Synapse

Volume 17, Issue 2 Fall 2014

Newsletter of the Spastic Paraplegia Foundation

Letter from the President

Dear Friends,

Like so many of you, the injustice of HSP and PLS makes me very angry. As I see it, we can deal with this anger in one of three ways. First, we can ignore it and hope someone else will take some action. Worse yet, we can deal with it self destructively with drugs, liquor or self pity. But, there is another answer.

The best answer is inspired every day by so many people in our community. Families like that of 4 year old Lucas Bloodworth, who has HSP and at our recent annual conference had such a gentle glowing smile and loving spirit that my whole family fell in love with him. But Lucas, like so many of us, needs hope for a better tomorrow.

Augustine said "Hope has two beautiful daughters; their names are Anger and Courage. Anger at the way things are, and Courage to see that they do not remain as they are." The mission of Our Spastic Paraplegia Foundation is to help you and I use our anger at the terrible injustice of HSP and PLS to bring forward the courage to correct that outrage by making a tax deductible donation of any amount you can afford toward the hope for a cure. Every donation whether large to small is necessary.

The Spastic Paraplegia Foundation is your ladder of hope. The steps of that ladder have been proven strong as they lean against the wall of 12 years of solid performance. Since 2002, The Spastic Paraplegia Foundation, a 501c3 nonprofit foundation, has supported over 5 million dollars worth of research on HSP and PLS and remarkable progress keeps taking place. In our last Annual Report I listed three pages of just some of the scientific progress that has just recently been made.

This year alone we are funding over $800,000 worth of cutting edge research. We are supporting two actual drugs that could be the breakthrough. We are supporting stem cell technology that continues showing more and more real promise for correcting the atrocity of HSP and PLS. We are supporting finding just how the microtubules of your long neurons support or block the signals from your brain to move your legs correctly. The list goes on and on.

We are so lucky to be living at a time in history when answers to questions about genetics and upper motor neurology are so within reach. Answers are unfolding at a blistering pace but answers demand more answers and answers cost money. Please remember that everyone on our dedicated, skilled staff is volunteer and over 80% of our funding goes directly to support research. We use the very objective rating system of our world renowned International Scientific Advisory Board to know which of the proposed research projects we should support. Science is so ripe for the picking that in the last couple of years they have been rating many more proposals as *rock-solid excellent* than we can possibly afford.

Your past donations are very much appreciated. Thank you. If it has been a while since your last donation, please use your courage from anger at HSP and PLS to do so today. What if you could increase your donation this year by just 20%? What about 50 or 100%? What answers we could joyfully share at the table of hope for a cure? Please mail your tax deductible gift in the enclosed self addressed reply envelope today.

Sincerely,

Frank Davis

President, SPF

CHASING A CURE

Federal Employees are Willing to Support Spastic Paraplegia Foundation

The Combined Federal Campaign or CFC is a fundraising campaign the Federal Government offers its employees to participate with each year. It is like a United Way campaign where employees pick which non-profits they would like to support. The CFC starts the end of August and usually runs through December.

Each year **Millions** of dollars are raised through the CFC campaign. The bad news is that the Spastic Paraplegia Foundation doesn't get much of the money raised, usually around $5,000.

Your help is needed to ask the Federal employees you know to pick the Spastic Paraplegia Foundation to contribute to. The best way to get a commitment is to ask personally. If that is too uncomfortable, feel free to send a letter or email. The Spastic Paraplegia Foundation CFC number is **12554**. Federal employees will need to know that so they can designate the SPF.

The following are examples of Federal employees: law enforcement, mail personnel, VA or Veteran's Administration employees, Medicare, Medicaid, military and many types of governmental jobs. If donors want to know more, please have them log on to www.sp-foundation.org

Many federal locations have CFC Fairs during the Fall. They allow representatives from organizations to visit with employees and provide information. If you hear about one in your area and would like to help inform federal employees about the SPF, please let me know. I can get you the materials you will need. All you will need to do is provide the time and a friendly attitude.

If you have any questions, suggestions or would like to help with a CFC fair, please contact Jim Sheorn at jmsheorn@comcast.net or 615-479-7369.

Please help us generate more resources for research.

CFC®
Combined Federal Campaign

Synapse

Volume 17, Issue 2 - Fall 2014

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

Synapse Editor
John Staehle.............................Senior Editor

Published three times a year, Spring Fall and Winter, for the HSP/PLS community. It is also available online at www.sp-foundation.org

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www.sp-foundation.org

SPF Medical Advisor:
John K. Fink, M.D., University of Michigan

Connections

SPF Illinois Connection

May 10, 2014

HSP: Sid Clark   PLS: Hank Chiuppi

We had a great spring meeting. Margaret McGovern-Denk (MS,OTR/L Northwestern Clinic) gave us a beautiful presentation on exercising and stretching given neuromuscular conditions. She also covered Spasticity Challenges and options. With several members in the hospital or recovering from Baclofen pump, members discussed their personal pump problems and experiences. We also discussed a number of other items of interest including hand controls for the car.

We had 13 in our group for this meeting which was held in the Buffalo Grove Arboretum clubhouse. For lunch we ordered Jimmy Johns' subs and had the driver take the connection picture. For our fall meeting several from Wisconsin have expressed an interest in attending and are most welcome. By each of us sharing we have learned a lot. Our next meeting will be mid-October. If you are interested and want to share and learn more about HSP/PLS, please let us know at SPFIllinois@gmail.com

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Iowa Connection

On July 19th, the Iowans plus a family from Kansas met to share some salsa and guacamole. We got to meet two new Iowans, Troy Martens and Dave Statzini. We enjoyed each other's company. Thanks to all of you that came!

L to R: Troy Martens, Karen Powers, Kim Richman, Jackie Wellman, Mari White, Ray White, Alex White, Redondo White, Dave Statzini, Lois Wagoner, Barbara Lawrence

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LEMONADE FOR SPF

By Tina Croghan
Missouri State Ambassador, SPF Board Member

Perhaps the biggest help comes in the smallest package! On a warm Spring day, two little neighborhood girls set out to do a kind of right of passage—to open a tea and lemonade stand at the end of their driveway. But what they did next astounded even one of the girl's mother, the girls decided they wanted to raise money, "…to help Mrs. Croghan walk better!"

