this historic 97 year old park.


A Big SPF Crowd in Utah

Over $50,000 in donations was raised by New Englanders for this event, Jim Campbell, chair of the TeamWalk reported. This year's committee consisting of Thurza Campbell, Matt Gattuso, Kathi and Ed Geisler, Ed and Joan Gilroy, Maureen Hall, Judy and Carl Johnson, Joel and Bobbie Seidman and Bob and John Swain was active in both event planning and soliciting donations averaging over 40 solicitation letters or email per family.

A memorable footnote to the day is that police, FBI and bomb squad people were all over the place. A Ben Affleck film "Town" was being shot in another part of the ball park. All of the uniformed officers we saw standing around were actually actors!

New England TeamWalkers listen to Chair, Jim Campbell after lunch

Pleasanton, California September 25th-26th

Linda Gentner, Chair

This year, 86 people found their way to a very sunny Pleasanton. Our Welcoming Dinner on Friday night brought 18 of us together. We had a nice dinner and then adjourned for chocolate cake, which was decorated with our SPF logo, and beverages. People put a lot of thought into asking questions and expressing concerns that were on their mind.

Saturday was hotter than it ever has been for our TeamWalks (approximately 100 degrees) so thank goodness we walked in the morning. After our walk, we had a nice luncheon and raffle. Jean Chambers and I talked about how connected people seemed this year with a "nice to see you again", "how are you


New England TeamWalkers in the Fenway Park press box

California TeamWalk

TeamWalkers in Pleasanton, California

Sue Meholick & Helen Kienlen in Pennsylvania

The Pennsylvania Teamwalk Turns 3

doing since last year?" Newcomers sought out others and compared symptoms. People seemed to have formed real bonds. We had 12 first-timers and many folks were eagerly inquiring about next year by the end of the weekend. I was amazed at how many raffle prizes we ended up with. The EVERYTHING CHOCOLATE basket drew many raffle tickets. Kris Brocchini won that one. If dark chocolate is medicinal, she should be well on her way to recovery. We also had lots of wine, golf packages, toys, and very unique items that brought in well over $1,000. Our community members included: Debbie Anderson, Scott Athearn, Ken Auer, Ryan Banks, Kris Brocchini, Rick Buchanan, Jean Chambers, Diane Dobrowolski, Angela Dixon, Malin Dollinger, Linda Gentner, Susan Jeiven, Jane Mitchell, Adele Pence, Cheryl Schmidt, Susan Stendahl, and Linda Worthington. We also made the official announcement that the SPF 2010 National Conference will be held in Los Angeles at the LAX Hilton on May 21-22-23, 2010 weekend. Start making your plans.

Knoebel's Park, Pennsylvania September 26th

Helen Kienlen, Chair

The Pennsylvania TeamWalk celebrated its third birthday this year. It turned out to be a great day despite the cold weather. We had 15 people for this event and we got to exchange stories and ideas for the roadblocks we face in life. The supporters enjoyed the lunch provided by the park. After lunch, the walkers participated in a joyful raffle. The items to be raffled included a basket containing various sauces and salsas, a gym bag containing a Frisbee and t-shirt, a basket of various beauty products, and a painting from a local artist. All the winners were very appreciative. After the raffle, the walkers participated in an invigorating and refreshing walk through the park. After the walk, supporters of this event were free to do whatever they liked. The walk was very enjoyable and the park provided a great venue for SPF as well as a kind and helpful security staff.

The remaining TeamWalks and Autumn in Carolina will be reported in the January issue of *Synapse.*

Upcoming Events

A Southern California Connection will be held on Saturday November 21, 2009, from 12 noon to 3 PM. Location and further details will be announced shortly, likely in the Los Angeles area near a freeway exit. We would be pleased to greet persons with PLS and HSP, and their family and friends, for sharing of our experiences, our problems, our victories, and ways of dealing with the obstacles we face. We also wish to meet "new people" with recent diagnoses, as well as people who recently became aware of the Spastic Paraplegia Foundation. If you are interested in attending, please email Malin at malind@cox.net or call him at (310) 378-4059. There will also be people attending from "outside" Southern California, e.g. Australia and Northern California, so we welcome you and encourage you to come, even if a bit of travel is involved.

In your message, please give your name, address, phone number, and email address, so we can give you specific information about the location. Tell me also who your "significant others"/ children are, and anyone who might attend with you. Giving this information is not making a reservation, but simply puts you on the "mailing list" so we are sure everyone knows about this. Knowing how many may be attending also helps me to plan."

