Synapse

Volume 16, Issue 1 Spring, 2013

Newsletter of the Spastic Paraplegia Foundation

Letter from the President

Dear Friends,

Spring is my favorite time of the year and this year, along with the budding of new leaves and flowers comes the budding of new hope for the cure for HSP and PLS. What's more, the research we are funding is more tangible than basic science. It involves real hope that we can all sink our teeth into.

We are just now announcing our funding of $370,000 for three grants for HSP and PLS research along with an exciting PLS Research Training Fellowship. The $200,000 Virginia Freer-Sweeney Clinical Research Training Fellowship offers two years of very significant salary and educational support to train exceptional junior clinical researchers to translate advances in neuroscience into treatments for people with PLS.

Dr. Christina Fournier at Emory University, Atlanta Ga. has been selected for this support. She has trained under two of the most respected ALS/PLS Clinician/Scientists in the world, Drs. James Russell and Jonathan Glass. She plans to continue to work collaboratively with Dr. Glass and develop a longitudinal PLS biomarker study.

Hiroshi Mitsumoto, MD, DSc is continuing his SPF funded groundbreaking study with PLS but the next phase of this study is even more appealing than the first. In this new phase, he will be analyzing patient samples for biomarkers and genetic signatures unique to PLS.

In HSP research, we funded two teams. Both are working on actual treatments. Tina H. Lee Ph.D. with Carnegie Mellon Univ., Pittsburgh Pa., will work to identify compounds to treat SPG3A associated HSP. Andrew Grierson, Ph.D., Kurt De Vos, Ph.D. Univ of Shefield, UK and Ludo VanDen Bosch, Ph.D. Univ of Leuven, Belgium are all focusing their work on a new therapeutic approach to treat HSP.

Dr. John Fink, our SPF medical advisor, put it succinctly at our annual conference last June when he said "there is a lot to be excited about!" and with these projects now in the works his proclamation is proving to be true.

I so much want to again thank everyone who has played a part in supporting our research and invite everyone to join with us in celebrating this exhilarating progress.

Sincerely,

Frank Davis

President, SPF

MARK YOUR CALENDARS!

The next SPF Annual Conference is scheduled for June 14 & 15, 2013 at the Renaissance St. Louis Airport Hotel in St. Louis, MO. For those that attended in 2009, it's the same hotel!

Registration opens January 7, 2013 so book early and we'll see you there.

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Volume 16, Issue 1 - Spring 2013

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
Allen Bernard.....Senior Editor

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2013 Annual Conference - Join Us in St. Louis

Dear Friends,

I am excited to be hosting the Spastic Paraplegia Foundations 2013 Annual Conference in St. Louis. We are putting together an exciting conference for everyone, starting with our Friday night keynote speaker, Rebecca Hart, a five-time USEF National Para-Olympic Equestrian Champion and two-time Paralympian in Equestrian Dressage (Beijing 2008 and London 2012). Rebecca will be speaking on how her HSP doesn't hold her back.

On Saturday, Dr. John Fink, Dr. M. Delgado, Dr. James Berry and Dr. James McNew will be speaking on a host of interesting topics from current research updates to children with HSP. We will also have numerous, informative breakout sessions hosted by SPF board members and other special guests.

The conference will be held at the Renaissance St. Louis Airport Hotel. For those of you who were there this is the same hotel as the 2009 conference. It is convenient to the airport and downtown. If you are going to be flying, the hotel offers a free shuttle. If you are driving, the hotel is offering SPF attendees free gated parking.

On Sunday (Father's Day), we plan to have a special event coupled with a Team Walk. If you'd like to explore the Gateway to the West while you here St. Louis has a lot to offer you. Checkout these websites: ExploreStLouis.com and StLouis.About.com.

Sincerely,

Tina Croghan
2013 AC Conference Chair

REGISTRATION FORM

SPASTIC PARAPLEGIA FOUNDATION - 2013 ANNUAL CONFERENCE

SPF SPASTIC PARAPLEGIA FOUNDATION, INC.

Research / Education / Support

June 2013 - St Louis, Missouri

Renaissance St. Louis Airport Hotel
1-888-340-2594
9801 Natural Bridge Road St. Louis, Missouri 63134 USA

Our group code: spfspfa

Friday, June 14th
Rebecca Hart – 5 time USEF National Para-Olympic Equestrian Champion and two-time Para-Olympian (Beijing 2008 and London 2012) in Equestrian Dressage.

Saturday, June 15th
Speakers include:
Dr. John Fink - University of MI,
Dr. James McNew - Rice University Biosciences Research--Houston, TX
Dr. James Berry - NEALS
Dr. Mauricio Delgado - Scottish Rite Hosp. & Univ. of Texas Southwestern Medical Center, Dallas, TX

Sunday, June 16th
Tour Busch Stadium home of the MLB World Series 2011 winning St. Louis Cardinals while participating in the St. Louis SPF Team Walk
(transportation provided!)