The two second-graders, Sophie Mabrey and Avery Parker (pictured) made their own sign and sold tea, lemonade and Girl Scout Cookies. They went to a few houses to "drum" up business and quickly sold out.

Later that afternoon, Sophie and Avery presented me with an unexpected surprise. The girls handed me a zip-lock baggie with crumpled bills and sweaty change, and proudly announced that they wanted to give me money for SPF!

I opened the baggie and counted the money. The two little girls had raised $22.05. Now you might be thinking, "That isn't much", but let me just point out—These are second-graders! Here are two little girls that have a simple concept—they wanted to help someone else.

Fundraising isn't about big money and relegated only to adults. All little kids everywhere should get involved. These girls would like to challenge other young children to do the same—have a lemonade stand or bake sale to raise a few dollars and to donate the money to SPF.

The contents of that baggie were priceless! The girls had given me hope, "…to help (me) walk better."

HOW TO USE EXERCISE BANDS AT HOME

By Harvey Mover

Lack of mobility seems to be one of the most difficult obstacles for people with HSP, and in the last nine months my personal goal was to overcome and beat this problem. I was fortunate to be invited to a seminar in Indianapolis in 2013 and spend time with the keynote speaker, Donnie Thompson.

In the power lifting world he is referred to as Mr. 3,000 for having been the first to lift a combined 3,000 pounds in bench press, squat and deadlift. Although this is an incredible milestone, the more interesting item is that he did this at 46 years old and had herniated disks and other injuries.

He told us about this and then shared with us how he overcame these hurdles using techniques for rehab, stretching and strengthening. He said ankles are the biggest asset to an athlete and, not kept in check, they can cause a complete breakdown of running, lifting or jumping for anyone. He talked about the four flexion points which, simply stated, are forward/backward and side to side. He suggested that we visualize how a duck walked and how their feet move.

For me, I noticed the biggest issue was pointing up and some side to side. He pulled me out of the audience since I was having the most issues and demonstrated for the audience how to do these techniques that I am using daily which has resulted in a noticeable improvement.

First he wrapped my ankle with a latex rubber band that is seven feet long. This compressed the joint. He then showed the group the four flexion points and provided resistance with his hands as he asked me to work the foot in the opposite direction.

As you can see from the picture, he then set up the exercise using two additional resistance bands with me holding one and him holding the other in opposite directions as he sat facing me with my leg extended. He used one band and I used the other and we pulled back and forth.

Although a little difficult the first time, I could feel my range of motion increasing over the next few minutes as we did the exercise. After we completed the exercise, I left the latex bands on for a total of 10 minutes and then removed them. The ankle area was very red with blood and the overall joint swelling had been reduced. Additionally I noticed a lot of tingling in the joint.

Since I have had some knee problems, I also wrap both the ankle and knee and do the ankle exercises and then remove the latex bands. Once the blood returns to the joints, it tingles and feels better. I was surprised how well this worked and in the last several months, I notice especially when I miss a day or two, how this negatively impacts my walking.

More detailed examples of how to can be found at the following websites by searching the following at YouTube.com:

- SuperDtv Flexion Your Ankles
- SuperDtv REHAB The Ankle, Fix that crappy ankle

The above should be something that many people with "mobility issues" can do and get some immediate improvements. Cost is minimal for the equipment needed and you can purchase online for under $50 including getting a set of three different resistance bands.

Once you have the resistance bands, you can do some other basic exercises with them to help increase muscle strength depending on how mobile you are. Leg extensions and abductors would be the simplest place to start.

For band leg extension, sit in a chair where your feet do not touch the ground. Take a band and put your foot in the opening and then run the rest of the band under and up the back of the chair and over your shoulder and then hold the other end of the band.

Extending your leg and allow it to slowly return after a pause. Start with a lighter resistance band and do what you feel comfortable with. Don't push yourself too hard on these first ones. The key is to be doing it right and feeling comfortable doing the exercise. Once you get stronger, increase the number that you do which you should try to target 12-20 but start slow and build up to 20 if you have to.

For band hip abductor sit on a bench and wrap your thighs together with the bands. Pull your thighs away from each or apart by stretching the band and then allow them to slowly return after a pause. Keep your feet solidly in place throughout.

These are a few key basic exercises that you can do to improve your mobility and avoid muscle atrophy. Once you master these, then you can either increase the number you do, add other exercises, or get bands with more resistance all of which you can do at your home.

Yes, these take a little work for some and more for others but you will reap the rewards. Stay positive you will see a difference!

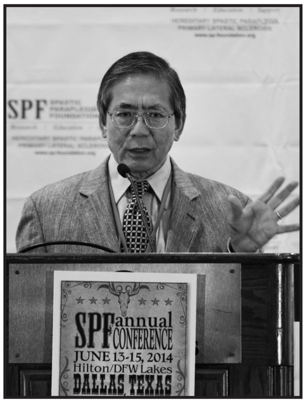
Dear SPF Family,

I would like to thank every sponsor, vendor, attendee and speaker who helped in making the 2014 SPF Annual Conference such a success.

There were so many who contributed time, money and so much effort in bringing the conference to a fun and absolutely informative event! We were so fortunate to have some of the most knowledgeable doctors on both HSP and PLS attend and speak at the conference. Scottish Rite Hospital opened its doors to give some of our young people special attention and time in regards to HSP. They also invited our Board of Directors to have their Annual Meeting at the hospital with food, tour and the use of their boardroom for the meeting.

A sincere thanks goes out to Craig Gentner for once again being our Master of Ceremonies. It is not an easy job to keep everything on time and running smoothly. Actually, I now know that it's almost impossible!

Thank you to all the wonderful doctors who took time away from their busy schedules to come to Dallas and share their knowledge with all of us in attendance. Dr. John Fink, Dr. Nazem Atassi, Dr. Hiroshi Mitsumoto, Dr. Mauricio Delgado and Dr. Jonathan Rios and Corey Braastad, PhD.

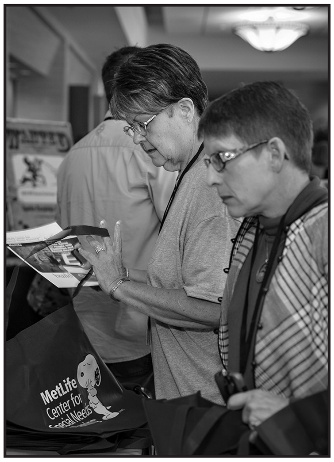


Our thanks to Jean Chambers (board member) who shared with us her wonderful canine companion, Annie, actually so much more than a canine…a bonified new member to the SPF Family. Those who were in attendance had the privilege of being able to love, spoil and meet Annie!