SPF Grant Summary

Year	Recipient	Title	Amount Of Grant	Duration Of Grant	Summary of Findings	For
2003	John K. Fink, M.D., Director, Neurogenetic Disorders Clinic, University of Michigan	"A Molecular Genetic Analysis of Primary Lateral Sclerosis".	$40,000	1 year	The project sought to determine if one of the binding partners of the protein Alsin (defective Alsin causes juvenile onset PLS, ALS and HSP) is Atlastin (defective Atlastin causes juvenile onset HSP). Although several proteins were discovered that interacted with Atlastin in a yeast model, Alsin was not one of them.	PLS
	Douglas A. Marchuk, Ph.D., Associate Professor of Molecular Genetics and Microbiology, Duke University	"A Mouse Model of Hereditary Spastic Paraplegia"	$40,000	1 year	Developed a line of mice into which the mutated, human gene KIF5A was incorporated. (Mutated KIF5A causes HSP.) These mice developed several symptoms consistent with human HSP. These mice were then cross-bred with another line of mice that had been developed by Dr. Larry Goldstein at the University of California, San Diego that had only one copy of the KIF5A gene. This was done to attempt to develop mice that developed more severe HSP-like symptoms and at an earlier age. By the end of the period of grant funding, those mice were too young to have developed symptoms.	HSP
2004	Vincent T. Cunliffe, Ph.D. and Jonathan D. Wood, Ph.D., Centre for Developmental Genetics and the Academic Neurology Unit of the University of Sheffield, UK	"Modeling the neuro-degenerative processes caused by mutation of the SPG4 gene in zebrafish and development of strategies for pharmacological intervention".	$90,000	2 years	Discovered a crucial role for Spastin (defective Spastin causes the most common type of adult onset HSP) in promoting axon outgrowth in the zebrafish embryo. The discovery was published in the high profile journal Human Molecular Genetics, and the journal also included an image from the paper on the cover of the issue in which the article appeared, further enhancing the visibility of the research.	HSP
	Dr. Teepu Siddique, the Director of the Neuromuscular Disorders Program at the Feinberg School of Medicine at Northwestern Univ.	"PLS Registry"	$90,000	3 years	Expanded the PLS Registry to include blood specimens from 214 PLS patients, including 118 sets of patients with an unaffected sibling, and 33 sets with a patient and both parents.	PLS
2005	Michael R. Hayden, M.D., Ph.D. and Blair R. Leavitt, Ph.D., Center for Molecular Medicine and Therapeutics, University of British Columbia, Vancouver	"Revealing the mechanisms underlying ALS2, a form of hereditary spastic paraplegia, using ALS -/- mice".	$149,896	2 years	Discovered that in flies, Spastin and Atlastin proteins act together in microtubule dynamics regulation to control neuron structure and function.	HSP and PLS
	Brett Peter Lauring, M.D., Ph.D., Department of Pathology, Columbia University College of Physicians and Surgeons, NYC	"Analysis of Spastin and Atlastin in the cell biology of neurons".	$96,701	2 years	Confirmed that loss of Spastin function results in prematurely terminating motor axons in C. elegans. Developed a line of C. elegans with mutated human SPG4/ Spastin genes. Published the results of his work in the July, 2006 issue of the The Proceedings of the National Academy of Science, the March, 2007 issue of the Journal of Cell Biology, the July, 2007 issue of the Journal of Cell Science, and the December, 2007 issue of the journal Traffic.	HSP
	Peter Hedera, M.D., Department of Neurology, Vanderbilt University, Nashville	"Invertebrate model of hereditary spastic paraplegia"	$90,000	2 years	Created a line of C. elegans (a microscopic worm) with HSP caused by mutations in their SGG6/ NIPA1 gene, and another line of these worms with HSP caused by knocking out their SPG2A/ Atlastin gene. Determined that HSP caused by mutations in the SPG6/ NIPA1 gene is associated with accumulation of misfolded NIPA1 protein, which triggers neuronal degeneration and programmed cell death. In a line of C. elegans with mutant SPG6/ NIPA1 genes and a mutation in another gene that causes the worms to be resistant to programmed cell death, observed that the disease course was slower and less severe. Published findings in the December, 2008 issue of the Journal of Neuroscience.	HSP
	Kendall S. Broadie, Ph.D., Department of Neurobiology, Vanderbilt University, Nashville,	"Mechanistic interactions among hereditary spastic paraplegia genes"	$54,673	1 year	Discovered that in flies, Spastin and Atlastin proteins act together in microtubule dynamics regulation to control neuron structure and function.	HSP
	Jeffrey Macklis, M.D., Director of the Massachusetts General Hospital--Harvard Medical School Center for Nervous System Repair, Boston	"Molecular genetic controls over the development, connections, and survival of upper motor neurons"	$121,660	2 years	Dr. Macklis's work for the SPF is ongoing. He is investigating the molecular controls involved in the development, connectivity and survival of motor neurons in the central nervous system. Has published eight medical journal articles on his groundbreaking work to date.	PLS