Friday Night Dinner - $45
($50 after May 17th)
Children - $35
Saturday Conference - $65
Sunday Team Walk & Tour - $20 per person

http://www.marriott.com/hotels/travel/stlsa?groupCode=spfspfa&app=resvlink&fromDate=6/13/13&toDate=6/17/13
www.Renaissance St. Louis Airport.com

Name #1:_____ Diorder: HSP_____ PLS _____SP_____
Name #2: _____ Name #3:_____
Children: _____ Age(s): _____ Name(s):_____
Address: _____
Phone: ( )_____ E-mail: _____ Text?: Y/N
I will be using a: Cane _____ Walker _____ Scooter_____ Wheelchair _____

Friday Night Dinner: QTY:_____@ $45.00* pp = $_____
Conference: QTY:_____@ $65.00* pp = $_____
Children: QTY:_____@ $35.00* pp = $_____
Team Walk & Tour: QTY:_____@ $20.00* pp = $_____
Special Meal Requirement:
_____Regular _____ Vegetarian _____Soft _____Gluten-free
SPF Donation: $_____
Total Enclosed: $_____
**Early registration price before May 18, 2013*

Please send your registration form to:

Spastic Paraplegia Foundation
c/o Linda Gentner
1605 Goularte Place
Fremont, CA 94539

SPF to receive $24,000 from ExxonMobile

As a result of the 2013 campaign ExxonMobile Employees' Favorite Charities Campaign (EFCC) The Spastic Paraplegia Foundation receive approximately $24,000. Over the past 10 years, the EFCC has generated over $300,000 for SPF. These donations were made possible by the dedication and hard work of former board member and long-time Exxon employee Annette Lockwood.

Annette is seen here accepting a check from ExxonMobile.

Volunteer Opportunities - State Ambassador

Being a state ambassador is so very important to help people when they are new to HSP or PLS. They are often scared and feel alone. Remember how you felt when you were told you have PLS or HSP?

If you'd like to help new people overcome their fears and meet the challenges of HSP or PLS head on, please consider becoming a state ambassador. Go to the SPF website and read the job description.

We ask a lot but if that seems a bit more than you feel comfortable with, we can change it to fit your comfort zone. We need people to send welcome e-mails so people know they are not alone. Even if there is an ambassador in your state, we can always use more and especially in the larger states. Plus you don't need to live in a state to be willing to help folks.

Please go to the website to check out this opportunity to make a difference: http://sp-foundation.org/get-assistance/state-ambassadors

Paralympic Champion Rebecca Hart Keynoting Friday Night

Rebecca was born in 1984 with HSP in Erie, Pennsylvania. She first got involved with the Paralympic movement in 1998 at a regional competition in Atlanta, GA. This competition served as the selection trials for the 2000 Sydney Paralympic Games. And it was there that Rebecca decided she wanted to aim for international competition herself.

Rebecca purchased her first horse and began training seriously for national and international competition. Over the years, Rebecca has represented the United States at several international events, including the 2003 and 2007 Para-Equestrian World Championships.

In 2006, Rebecca was the National Champion at the USEF National Para-Equestrian Championships. With her current competition horse, Norteassa, Rebecca won the 2008 National Para-Equestrian Championships. Rebecca represented the U.S. at the 2008 Paralympics in Hong Kong where she went on to place fourth individually.

Rebecca also had a strong 2009 season once again claiming the National Para-Equestrian Championship.

In 2010, Rebecca not only defended her national champion title but also claimed reserve champion honors, as well. Rebecca won first prize with the Dutch Warmblood gelding Kazan and reserve honors with her 2008 and 2009 National Champion and 2008 riding Norteassa.

When not riding Hart enjoys kayaking, sailing, camping and rock climbing. She is currently a student at Penn State University and is majoring in international business and accounting. Rebecca is looking to become a forensic accountant upon graduation.

Paralympic Champion Rebecca Hart will be speaking Friday night about her experiences with HSP and how it has not stopped her from pursuing the things she loves. Please join us for Rebecca's fascinating story of hope and courage.

My Life with HSP

By Mitzi Probst – 8th Grade

The first time I wore AFO's (ankle-foot orthotic) to school was a day I will never forget. When I first left the doctors' office they felt like horseshoes dragging me down. How was I going to fit it at school with these things? My mom said I could take the day off, but I thought it would be worse if I waited. The monster would just get bigger.

When we got to school, I realized that I had gym in less than one hour. Panic! I figured out that hiding or panicking would not make it better, so I just plugged along. The surprise was that my gym teacher met me in the hall and let me skip the whole locker room thing. I brought sweatpants after that until I have enough guts to wear shorts.

My life changed drastically after that. At first most people had no idea I had a physical disability. I did not want to tell anyone about my HSP and tried to keep it a secret. My AFO's did not allow me to keep it a secret anymore. Eventually, I realized that hiding it only made my problem worse. It was not like I was hiding the secret identity of a superhero, it just confused people.