Thanks to Angela Dixon for being able to hear and visualize the concept for "Rounding up a Cure." Her artwork went above and beyond. Angela and James not only provided the artwork but donated their time and talent to provide the programs for the conference.

Thanks to Cory Weiser with MetLife who was a first time speaker to the SPF Conference this year. Thanks for MetLife in becoming a new sponsor for the SPF organization.

Rebecca Hart, we thank you for sending the most touching and wonderful video for the attendees to watch and enjoy. Rebecca actually gets the award for trying so incredibly hard to attend the conference, spending the most time at the airport and still not being able to fly to Dallas due to weather conditions.



Thanks to all the sponsors this year. Without this group of sponsors there would have been no wonderful fun times and great meals, and the "best of the best" speakers. So a special thanks goes out to: Kris Brocchini, Ned and Dee Davis, The Texas Rangers, Pete and Donna O'Brien, MetLife, Boggus Auto Group, Mike and Ann McLelland, Pittman&Davis, Texas American Insurers, Athena Diagnostics, GNS Foods, Visit Grapevine, Angela and James Dixon, The Christopher and Dana Reeves Foundation and Frank and Claudia Davis.

Thanks for the vendors who participated and did such a great job: Easy Access Travel, Snapit!, Life Vantage and MetLife. It would be great if you have time to let them know that you appreciate their time and participation.

There is a saying that "it takes a village" and this is especially true when putting on a conference. I could not have ever done this job without the help of my family, friends and the volunteers from the SPF community. I would like to thank Linda Gentner, who had to put up with an amazing number of emails from me asking way too many questions.

Thanks to all who helped make the 2014 Conference such a wonderful experience for me. I can honestly say I had a great time meeting everyone and being the conference coordinator. Thanks to everyone who made up the "village." I look forward to our 2015 SPF Annual Conference in Seattle!

Last but not least, I am sorry it has taken so long for the video to go on the website. It should be available in another week. It is truly a learning experience doing this job. Hopefully, next year I will have more of "my ducks in a row." Thanks for your patience.

Sincerely,

Claudia Davis

2014 SPF Conference Coordinator
claudiadavis6@yahoo.com

Rounding Up a Cure - SPF Annual Conference

June 13 – 15, 2014

Friday, June 13th
Reception and Dinner

"10 Basic Financial Steps for Special Needs Caregivers"

Cory Weiser, CIMA®, CMFC®, CRPC®, MBA
Financial Services Representative, Special Needs Planner, Investment Advisor, Representative for Bridge Financial Group, an office of MetLife.

Cory Weiser presented 10 steps to help caregivers get started in preparing for the financial future of their dependent with special needs. Each circumstance is unique, so caregivers should consider their own situation carefully.

1. **Plan for future medical, educational and housing needs for your dependent.** Start by thinking about what your dependent's needs will be in the future – and develop your financial strategy based on these projections.

2. **Review beneficiary designations.** To continue to receive federal aid, dependents with special needs cannot have any assets in their name (cash, art, jewelry) that is worth more than $2,000. It's important you, your friends and family leave inheritance and insurance benefits to your dependent's special needs trust (see number eight on this list for information on trusts).

3. **Have a family meeting to discuss your dependent's future needs.** Meet with your family members to discuss their concerns and options for future care. This is also a good time to broach the beneficiary designation issue discussed above.

4. **Speak with a special needs financial professional and create a team of professionals to assist you in planning.** It's important to pull together a support team that can help guide you through the variety of options available to you and your family.

5. **Contact local nonprofit organizations for additional resource support.** Your local nonprofit may be able to provide resources that can help with planning or that supplement the standard services provided by government agencies.

6. **Apply for government benefits.** Government benefits – such as Medicaid and Social Security (www.ssa.gov) – may help provide for your dependent's needs in the form of medical treatments and supplies, equipment, financial assistance and more.

7. **Prepare your Last Will and Testament (review and update periodically).** A Will declares how you want your estate to be distributed and allows you to select a guardian for your dependent when you pass away.

8. **Consider setting up a special needs trust.** This allows caregivers a way to provide for their dependent's care and quality of life, without disqualifying them for federal assistance. The money in the trust must be used to enhance the dependent with special needs quality of life, and can help to supplement standard services and benefits provided by government agencies.

9. **Apply for guardianship and conservatorship, if applicable.** Caregivers must apply for a guardianship or conservatorship to maintain legal control over financial and healthcare decisions once a dependent reaches the age of 18.

10. **Prepare a Letter of Intent.** Although not legally binding, this document is important for providing direction for the person or persons who will care for your dependent with special needs and should be stored with other vital documents, such as your Will.

* * *

"SPF Funded Research: Great Donations Are Driving Great Science"

Corey Braastad, PhD, Senior Scientific Director at Athena Diagnostics, SPF Board Member since 2008.

Dr. Braastad reported that SPF would be awarding about $800,000 of research funding in 2014. Dozens of grant applications have been received and are being reviewed by SPF's Scientific Advisory Board at this time. Awards are granted in each of the HSP and PLS research fields or for the more general field of Upper Motor Neuron diseases (HSP and PLS are both UMN diseases). He also told the group that, in collaboration with the NEALS, the first SPF Virginia Freer-Sweeney Clinical Research Training Fellowship in PLS was awarded to Christina Fournier, MD, of Emory University in Atlanta, GA.

Dr. Braastad further discussed the major basic research and clinical research areas of focus and the principal researchers in each area:

Primary Lateral Sclerosis (PLS)

- Longitudinal characterization of disease (Fournier, Floeter)
- Biomarker discovery (Fournier, Mitsumoto, Siddique)
- Exome sequencing (Siddique)
- Outcome measures for use in clinical trials (Fournier, Mitsumoto, Floeter)

Hereditary Spastic Paraplegia (HSP)

- Outcome measures for use in clinical trials (Fink)
- Models (Li – iPS cells, Rolls – flies, Sherwood – flies)
- Functional analysis of known causal genes/ proteins
- Exome sequencing (Chen, Zuchner, several labs)
- Drug screening (Mackay-Sim)

He then presented some biology facts and SPF's funding of and/or participation in the research being done in those areas.