SPF Grant Summary

Year	Recipient	Title	Amount Of Grant	Duration Of Grant	Summary of Findings	For
2006	John K. Fink, M.D., Director, Neurogenetic Disorders Clinic, University of Michigan	"Developing treatment for childhood onset hereditary spastic paraplegia (SPG3A HSP)"	$120,000	2 years	Dr. Fink's work for the SPF is ongoing. Results have not been published or made public.	HSP
	Jeffrey D. Macklis, M.D., D.HST Dir. of the Harvard Stem Cell Institute, MGH– Harvard Medical School Center for Nervous System Repair. Paola Arlotta, Ph.D.,Assistant Professor at the Harvard Stem Cell Institute and the MGH Center for Regenerative Medicine	"Molecular Controls over the Development, Connections, and Survival of Upper Motor Neurons"	$125,000	1 year	Drs. Macklis's and Arlotta's work for the SPF is ongoing. They are investigating the molecular controls involved in the development, connectivity and survival of motor neurons in the central nervous system.	PLS
	Nina Tang Sherwood, Ph.D. Assistant Research Professor, Duke University	"Understanding the ameliorative effects of temperature in fruit fly models of AD-HSP"	$120,000	2 years	Dr. Sherwood's work for the SPF is ongoing. Results have not been published or made public.	HSP
	Kendall S. Broadie, Ph.D., Department of Neurobiology, Vanderbilt University, Nashville	"Mechanistic interactions among hereditary spastic paraplegia genes"	$57,070	1 year extension of '05 grant	Discovered that in flies, Spastin and Atlastin proteins act together in microtubule dynamics regulation to control neuron structure and function.	HSP
2007	Bruce Horazdovsky, Ph.D., Associate Dean of the Mayo Clinic College of Medicine, and senior consultant to the department of biochemistry and molecular biology and the Mayo Clinic Cancer Center (Rochester, MN)	"Development of a cell culture system to analyze defects associated with Primary Lateral Sclerosis"	$58,000	1 year	Dr. Horazdovsky and his team's preliminary results defining Alsin's role in IGF-1 signaling and receptor trafficking have been submitted for publication and they report that they are confident that we will gain new insights into the role Alsin plays in motor neuron maintenance.	PLS
	Peter W. Baas, Ph.D., a professor in the departments of neurobiology and anatomy, in the College of Medicine, at Drexel University (Philadelphia, PA)	"Mechanistic Basis of SPG4-based Hereditary Spastic Paraplegia"	$140,000	2 years	In Dr. Baas's published works referencing the SPF as sources of funding, he and his team explore an alternative theory for how the defective Spastin protein expressed by mutated SPG4 genes can cause axonal degeneration in adults through a novel gain-of-function mechanism. Baas proposes that the mutant spastin protein may be toxic to the axons, and cause them to degenerate due to this toxicity. Understanding the molecular function of Spastin in a neuron is critical in order to intelligently design therapies and cures for HSP in our lifetime.	HSP
	Stephan Zuchner, M.D., the director of the Center for Human Molecular Genetics at the Miami Institute for Human Genetics in the Leonard M. Miller School of Medicine (Miami, FL)	"Molecular and genetic analysis of the SPG31 gene REEP1".	$135,561	2 years	Dr. Stephan Zuchner's group at Human Genetics used SPF funding to further characterize the REEP1 (SPG31) gene. Zuchner's group will provide a complete report on the findings to the SPF in early 2010. Understanding the molecular function of REEP1 could help guide the design and testing of therapies and cures for HSP.	HSP
2008	Paola Arlotta, Ph.D., Center for Regenerative Medicine at Massachusetts General Hospital (Boston, MA)	"Directed Differentiation of Neural Progenitors and iPS Cells into Corticospinal Motor Neurons".	$60,000	1 year	Dr. Arlotta's work for the SPF is ongoing. Results have not been published or made public.	HSP and PLS
	Janine Kirby, Ph.D., Christopher John McDermott, Ph.D., and Pamela Shaw, Ph.D., Academic Neurology Unit at the University of Sheffield Medical School (Sheffield, United Kingdom)	"Elucidation of upper motor neuron vulnerability in Primary Lateral Sclerosis".	$103,375	2 years	Drs. Kirby, McDermott, and Shaw's work for the SPF is ongoing. Results have not been published or made public.	PLS
	Yasushi Kisanuki, M.D., Department of Neurology at Ohio State University (Columbus, OH)	"Paraplegia in HSP Rat: Analysis and treatment".	$120,000	2 years	Dr. Kisanuli's work for the SPF is ongoing. Results have not been published or made public.	HSP
	Jeffrey Macklis, M.D. D.HST., and Paola Arlotta, Ph.D., Harvard Medical School Center For Nervous System Repair at Massachusetts General Hospital (Boston, MA)	"Molecular-Genetic Controls over the Development, Connections, and Survival of Upper Motor Neurons".	$120,560	2 years	Dr. Macklis's work for the SPF is ongoing. His most recent results have not been published or made public.	HSP and PLS

disAbility Awareness Day

Inspired by and Contributed by Ronnie Grove

Setting up Friday after 9 months of planning, it was like having a baby---the size of an elephant. Saturday, September 12 was like most days the day after birth; pure joy and happiness. What a day!