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Saturday, June 14th

"Clinical and Genetic Aspects of HSP in Children"

Mauricio R. Delgado, MD, Director of Neurology, Texas Scottish Rite Hospital for Children

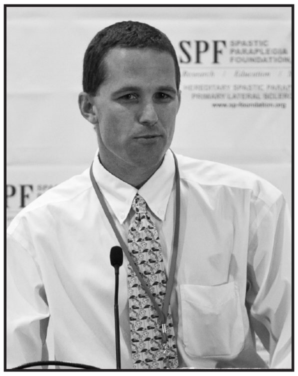
Jonathan Rios, PhD, Division of Molecular Genetics, Texas Scottish Rite Hospital for Children

Dr. Delgado began with background on the HSP Clinic formed at the Texas Scottish Rite Hospital for Children three years ago for studying childhood-onset HSP. One of the bigger problems they face is the lack of prior pediatric HSP studies. HSP, with all its variants, is a rare orphan disease, only 4–10 per 100,000 people. In the US, that means there are between 12,400 and 31,000 cases of HSP, the majority being adult-onset. Many other childhood conditions have initial symptoms that are nearly identical to HSP. This often leads to a frequent misdiagnosis as cerebral palsy which is 40 to 70 times more common than HSP.

Athena Diagnostics has a panel that tests for 11 of the 70 gene variants associated with HSP. That technology is being used to identify those in the Clinic's 120-child database that have the most common autosomal dominant forms of childhood HSP (AD-HSP) which are SPG3a (10% of AD-HSP cases), SPG4 (40-45% of pure AD-HSP) and SPG31 (4.5-6.5% of AD-SP). To date they have tested 83 patients using the Athena HSP panel and got positive results for 15 – that's only an 18% yield. Seven were SPG3a, 7 were SPG4 and 1 was SPG31. Of those 15, 10 were infantile onset (less than 2 years of age) and 7 of those 10 were SPG4 and 3 were SPG3a.

In addition to genetic classifications of HSPs, they also are classified by their mode of inheritance, Autosomal Dominant, Autosomal Recessive and X-linked. There is also a non-inherited mode known as Sporadic. The third classification of HSPs is their Clinical Presentation consisting of: clinical manifestations that are indicative of uncomplicated or pure forms of HSP, where neuro signs are limited to lower limbs, manifestations that are indicative of complicated forms of HSP, and age of onset: Early (separating infantile from childhood/ adolescent) and Late onset.

The HSP Clinic also performs an assessment of motor impairment. They use the ICF: International Classification of Functioning, Disability and Health as a conceptual framework to see the "full picture" of a patient's abilities, including the activities the patient can and cannot do (limitations), in what activities they can and cannot participate (restrictions) and what environmental and personal factors affect their abilities. The Clinic also developed a Gross Motor Function Classification of Pediatric HSP Patients and applied it to the 15 confirmed pediatric HSP patients in order to develop individualized treatments for each of them.



Dr. Rios took over to explain the difficulty in identifying the genetic connection the other 82% of the patients have with HSP. Genetic testing is limited today to 11 specific genes. Therefore, many genetic test panels still come back as "negative" because the patients' HSP genes are not one of the 11. Other results of these tests can be positive for one of the 11, a new variant of a known SPG gene or a new HSP gene.

Dr. Rios reminded the group why TSRHC is studying the genetics of HSP: most children with HSP treated at TSRHC have no family history of HSP and the success rate for genetic testing is very low (18%). Gene research will help get better and earlier genetic testing and diagnosis and faster access to treatments.

Much more is now possible with whole exome sequencing. Instead of 11 genes, researchers can now sequence over 20,000 genes. There are currently 14 clinical exome sequences that have been done and of those, 2 were confirmed as HSP, 4 as not HSP, 5 as negative, and 3 as unknown. The interpretation of these results is limited by our knowledge of the gene and its variants. New knowledge is gained through research and is not limited by prior knowledge.

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"Therapy Development for People with Upper Motor Neuron Diseases"

Nazeem Atassi, MD, MMSc, Assistant Director of the Neurological Clinical Research Institute at Massachusetts General Hospital.

Dr. Atassi began his presentation with some background information on NEALS, the Northeast ALS Consortium. Established in 1995, NEALS now has 400 members (physicians, neurologists, physical therapists, researchers, clinics) and 100 sites in the US. NEALS has a commitment to collaboration, sharing with any ALS researcher that needs them, rich data sets from previous NEALS trials and samples and tissue from NEALS' repository of more than 20,000 cryovials of plasma, serum, corticospinal fluid, urine, whole blood and extracted DNA. This is all in support of NEALS' main goal to "translate advances in ALS research into clinical trials for patients as quickly as possible."

After SPF members on the NEALS Upper Motor Neuron Committee began to question why all the organization's efforts were directed at ALS and none at PLS, also an UMN disease, they agreed to form an SPF-NEALS Collaboration. The goals of this collaborative effort are: (1) Improve HSP/PLS clinical care; (2) Connect HSP/PLS patients with experienced NEALS clinicians; (3) Launch biomarkers studies; (4) Use the NEALS infrastructure to plan and conduct HSP/PLS clinical trials; and (5) Encourage NEALS members to submit proposals to SPF's Research Grant Program.

To date, the collaboration has produced a list of NEALS centers that also treat PLS and HSP; a survey of patient needs; conduct regular webinars on HSP and PLS research; established the first Upper Motor Neuron (UMN) Registry with the objective of understanding the natural history of PLS; and established the 2-year SPF-NEALS sponsored Virginia Freer-Sweeney Fellowship in PLS, awarding its first such fellowship for 2013-2015 to Christina Fournier, MD, Emory University.

Tasks yet to complete are (1) Make the PLS diagnosis earlier by developing diagnostic tests and increasing neurologists' awareness of PLS symptoms; (2) Understand disease mechanisms – the genetics involved, from studying disease models (mouse, animal, stem cells); and from studying the disease mechanisms in people; (3) Understand how the disease progresses using the information gathered from the natural history studies; (4) Measure response to treatments using biomarkers and clinical outcomes; and (5) Design and conduct clinical trials that address the symptoms of PLS and also those that address modifying the disease.

Next steps are to increase the use of imaging with dedicated PLS studies with yearly PET-MRI scans; develop more PLS biomarkers, both fluid and genetic, to ultimately improve clinical care and accelerate drug development; build on the existing pilot UMND Registry by increasing the number of sites and patients whose natural histories are recorded there; conduct more clinical trials, especially those that determine the effect of exercise on the PLS; and train additional young investigators dedicated to UMND by continuing the SPF-NEALS Fellowship in PLS. All of these require funding for PLS/HSP research from organizations like SPF.

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“Progress in Understanding Spastic Paraplegia”

John K. Fink, MD, Neurogenetic Disorders Program, University of Michigan, Medal Advisor to the Spastic Paraplegia Foundation.

Dr. John Fink, Director of the Neurogenetic Disorder Program at the University of Michigan and medical advisor to the SPF Foundation spoke to the conference about the significant progress being made in research for both HSP and PLS. He called the past year “a landmark year” in research and new discoveries related to those two conditions.

According to Dr. Fink, there are now more than seventy known gene mutations that are part of the HSP condition, and he indicated that eighteen or twenty of those had been discovered within the last six months. He also stated that researchers are finding more gene mutations that affect PLS as well. There is also an advance in stem cell models, particularly for drug testing purposes.

Dr. Fink spoke at length concerning the process for completing clinical trials. First, researchers must understand the molecular processes involved with any testing circumstance. Then, various clinical assessments must occur. Researchers like to sever compounds in cellular cultures, and then test in animals before conducting clinical trials. He stated that testing two drugs in mice will cost as much as $100,000.00. That is the reason it is so important to raise funds to assist with the ongoing cost of research.

Many attendees were interested in and questioned Dr. Fink regarding clinical trials for ampyra, a multiple sclerosis drug, that some are finding provide positive results in regard to improved gait issues. He indicated he believed we are getting closer to initiating those clinical trials for HSP and PLS purposes.

Summarized by Greg Pruitt, SPF Board Member

* * *

“My Story, Having HSP and How Riding Has Made a Difference in My Life”

Rebecca Hart, Para Equestrian – Dressage

Due to weather related flight delays, Rebecca was unable to attend the conference. In lieu of her presentation, attendees were shown a 6 ½ minute video, narrated by Rebecca, about her disease and the Para Equestrian Dressage competition in the 2016 Para Olympics. The video is available for viewing on the SPF website, www.sp-foundation.org, by selecting “2014 Annual Conference Recap” on the home page. The following article appeared online the day after the annual conference concluded.

Para-Equestrian Sets Sights on Gold in 2014

Para-equestrian champion Rebecca Hart proves that having a disability doesn’t derail your dreams.

UNIONVILLE, Pa., June 17, 2014 /PRNewswire/ -- Born with a very rare genetic condition called hereditary spastic paraplegia (HSP) that keeps her from walking across a room unaided, six-time U.S. National Champion para-equestrian rider Rebecca Hart has set her sights on winning Gold at the 2014 AllTech FEI World Equestrian Games (WEG) in Normandy in August.

After winning her sixth national championship on June 6, placing fourth and just behind the Bronze at the Para-Olympics in 2008 and 2012, Hart is determined to make it to the podium this time around. Hart recently returned from Denmark where she purchased what she believes is just the right mare for the job, Romani, from the prestigious Straight Horse Farm.

“What is special about this horse is she combines the athleticism and skill of a high- performance dressage horse with a very giving and adaptable intelligence and temperament,” says Hart. “It took a long time to find her, but with a horse like Romani, gold is definitely in reach.”

Riders like Hart, whose legs are less responsive and nimble than more able-bodied riders, require an animal with just the right mix of attitude and performance. If the horse is too responsive, then Hart will have trouble controlling the animal. If the horse is too slow, then Hart will not be able to complete the complex and precision steps Dressage demands.

This requires para-equestrian riders to become far more proficient than their able-bodied counterparts.

Backed by sponsors Starbucks, Margaret Duprey of Cherry Knoll Farm, William and Sandy Kimmel, and many friends and family, Hart is also riding in support of the Spastic Paraplegia Foundation and its efforts to find a cure for HSP and its ultra-rare cousin, Primary Lateral Sclerosis (PLS). Both HSP and PLS are related to other serious central nervous systems disorders like ALS and Huntington's disease.

This year's WEG is special because it is the only international competition where para-athletes compete on the same stage as their able-bodied counterparts. Usually, para-equestrian competitions are held in the same venues but on separate dates thus lowering the profile of the riders.

"To be able to be there, we can show we are high-powered riders and equally qualified to compete on any stage in the world," says Hart. "Yes, we have disabilities, but our abilities parallel those of able-bodied competitors. Having our events going off at the same time, being side by side, we can effectively demonstrate this."

Rio or Bust

While the WEGs are perhaps more prestigious than the Olympics, Hart has her sights set squarely on winning Gold in Rio in 2016. If she is successful, she will significantly raise the profile of American para-equestrian riders everywhere and give the sport the big boost it needs to survive and flourish in the U.S.

"That is our ultimate goal," says Hart. "I've been close to the podium a couple of times, and it would be as much their win as it would be my own. It would be amazing to stand on the podium and see our flag go up."

Hart rides out of Blue Hill Farms in Unionville, PA and, like all top-level athletes, trains at least six days a week -- in between fund raising events and a full time job as a financial analyst as well as another part time job at Starbucks. She will be heading to West Palm Beach, FL in March to compete in the Certified Para-Equestrian Dressage International (CPEDI) qualifier and then on to Europe for two more qualifiers in Belgium and France.

About United States Para Equestrian Association
The United States Para-Equestrian Association (USPEA) includes every equestrian discipline that is practiced by athletes with physical disabilities. Each rider or driver competes under the eighth discipline, The Para-Equestrian discipline, of the United States Equestrian Federation. The USPEA is a network of riders, judges, national federation board members, and equestrian enthusiasts. The association gives athletes the ability to get involved and expand their knowledge and experience in the sport of Para-Equestrian.

Article written by Allen Bernard
Media Advisor
Media Contact for Rebecca Hart and the SPF:

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"My Canine Companion Journey"

Jean Chambers, RN, SPF Board Secretary

Dogs are supposed to be man's best friend, so it is understandable that a support dog is the natural choice for a person with limited abilities. Annie is the three-year-old, yellow lab support dog for Jean Chambers, the Board Secretary for the Spastic Paraplegia Foundation. Jean Chambers led an afternoon session on support dogs at the SPF Annual Conference in Dallas this June. "Annie does so much more than just retrieve," said Chambers.