I never did get a count but I would say we had between 20 and 30 vendors set up from many different sources. With the vendors, visitors and volunteers it is hard to say exactly how many attended this fabulous even but I would hazard a guess that there were 100 people present at all times throughout the day. We had use of the local high school gym with adjoining cafeteria and across the hall the auditorium. There was something going on at all three places all the time. I had the TTY and a phone set up for relay calls, one blind person was showing his talking paper money changer and an eleven year old girl who is severely visually impaired by Aridia---the absence of the iris of the eye---was showing her Braille writer. We had two accessible mobility dealers. One had more walkers, scooters and wheelchairs than I could count. The Lions Club brought a black lab named Bernie that is learning to be a service dog. I think he was the hit of the show. Also, in the vision section we had the gadget that you wear to show what low vision is for normal people. There were lots of visitors at this table.

I could not believe how many handicapped people came out for the event. We even had a handicapped dog. It was a very small dog whose back legs did not work and it was in one of those little carts. We have a group in our community that does religious puppet shows. They put on a fabulous show called "It's No Fun Being Left Out." The show was a hit with the kids and adults.

Everything was just grand. I know we made a difference. The day, without a doubt, was a huge success. People came, they saw, they learned and that pleased me very much. I think there was just too much to see and do and not enough volunteers to be every where that we needed them. (We also learned a lot). I know there is more to come.

I learned there was a man from Winchester (40 miles away) who came specifically to learn about Dawn O'Leary's bionic hand. That alone made it all worthwhile. As I looked around that gym on Saturday one thing came to mind: Steve Urkel's famous line---"Did I do that?"

Follow-up editorial from The Morgan Messenger

Different Abilities Rely On Strength Of Spirit - 9/16/2009

Most of us will wake up tomorrow, get dressed and head off to work or school without hesitation. Many of us will go about our daily routines with a few aches and pains that go along with the aging process, or a cold that will disappear in a day or two. And then there is that percentage of the population that faces each new day with a special physical challenge. For some, it is a limb that works differently; for others, it is a medical condition that requires daily monitoring, tests and injections. Some disabilities are temporary and others are life-changing. Some of our neighbors are born with their disability, yet others develop them by illness or accident. Regardless, disabilities and the challenges they bring don't discriminate — they strike newborns, the aged and everyone in between.

For the last few weeks *The Morgan Messenger* has featured stories about people from the area living with their disabilities. For every story that was told, there are hundreds that weren't. The newly formed group, Morgan County Advocates for disAbility Awareness, held their first disAbility Awareness Day last Saturday. Not only was the day full of activities for those with a disability, but for their caregivers, families and friends. The group's main goal is to make everyone aware of the resources available to those in the area, and to make those without disabilities aware of the special challenges that face those whose abilities are changed by disease, heredity or circumstance. Their message is: "Having a disability doesn't change who you are...it only changes what you can do." We could all learn from their message. We should be grateful for the things we can do — for ourselves and for others, and be mindful that a strong spirit can make up for any shortcomings of our bodies.

We were inspired by the stories of our neighbors who exhibit such strength of spirit, and hope that you were, too. And we applaud those organizers who have brought this issue to the forefront of our minds.

National Super Kid's Classic Soapbox Derby

Contributed by Carrie Cottrill

My son, Nickolas Beekman has become a local celebrity!! He has won the local Super Kid's Classic Soapbox Derby in Lancaster, Ohio. At the National Super Kid's Classic in Akron in July, Nickolas placed 6th out of 56 kids. He actually got to go up on stage and get his trophy. The top six kids got to be included in the awards ceremony. We had a great time. Nick enjoyed it very much!

Here is their website information. www.nationalsuperkids.com. A group of Vietnam veterans in 1980 in Akron, volunteering at the All-American Soap Box Derby decided to start races for special needs kids. Local races have gone on in many places since. The local Super Kids Classic committee participants and board members, while working on the annual event year after year, also had a much bigger dream - a national race in which youngsters who were not able to participate in the All-American Soap Box Derby race could experience a race of their own. On August 9, 2003, the National Super Kids Classic in Akron, Ohio became a reality. The same hill, the home of the All-American Soap Box Derby, known as Derby Downs, became the home of the National Super Kids Classic.

Although Nick is in a wheelchair because of his HSP/FSP he is still treated like a typical child. They are all calling him "Champ." These people are fantastic and the experience has been one that we will remember for a long time.

Giving up your Keys

Contributed by Thurza Campbell

I hope I might be able to encourage HSPers and PLSers to think outside the box regarding when it is time to give up your driver's license. I write as a woman who voluntarily gave up her license in my early sixties (4 years ago). I have PLS, as many of you know. I realized I no longer had the coordination to quickly slam on the brakes or spin the steering wheel in time to react in an emergency. I've had to make many adjustments in my daily routine, but I have not become housebound. I depend heavily on my husband Jim, for driving our handicapped car to installation sites of our landscape design business. He and others help with errands. I primarily shop on line.

It seems to me that people consider driving a right. I believe it is a privilege we earned as a teen but now should objectively consider if we should each be behind the wheel. Here are some considerations which might not have occurred to you. Consider the financial rewards for not driving. Add up the annual cost to purchase or lease a car + insurance + maintenance + gas. For that total you spend annually, you can use local transportation or even taxis for much less.

Giving up your license is only one of life's many transitions and adjustments. It doesn't have to mean total loss of independence. It means initially, objectively acknowledging to yourself your physical ability. How quickly can your reflexes, eyesight and hearing react in an emergency? Then using your life experience, creativity and maturity think through how you will adapt your routine without your license. The life that might be saved by an accident prevented is very precious to someone.

How to Get your Genetic Profile

Contributed by Jennifer Thomson

I'm wondering how many might be interested in getting their genetic profile done. I had mine done through www.23andme.com for $399. You just spit in a tube (they send you the kit), they extract your DNA from your saliva, analyze it and deposit the resulting genetic data in your account at www.23andme.com. You can then review your profile, compare it to others, see what diseases you might be a carrier for, have a greater risk, or a decreased risk of getting. You can find information about drug reactions based on your genotype. It is really interesting.

Here are examples: SNP rs10260404, my genotype is CC: This study examined 1,767 Europeans with ALS and 1,916 healthy controls. The authors found that each C at rs10260404 increased a person's risk for the sporadic form of ALS by about 1.3 times compared to those with the TT genotype. SNP rs7538876, my genotype is GG:

This study compared 2,137 basal cell carcinoma (BCC) patients with 35,921 healthy controls from Icelandic and Eastern European populations and found that rs7538876 in the PADI6 gene is associated with the disease. Having an A at both copies of rs7538876 increased a person's odds of developing BCC about 1.3 times compared to the AG genotype; the GG genotype decreased a person's odds about 1.3 times. SNP rs762551, my genotype is AC:

Caffeine is primarily metabolized by the liver enzyme cytochrome P450 1A2 (CYP1A2). The form of the SNP rs762551 a person has determines how fast CYP1A2 metabolizes caffeine. In this study, people with the slower version of the CYP1A2 enzyme who also drank at least two to three cups of coffee per day had a significantly increased risk of a non-fatal heart attack. The study found that fast metabolizers, on the other hand, may have actually reduced their heart attack risk by drinking coffee. You can also use it for following ancestry lines, compare your profile with others in your family, etc.

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Gawkers in a Restaurant

Contributed by: Maryann Mayer, Ohio Ambassador

My brother who also has HSP came to visit this weekend. We went out to eat Friday evening. Imagine this. My mom is 87, walks bent over almost at a 90 degree angle and uses a walker. Then there's my brother with his cane who really has a hard time lifting his legs and uses his wife's shoulder along with his cane to walk. Then there's me with my cane and my HSP limp on the right. And of course my husband, who, thank goodness has no problems. Later on that evening my husband told me how the people who were sitting at the table next to us were gawking at us when we got up to leave. He said the stares followed us all the way out the door as our parade processed out. I used to feel all those stares and they would cause me to stiffen more and walk even worse. Friday, I didn't even notice. It takes time. Fortunately there are those who are more helpful and open doors or offer their help, etc.

"Never be bullied into silence. Never allow yourself to be made a victim. Accept no one's definition of your life; define yourself."

~ Harvey Fierstein ~

"Reading Rocks!"

Contributed by Joan Burton (HSP); excerpted from an interview on CKWS TV, St. Louis

Photo credit: Norman Wong

When she was just 12 years old, Melanie Burton helped create "Reading Rocks!"-- a literacy program aimed at helping young kids learn to read. This is how Moira secondary student Melanie Burton spent her summer, teaching young kids how to read. Six years ago there were just 11 kids in the group. Today, the "Reading Rocks!" program helps 40 to 60 kids a year. It's such a success -- Belleville's city council recently honored Melanie for all her volunteerism. "It's a lot of fun. It was never anything I expected or was aiming for."

So accomplished are Melanie's efforts *Time Magazine* even picked her as one of 30 volunteers honored before major league baseball's All-Star Game in St. Louis. But she says, "despite all the praise, it's still all about teaching young kids how to learn to like reading - especially the kids who come in and who look at you like you're crazy when you say you're going to read 100 books. They're like 'that's not going to happen lady.' Then you're celebrating with them and they've read 500 or more."

Completely free of charge for young minds, the "Reading Rocks!" literacy program is 100-percent volunteer driven. Melanie's efforts have even encouraged other teens to give back. "The biggest thing for me is getting to see how other people are volunteering in their communities at all different ages. So it's not something that has to stop because I'm away in college."

Caregiving and Medical Emergencies

By Linda Foster, MA
Medically reviewed by Lindsey Marcellin, MD, MPH

Caregiving can be a demanding and intimidating responsibility, especially when you know the person you are taking care of might one day be facing a medical emergency. Should that happen, the situation will go more smoothly for you as the caregiver and for your loved one if you're prepared in advance and know exactly what you'll need to have handy and what steps you'll need to take.