A support dog, when wearing their working vest, is all seriousness and no play. If someone comes up and asks to pet Annie, Jean commands Annie to "sit" first before indulging in all the friendly pats. According to Chambers, Annie was a "love sponge" the weekend of the conference. Jean also had many people waiting in line and asking if Annie needed a walk. When Annie isn't working, she lies obediently at Jean's feet waiting to respond to her voice and needs.

Annie is from Canine Companions of Santa Rosa, CA. After Annie completed a required 24 months of obedience and advanced training, Chambers attended a two-week bonding/training period in San Jose where Annie was matched with her. "The dogs really do the choosing," said Jean. "I had my eye on another dog in the litter, but Annie and I ended up being a perfect fit."

Together they visited several public places where Annie displayed the ultimate in control amid a myriad of distractions. Jean Chambers dropped her leash to Annie. Annie responded by staying by Chamber's side.

Many people wanted to know if that strap across her nose was a muzzle. "I use a 'gentle leader.' She is free to open her mouth, lick, eat, but it is just a way to keep her from pulling me, since my balance isn't the greatest."

When asked what Annie does for her, Jean Chambers responded that there are several enrichments that she can accomplish like picking up dropped articles including her own leash and opening/closing doors. But it's the intangibles that Chambers never realized that Annie encourages. "Annie makes me get up and let her out three or four times a day. She needs walking then, not just letting out." Chambers went on to say, "If I go anywhere, I must allow an extra 10-15 minutes for Annie to socialize. I never meet a stranger. I have a built-in conversation ice-breaker."

When asked if Annie cost anything to acquire, Jean Chambers responded, "No. I had to fill out a lengthy five stage application which included the submission of several pictures of my home and all of my mobility devices. Then an interview followed. After that, my name was put on 'the list.' I waited another 14 months after being approved when I finally got the call from Canine Companions. I drove down to Santa Rosa, CA, where I stayed at their facility. I was paired up with several different dogs, but none had that instant 'click' like Annie and I. After 'graduation,' Annie and I came home to Vancouver, Canada. She's been by my side ever since, cheering on the Canucks!"

Canine Companions has 5 centers in the US – two in California, and one in Florida, New York and Columbus, Ohio. In 2015 they will be opening a new center in Dallas that will serve people from Texas.

Summarized by Tina Croghan, Missouri SPF State Ambassador, SPF Board Member

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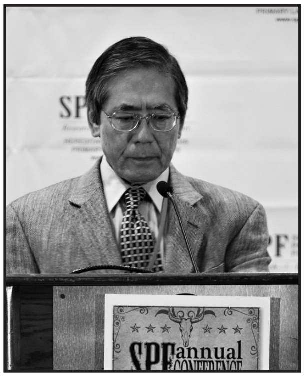
"PLS Cohort Study of Multicenter Oxidative Stress (ALS/PLS COSMOS) - Early Results of PLS COSMOS Projects"

Hiroshi Mitsumoto, MD, Eleanor and Lou Gehrig MDA/ALS Research Center Medial Director



PLS is considered to be a "pure" Upper Motor Neuron Dysfunction/ Disease (UMND) and it is exceedingly rare, only 1% to 3% as many patients as there are for ALS. It is pure in the sense that it is only a UMND whereas ALS is both a UMN and a LMN disease. Life expectancy is probably not affected by PLS but functional impairment is great. It is a much neglected and obscure disease with very few studies performed in the past. PLS' rarity makes it difficult to develop and execute studies so most have used patient histories.

Patients considered for the COSMOS study are more than 20 years old, able to understand the study, have had PLS for more than 5 years and less than 15, have had a normal electromyogram (EMG) within the past 12 months, have no other active or unstable diseases, and have no other neurological disease to explain UMND.

The initial study group totaled 40 patients, the goal was 50. However, SPF publicized the fact that additional PLS patients for the study were desired and COSMOS received calls from several potential candidates to add to the group. Dr. Mitsumoto then presented the results of biomarker measures and clinical features for 355 ALS patients and 40 PLS patients and commented on the comparative differences and similarities of the two groups. [These results and his comments may be viewed on the SPF website, www.sp-foundation.org, by watching the video of Dr. Mitsumoto's presentation at the conference. Ed.] Analyses yet to be performed are Paraoxinase 1 (PON1) to determine if oxidative stress affects PLS, occupational exposure, lifestyle and hobbies, dietary factors and psychological factors.

In summary, this is the first time PLS has been comprehensively designed and studied in a multi-centered environment. The study found genetically known molecular mutations in about 20% of patients with clinically definite PLS. A preliminary cluster analysis of clinical features showed two distinct phenotypic diseases in the PLS category. Biomarker studies indicate that PLS resembles ALS. However it is still unclear if the rest of the PLS group, after excluding known mutations, consists of one disease. Undoubtedly, Dr. Mitsumoto concluded, “We need to study a larger number of patients with clinically definite PLS.”

Breakout Sessions

Q&A Session with John Fink, MD.

During the afternoon question and answer session, Dr. Fink made a number of important points concerning the importance of stretching and strengthening exercises:

- Exercise may help reorganize/retrain your leg and other muscles. Upper motor neuron issues cause gait issues, and the repetition of improper walking technique can retrain those muscles the wrong way. So, exercise and stretching often is part of attempting to retrain proper technique.
- Swimming pool walking, when possible, is an excellent exercise. Concentrate on good heel strikes.
- Stationary bicycles and ellipticals can be very beneficial exercise tools, but go “backwards” sometimes in order to work different muscles important to proper gait.
- When possible, stand every hour for at least a minute or two, longer if possible.
- Have an exercise partner who will motivate and keep you focused and committed to exercising frequently.
- Frequency of exercise is more important than the duration of one session. Better to have shorter sessions more often than longer sessions, less frequently.

Summarized by Greg Pruitt, SPF Board Member

GREAT NEWS!

We just received a generous donation of $10,000 from a company called Invitae. Invitae is in the same business as another large donor to SPF, Athena Corp. Both companies do genetic testing for the different known HSP types. Please see the Invitae web site at <u>www.Invitae.com</u> where you will see that they run the full panel HSP test for $1500.00. Their toll free number is: 800-436-3037.