4 STEPS TO TAKE IN AN EMERGENCY SITUATION

Keeping your wits about you will be key, should an emergency arise. Practice these steps so that you'll know exactly what to do in an emergency:

1 ASSESS THE SITUATION. Is your loved one bleeding or did he have a blow to the head, a fall, or an allergic reaction? Is he experiencing any symptom specifically related to his illness? What were you both doing just prior to the emergency? Is he responsive? Are his pupils enlarged and are they the same size? Was there a complaint of pain or anything else relevant? What is different or unusual about your loved one? "Observe, observe, observe," says Guerra in anticipation of calling 911.

Caregiving for someone who is seriously or chronically ill is a daunting task, and an emergency can be frightening. Taking steps in advance will help you handle a crisis, should one occur.

2 CALL 911. Do this when you have the even slightest hint that your loved one is facing a life-threatening emergency. "A call to 911 is the best thing in any emergency," says Guerra. Do not attempt to take anyone with a potentially serious problem to the hospital yourself; instead, call 911 immediately. Give the 911 operator as much information as you can, so that emergency personnel can be fully prepared to assist your loved one when they arrive, says Guerra. It's crucial to accurately describe the situation and speak slowly and clearly when talking with the 911 operator. Mention any pre-existing conditions, such as a history of heart attack, diabetes, a bleeding disorder, or asthma.

3 LOOSEN ANY TIGHT CLOTHING. Make sure your loved one has nothing constricting the airways, like a restrictive shirt or tie, and keep them in a comfortable position while you wait for help.

4 COMFORT AND COMMUNICATE. Talk to your loved one until 911 arrives. Guerra recommends that you keep your loved one awake by talking to them, but don't encourage them to talk. Take slow, deep breaths to help yourself stay calm as well.

For a caregiver, an emergency can be both alarming and frightening, but when you're prepared, you can make a tremendous difference in your loved one's well-being.

5 BASICS STEPS TO EMERGENCY PREPAREDNESS

Basic training and first aid supplies will go a long way toward giving you a greater comfort level as a caregiver:

1 TAKE A CPR CLASS. You can use CPR to revive someone whose heart has stopped beating or who has stopped breathing. Bill Guerra, RN, BSN, of the Seven Hills Surgery Center in Henderson, Nev., recommends that all caregivers take a CPR class so you understand the ABCs of emergency response: **A** *(airway*), **B** *(breathing)*, and **C** *(circulation)*.

2 LEARN THE HEIMLICH MANEUVER. CPR training also involves learning the Heimlich maneuver — how to clear someone's airway in the event a foreign object or food becomes lodged in the throat. Attempting the Heimlich maneuver without proper training can injure your loved one. Visit the American Heart Association Web site to learn about the Heimlich maneuver and where to take a CPR class. Also, find more on the warning signs of heart attack and stroke.

3 MAINTAIN A WELL-STOCKED FIRST AID KIT. Thoroughly read the manual as soon as you buy your first aid kit. Check monthly to make certain you have adequate supplies, and replace any items that may have expired. An ear thermometer may be the best choice if your loved one might accidentally bite down on an oral thermometer. Keep a 2nd first aid kit in your car; keep both out of the reach of children.

4 CREATE A MEDICAL PROVIDER LIST AND KEEP COPIES HANDY. "Always have all doctors' numbers in a central, convenient place; put one copy of the list in your purse or wallet and one on the refrigerator. Include a list of all medications and other health facts and conditions. Make another list of family members to notify," says Guerra. When applicable, have phone numbers of people who can watch your children or pets and secure your home, should you need to take your loved one to the emergency room.

5 BUY AN AUTOMATIC BLOOD PRESSURE CUFF. You can buy a good one at any local drug store. Learn how to use it and practice using it regularly. On regular doctor visits, take the cuff with you to check its accuracy against the physician's blood pressure monitor and to ensure you are using it correctly.

Medical Research

Betsy Baquet, editor

Neuralstem Receives FDA Approval To Begin First ALS Stem Cell Trial

Neuralstem, Inc. announced that the FDA has approved its Investigational New Drug application to commence a Phase I trial to treat ALS with its spinal cord stem cells.

"We are very excited about this clinical trial," says Dr. Eva L. Feldman, M.D., Ph.D., director of the University of Michigan Health System ALS Clinic and the Program for Neurology Research & Discovery. "This is a major advancement in what still could be a long road to a new and improved treatment for ALS." "In work with animals, these spinal cord stem cells both protected at-risk motor neurons and made connections to the neurons controlling muscles. We don't want to raise expectations unduly, but we believe these stem cells could produce similar results in patients with ALS," Dr. Feldman concluded.

This first trial, which will primarily evaluate safety of the cells and the surgery procedure, will consist of 12 ALS patients with varying degrees of the disease. The patients will be examined at regular intervals post-surgery, with final review of the data to come about 24 months later.

Neuralstem CEO and President Richard Garr stated "While this trial aims to primarily establish safety and feasibility data in treating ALS patients, we also hope to be able to measure a slowing down of the ALS degenerative process."
SOURCE: http://www.alsa.org/news/article.cfm?id=1510&CFID=4268356&CFTOKEN=7ebacfda26251835-FDD87E2A-188B-2E62-80B23D81567888AD; http://www.als-mda.org/research/news/090923als-neuralstem.html

Scientists Discover Faster Protein Aggregation Means Faster Disease Progression in ALS

A team of researchers have discovered that ALS progresses faster in patients whose gene mutations cause faster clumping of the SOD1 protein, linked to approximately 2 percent of all ALS cases. Aggregation – or clumping – of the SOD1 protein is believed to be a crucial step in the disease process.

The finding suggests that aggregated proteins may elude normal cellular "housecleaning" methods, or their formation is heightened by stress conditions in the cell. As people with ALS begin to experience symptoms, the buildup of protein is rapid and dramatic. However, it is well established that significant damage to the nervous system occurs well before the patient has any symptoms.

"By linking speed of aggregation with the rate of disease progression, this study tells us that interrupting aggregation with drugs is likely to lead to a viable treatment strategy. If we can target drugs to stop the aggregation, we can stop the progression of ALS," said Lucie Bruijn, Ph.D., senior vice president of Research and Development at The ALS Association.
SOURCE: http://www.alsa.org/news/article.cfm?id=1491&CFID=4268356&CFTOKEN=7ebacfda26251835-FDD87E2A-188B-2E62-80B23D81567888AD

Protective Gene Enables People with ALS to Live Longer, Study Finds

A new genetic discovery may help researchers understand factors that improve survival in people who have ALS. The discovery also strengthens the theory that changes in cellular transport contribute to the death of motor neurons.

Every person carries two copies of the gene called the KIFAP3 gene, which plays a significant role in the transport of cellular cargo. The researchers found that patients whose two genes both contained a certain type of genetic material called "C form," lived about 14 months longer than those whose two genes were both of the "T form."

The study was carried out in 3,000 patients with "sporadic" ALS, those for whom no genetic cause or family history is known. About 90% of all ALS is sporadic. The researchers showed that the KIFAP3 gene forms did not influence the risk of developing ALS, only survival once the disease began.

"This discovery is important, because it provides an important clue about progression in ALS," said John Landers, Ph.D., of the University of Massachusetts Medical School. "As we learn more about this gene, we may be able to develop therapies that mimic the protective function of this newly discovered variant, which may benefit all ALS patients."
SOURCE: http://www.alsa.org/news/article.cfm?id=1484&CFID=4268356&CFTOKEN=7ebacfda26251835-FDD87E2A-188B-2E62-80B23D81567888AD

NIH Cuts Short ALS Lithium Trial; MDA's Trial Continues for Now

The investigators on a study of lithium carbonate in ALS, funded by the National Institutes of Health, the ALS Association and the ALS Society of Canada, announced Sept. 23, 2009, that they will stop their study after an interim analysis showed the drug was not beneficial. MDA is not a funder of this study.

The MDA-supported study of lithium (See Lithium Open-Label Study) will remain open for the time being. The MDA trial is separate from the canceled NIH trial, and the dosages and trial design are not the same.

Clinical trials of the effectiveness of lithium in ALS were undertaken after a small study in Italy was published in 2008, reporting that lithium appeared to dramatically slow the course of the disease.
SOURCE: http://www.mda.org/publications/Quest/extra/sept09/als-lithium.html

Enhancing Cellular Cleanup Mechanism Helped ALS Mice Eliminating the XBP1 protein enhanced cellular cleanup, saved nerve cells and prolonged survival in female ALS mice

New research has shown that augmenting a process that naturally occurs in the nervous system to clean up and destroy cellular debris delayed onset and prolonged survival in female ALS mice. The cleanup process, autophagy, was enhanced by the absence of a protein called XBP1. This protein normally helps cells to respond to various types of physiological stress. To the surprise of researchers, eliminating this protein flipped a switch in the nervous system and caused an enhanced cleanup process. As a result, the female SOD1 mice developed ALS later, and lived a significant amount of time longer. The male SOD1 mice who were deficient in XBP1, however, were not helped.

Although there is no immediate benefit for patients, if the findings in this study are confirmed, it may prove worthwhile to develop strategies for ALS treatment that enhance autophagy.
SOURCE: http://www.mda.org/publications/Quest/extra/sept09/enhancing.html

Drug Offers Control for Laughing/Crying Outbursts Avanir's experimental drug Zenvia appears safe and effective for involuntary emotional expression in ALS

The pharamaceutical company Avanir has announced positive results for its phase 3 trial to treat unwanted episodes of laughing and crying in patients with ALS and multiple sclerosis using its experimental drug Zenvia.