Q&A Session for Parents of Children with HSP

Linsley Smith, RN, Texas Scottish Rite Hospital for Children

The breakout session for parents of kids with HSP was really a fantastic group of parents/grandparents. The room was full. It was led by the team from Scottish Rite Hospital and it was a very informal question and answer period. It was a great big support group. I did not take many notes because of the great conversations. The research they are doing and how to get involved was discussed. They have the capacity to do genetic testing, blood tests, gait analysis, oxygen consumption and energy consumption studies, and clinical neurological visits. It can be pure research which is then covered for free but a parent does not get the results. If you go as a clinical patient then they will give all the results. Many times insurance will pay for things when you go as a clinical patient but not always. Once you get accepted they would like to monitor the children at yearly intervals to observe progression. They are currently enrolling kids. You need to contact the Clinical Nurse and send in all medical records for them to review and they will contact you to set up the appointments. The hospital clinic is multidisciplinary in that many different specialists test the kids. The hospital is really fantastic and is completely geared towards kids.

Notes provided by Mari White, Kansas SPF State Ambassador

Sunday, June 15th

TeamWalk and Tour of AT&T Stadium

On Sunday morning 45 conference attendees departed from the hotel to participate in the Team*W*alk and Tour at AT&T Stadium in Arlington, home of the Dallas Cowboys. At the end of the tour, the group gathered on the field for photographs.

Up Close and Personal

"An Ounce of Prevention…"

By Don Wilson
SPF North Carolina Ambassador

I can't claim to be any sort of expert, however, Bettie Jo has been in a power chair for about 12 years and about 7 to 8 years she has been in it 24/7, including sleeping. She is not blessed with abundant glutes, so she has little natural cushioning. We are very proactive in protection against problems and during that time, Bettie Jo has not experienced a pressure sore (ulcer) or even a "hot spot."

Pressure ulcers are much easier to prevent than to cure. Dolores has listed the people that are involved in the first steps to take in seating systems and nutrition. I want to add three other keys to prevention: clean, dry, and monitoring. Funny thing, these are the same requirements I learned to keep my feet blister free in the army and passed on to my Boy Scouts on those long hikes.

Both skin and clothing must be clean. It does not take much dirt at a pressure point to provide enough friction to initiate skin breakdown.

Keeping the area dry is just as important as keeping it clean. If working with bed sores, a light application of powder produced for that purpose to the sheets may help reduce friction. Check with a medical supply store for the best powder, not baby or talcum powder on local store shelves. When our kids were in diapers, we used Aquaphor to keep their bottoms dry and protect against diaper rash. Hospitals use a cream usually with a high percentage of zinc oxide, up to 100%. I have found that generic CVS 40% zinc oxide cream works best. If applied to the areas in most danger of developing a pressure sore (ulcer) it protects against moisture and from dirt particles.

The most important activity is monitoring to insure that a possible problem is caught and solved before it becomes a problem. That means continually checking the areas that may be susceptible to pressure sores (ulcers). That does not mean just the areas that may in constant contact with materials, as the bottom is in constant contact with the cushion of a wheel chair. Also check where constant contact and friction may happen. For an example, spasticity has drawn Bettie Jo's feet toward each other, normally with heals in constant contact. Keep those areas in mind when checking for possible problems. Look for any reddened area on the skin that, when pressed, is "non-blanch-able," meaning that when pressed with a finger, it does not turn white. This is the first stage announcing that skin ulceration will follow if not treated. In cases where skin is darker, discoloration of the skin, warmth, swelling, or hardness may also indicate a problem. This would be considered Stage One in a Four Stage evaluation, and the only stage that may not require professional care.

First aid for Stage One: 1) Relieve the pressure to the involved area, using pillows, special foam cushions and sheepskin. 2) Keep the area clean and dry. 3) Work on improving nutrition and any other underlying problems.

If the ulcer is at Stage Two or worse, the health care provider should provide specific instructions and special dressings and antibiotics if necessary.

DO NOT: 1) Massage the area of the ulcer as that can cause tissue damage under the skin. 2) Do not use do-nut shaped or ring shaped cushions because they can interfere with blood flow to the at risk area and cause additional complications.

I will share a happening. My cousin, a retired nurse, was observing me giving Bettie Jo a shower, with the thought that in an emergency, she could help out. I finished the shower including washing her hair and then toweled her dry. I laid the towel over Bettie Jo and gathered a handful of toilet paper and began to get down on my knees *(knees don't hurt when I am crawling, but it is a bitch to get down and then back up)*. My cousin asked "What in the world are you doing?" I replied that I was going to check Bettie Jo's "running gear." I used the tissue to dry her bottom and insured that all of the skin "blanched" correctly. I do this as part of each shower.

As I said, prevention is much easier than curing.

…A Pound of Cure

John Staehle
SPF Ambassador, North Texas, SPF Board Member

As I write this, I have been home for only a week. Prior to that I had spent a total of 126 days (18 weeks) away from home. During those 126 days, I had four surgeries at two different hospitals, spent 60 days at a recovery hospital followed by 58 days at a skilled nursing facility for continued recovery and rehabilitation. All because I allowed a pressure ulcer to get so bad that it required surgical repair.

Before I get into the mistakes I made, let me tell you a little about my HSP journey. Though I had symptoms of an UMN disorder for many years, I tended to just wrote them off as part of growing older. I had never heard of HSP. My first gait problems were identified when I was 50 years old and in 1999, after numerous tests, I was diagnosed by a neurologist to have HSP. No one in our family who was still alive at the time had, or knew of other family members that had, any symptoms similar to those of HSP, so I suspect I have either the recessive or the sporadic form. My HSP progressed steadily and within 9 years I was permanently off my feet. That's when my experience with pressure ulcers first began.

After I discovered I could no longer support myself on my legs, I began using a manual chair at home and a 3-wheel scooter that I hauled to and from work on the back of an SUV. I was on the scooter all day – at my desk, in meetings, at lunch - and switched to the chair when I got home. The seat on my scooter was not intended for all day use and soon I developed several sores on my bottom. These were my first encounters with pressure ulcers. Mistake number one was to ignore the sores when they first appeared. After they began to bother me so much I couldn't find a comfortable sitting position, I finally got out-patient treatment from wound care specialists at a local hospital. However, it took three months of twice weekly treatments for those sores to heal. My second mistake was to not reposition myself on my chair every 30 minutes or so to relieve pressure on the areas previously affected by sores. Over the next couple of years, I experienced multiple recurrences of sores in the same general areas where I previously had pressure sores. Each occurrence was treated "after the fact."

In November of last year, I developed a small sore on my left side beneath my ischium bone (the part of the pelvis that protrudes down on both sides). I started wound care treatments in December at the hospital, going twice each week. But this sore was persistent and refused to improve even though I continued to receive wound care. On my wound therapist's advice I began to break up my day with a 2-hour period out of my chair and off my butt - still no improvement. Continued debridement just widened and deepened the sore. On several occasions, the wound therapist suggested I pursue a more aggressive treatment – specifically surgical repair. I acknowledged his suggestions but put them off each time because there was so much I had to do first. That was my third mistake – putting deferrable tasks ahead of my health. At the end of March, I finally agreed to consult with a general surgeon about surgical repair of the sore. Pre-op tests showed that I had a fever and a high white blood cell count. Upon seeing that, the doctor ordered me to immediately go to the emergency department and get admitted to the hospital.

After 2 surgeries by a plastic surgeon to clean out the wound and a week's stay in the hospital (during which I experienced some bizarre dreams and hallucinations from a drug reaction), I was transferred to a recovery hospital. Four weeks into my 8½ week stay there, I had a third surgery to cover the nearly healed wound with a "flap." This flap was not a skin graft as I originally thought. It was a 12" long piece of skin, fat tissue and muscle from the back of my thigh that was stretched to cover the wound area. Lots of stitches and staples held it in place for 6 weeks during which I had to lay on my back in a special bed to not stress the sutures. Complications required a fourth surgery 11 days into the original 6-week recovery period which reset the 6-week clock. Once those six weeks passed and the sutures and staples were removed, I began the arduous process of restoring the muscle mass, tone and flexibility I lost being in a hospital bed for more than three months. I've ended my stays away from home, but will continue to have restorative therapy for several additional months and take preventive measures so I never again have this experience.

The lessons I learned from this experience are:

- Do not ignore even the smallest irritation in areas subject to the development of pressure sores (ulcers).
- Seek professional treatment from a wound care therapist as soon as a sore appears.
- My health is more important than any deferrable task I have to do.
- Reposition, reposition, reposition – at least three times each hour.
- "An ounce of prevention is worth a pound of cure." *Benjamin Franklin*

Take care of yourself, treat those areas susceptible to the development of sores and if a sore develops, get immediate professional treatment. Do not assume you can deal with it yourself.

Pretty Shoes

By Tina Croghan
Missouri State Ambassador, SPF Board Member

Twelve years ago I hadn't heard of HSP. I stumbled, but hey, my mom always called me, "Clumsy." I tripped often and walked funny. But I figured I was just a klutz and I grew into, "the crazy drama teacher," so my eccentric movements were expected and accepted.

Later, I remember walking on the beach with my husband. I had taken him to Gulf Shores, Alabama, to celebrate his 40th Birthday. Earlier that morning, I sat on the deck enjoying the ocean breeze looking out at our footprints in the sand. Horrified and embarrassed, I was certain that other people had noticed. My husband's footprints were these perfect indentations in the sand and mine were elongated trenches! I made a mental note to "pick up my feet" like my mom always told me.

The next day I tried. I really did! Once again at breakfast, I scanned the beach for my footprints. There they were! Just slightly better than the previous day. It still looked like I was dragging my toes. But at that time I could run, jump, swim, wear pretty shoes…*(heavy sigh)* Guys, for a woman this is a MAJOR deal!

Twelve years have passed since that day. Now I only drive on streets around my house. I never drive at night or on the highway. I can no longer just "run in" any place for something. First of all, I look for places that have a drive-up window. People are usually very willing to help, I just have to ask.

It wasn't always like that. I just walked really "funny." I looked like I was drunk! When I was at a restaurant, I wouldn't get up to go to the bathroom. I was afraid other people would think that I had too much to drink.

You see--that was *MY* problem. I was afraid of what others thought of me. I knew what I thought of the person in the motorized cart at the grocery store-- "All she needs to do is get up and walk!"

Without a clear diagnosis, I have bouts of major denial. I've let the demons convince me that, "it's all in my head." But hard as I try, I can't make myself run, jump, swim or wear heels!

I had to finally come to the realization that I had only so much "gas in my tank" and how did I want to spend it? If I went grocery shopping, I could only push the cart halfway through the store. If I conceded to use a motorized cart, I could make it all around the store and even backtrack for an item I suddenly remembered.

It still took me a while to use a scooter in front of my friends. Why?! Others are immediately relieved of "duty" and all can enjoy the outing. I can even help out by carrying heavy or awkward packages and hey--I even come with my own seat!

There's still that nagging self-doubt. It would be so much easier *(I think)* if I just knew--if I had a face or enemy to fight and subsequently defeat!

I guess that my whole point is that no matter how positive I am some days, there are many days of self-doubt and "woe is me!" That's OK, too. That's what makes me human.

I do everything I can while I still can. I act like I'm running on my recumbent bike. I hold on to my walker with both hands as I try to make my feet come off the ground in a jump. I do water therapy twice a week exercising my legs and walking. The other day I wore ballet flats to a friend's wedding. They weren't heels, but they weren't black or white leg braces either!

You Can Make Money for SPF by Just Lifting (and Pressing) Your Finger

HSPers and PLSers....along with your friends and families.

The Online Shopping season is approaching. To donate to the Spastic Paraplegia Foundation without costing you a penny please use goodshop when you shop online. In fact, whenever you have that online shopping urge, go to **goodshop.com**. Enter Spastic Paraplegia Foundation as your charity of choice and then enter in the blank your favorite store…Amazon, Kohl's, Target, Barnes and Noble, Land's End, etc…hundreds of stores. If you enter the store's website this way, the retailer will donate from 1% - 6% (and sometimes more) of your purchases to SPF. Donating without costing you a penny more and requiring only about 5 seconds of extra time…how can you beat that? Please do this and pass this information on to everyone you know.

Also if you ever search the Internet (who doesn't?), use **goodsearch.com** and a penny will be donated to SPF for every search. Goodsearch is powered by YAHOO!. Just think, if everyone reading this, including their friends and families, used **goodsearch.com** and **goodshop.com**, SPF would have enough money for more research grants or for more clinical trials.

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