Uncontrollable laughing or crying, sometimes called "pseudobulbar affect" (PBA) is thought to be caused by degeneration of nerve pathways from the upper brain to the lower (bulbar) brain.

Two dosage formulations were used in the trial, and Avanir reports both dosages significantly reduced episodes of PBA compared to a placebo. The drug was generally safe and well tolerated. The company intends to announce complete results from the phase 3 study this fall and to follow guidance from the U.S. Food and Drug Administration (FDA) to gain market approval for Zenvia to treat PBA.

SOURCE: http://www.mda.org/publications/Quest/extra/aug09/outbursts.html

Survival Gene: A new study shows a small change in the KIFAP3 gene lengthens ALS survival time.

A variant version of the gene for a protein known as KIFAP3 has been found to increase survival time in people with sporadic (nonfamilial) ALS by an average of 14 months.

They analyzed the genomes (entire set of genes) of 1,821 people with sporadic ALS and 2,258 without the disease from the United States and Europe. Included in the analysis was the survival time in 1,014 people who had died of ALS. The researchers found a single variant in the gene for KIFAP3 that significantly correlated with ALS survival time. People with ALS who had the survival-enhancing KIFAP3 gene variant on both copies of chromosome 1 survived an average of 14 months longer than people with this variant on only one chromosome 1 or on neither chromosome 1.

The investigators say they don't know why having less KIFAP3 protein would prolong survival in ALS, but they believe, based on the normal function of this protein, that its reduction may reduce transport of toxic molecules inside nerve fibers.

"Few genetic factors that modify ALS survival are reported," the authors write, noting that none have been identified in previous genome-wide association studies in ALS. "The identification of KIFAP3 as a determinant of progression rate of sporadic ALS is therefore promising."

SOURCE: http://www.mda.org/publications/Quest/extra/may09/kinesin.html

Michigan Tech scientists identify genes linked to Lou Gehrig's disease

Michigan Technological University researchers have linked three genes to the most common type of ALS. Professor Shuanglin Zhang, who suffers from sporadic ALS, leads the team of mathematicians that isolated the genes from the many thousands scattered throughout human DNA. He notes that their discovery does not mean an end to ALS, but it could provide scientists with valuable clues as they search for a cure.

SOURCE: http://www.eurekalert.org/pub_releases/2009-09/mtu-mts090909.php

Little-known protein found to be key player: 'Atlastin' builds critical structures; does job in fundamentally new way

HOUSTON -- (July 29, 2009) -- Italian and U.S. biologists this week report that atlastin, previously implicated in HSP, plays an unexpected and critical role in building and maintaining healthy cells. Even more surprising, their report in the journal *Nature* shows that the protein, does its work by fusing intracellular membranes in a previously undocumented way.

Using a range of tests on purified proteins, live fruit flies and cell cultures, researchers examined the effect of both an overabundance and a scarcity of atlastin on cell function and on fruit fly development. They also created mutant versions of the protein to see how it functioned -- or failed to function -- when some parts were disabled.

The tests showed that cells with extra atlastin had an overdeveloped endoplasmic reticulum (ER), a system of interconnected membrane tubes and chambers that's critical for normal cell function. The tests also showed too little atlastin led to a fragmented ER. Flies with defective atlastin were sterile and short-lived.

"We hope the findings lead to a better understanding of hereditary spastic paraplegia (HSP)," said Daga, a researcher at the Eugenio Medea Scientific Institute in Conegliano, Italy. Daga said atlastin's role in building and maintaining a healthy ER may help HSP researchers better understand why neurons are affected first.

SOURCE: http://www.eurekalert.org/pub_releases/2009-07/ru-lpf072909.php

From nerve roots to plant roots – research on HSP yields surprises

A new study from the National Institutes of Health and Harvard Medical School suggests that neurons and plant root cells may grow using a similar mechanism.

"This study provides us with valuable new insights that will stimulate research toward therapies for hereditary spastic paraplegias," says Craig Blackstone, M.D., Ph.D., an investigator at NIH's National Institute of Neurological Disorders and Stroke (NINDS) and an HSP expert.

In this study, researchers propose that a defect in the endoplasmic reticulum (ER) in both patients and a plant called Arabidopsis cause a common problem. Misshapen ER are a common cause of HSP, and causes impaired growth or maintenance of long corticospinal axons in humans. Similarly, defective ER in the Arabidopsis impairs the growth of the plant's root hairs, which are wispy, microscopic projections that grow from the plant's individual root cells.

Atlastin has previously shown to be defective in 10% of HSP cases and plays a role in axon growth. The new study shows that the Atlastin protein is also necessary for maintaining the shape of the ER.

Since Arabidopsis contains an analog of Atlastin, and this connection between axon growth and root hair growth withstands further study, Arabidopsis could be a useful tool for investigating mechanisms of HSP.

SOURCE: http://www.eurekalert.org/pub_releases/2009-08/nion-fnr080309.php

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