People could tell that something was wrong and they often just filled in the blank with other odd or demeaning conclusions. By disclosing my secret, it allowed other people to see me as I am and not draw incorrect conclusions about me or my abilities.

Being a middle-schooler with HSP has been no easy task. Socially, the trick has been to find people who enjoy me and do not care that I cannot run real fast or that my handwriting is on par with that of a second grader. I am a total geek *(hence the superhero joke earlier.)* and often hang out with people who share my same interests.

HSP has not stopped any of my dreams. I still love writing and history. Typing has helped and I can type twice as long and fast as I write. I am very outgoing and often do volunteer work. I participate in student council and National Junior Honor Society. I still swim competitively and have begun to swim with an adaptive program. I found a place for myself on my school swim team by concentrating on distance swimming when I could no longer compete against sprinters.

HSP has actually helped me discover talents and abilities I did not know I had. For example, I needed to switch out of band to choir. Turns out, I got a lead in our last concert because of singing abilities that I did not know I had. I also learned that there is plenty of room for all kinds of dancers in community theater. I have sung and danced in several plays now; including my school's spring musical.

I don't have to be perfect, just willing to do things differently sometimes. I just have to remember to save my energy and that I often need more sleep than the average teenager. However, there are days where I get frustrated with everything and break down inside. I would tell you that the key is never to give up, but I am not one for sappy advice. Sometimes I have to let emotions scream in my ear before I can get back to thinking logically. Because I know that wallowing in self pity doesn't get me anywhere, I try to soon get over it and move on.

I have found that if I focus on what motives me I can get through each of these challenges. For me, managing HSP is all about finding little "work-arounds" that make my day easier. I use a laptop and I occasionally take the day off from gym. Most of the time I have teachers who are great and follow my accommodation plan, but occasionally I have a teacher who doesn't understand.

I think everyone has had a teacher they don't connect with, but that is different from one who assumes that my physical limitations equate directly to cognitive limitations. Like everything else, this has actually served to motivate me academically. I have become extremely focused and goal-oriented. I also work very hard to make myself stand out from my non-disabled peers. By doing so, those teachers have gotten to know me and most of the time things have worked out well.

Throughout history there have been many people with disabilities who have been very successful, including Franklyn D. Roosevelt, John F. Kennedy and Professor X (X-Men). Why not me? That is where I am going to place my focus and determination.

All in the Family

by Robin Grossbier

In July of 1989, I was the first of my siblings to be diagnosed with HSP. I grew up watching the disease steal the strength and self-esteem of my father. The picture of my father, Alex Grossbier, crying in his wheelchair knowing that his youngest daughter had been diagnosed with HSP makes my heart cry. What I hope he knows while he is enjoying the wonders of heaven, is that he has given me and my other siblings who also have HSP, the courage and inspiration to walk (or roll) this role on earth. Dad never gave up, he always met the challenges with all he could offer.

Each day gets a bit more difficult, but I know that God's vision for my life is not for pain, but for the good of His kingdom. My pain has been used so many times to minister to others in pain. Joni Eareckson Tada said it best, "God looks at my HSP through 2 lenses. The narrow lens, he sees the tragedy, hurt and weeps with me. The wide angle lens, He sees my disability in relation to everything flowing from it. He sees me in His mosaic stretching to eternity--this mosaic brings him much delight."

I don't say all this to make everyone think that all days are bright and filled with joy. Many are filled with pain, frustration, and difficulty. My choice in my life is to offer a sacrifice of praise through what I can do, and give thanks for each breath, each card I can make, each phone call I use to cheer others, and finally each prayer for those who are also hurting in their lives.

Physically I am in a wheelchair for 90% of the time. I try to use a walker in my home to maintain any mobility I currently have. I also have bulging discs and back pain most of the time. My newest development is pain and muscle stiffness in my hands. I am not sure if it is HSP or just cold weather etc. I have a great friend who serves as my caregiver as needed. Having been single all my life, I am so grateful for her sense of selflessness as I ask Laura to get this, do that, and help me with… if not for my sister in Christ, I would find it difficult to be on my own. Thank you for sharing in my journey, Robin Grossbier.

It's All Greek to Me

If you've ever wondered why finding a cure can be so hard this paragraph should help. This is from *The EMBO Journal* and published in January. The title of the abstract is:

Structural basis for conformational switching and GTP loading of the large G protein atlastin.

The research, outlined in the following abstract, was conducted by researchers at Cornell University:

"Atlastin, a member of the dynamin superfamily, is known to catalyse homotypic membrane fusion in the smooth endoplasmic reticulum (ER). Recent studies of atlastin have elucidated key features about its structure and function; however, several mechanistic details, including the catalytic mechanism and GTP hydrolysis-driven conformational changes, are yet to be determined. Here, we present the crystal structures of atlastin-1 bound to GDP•AlF_4 – and GppNHp, uncovering an intramolecular arginine finger that stimulates GTP hydrolysis when correctly oriented through rearrangements within the G domain. Utilizing Förster Resonance Energy Transfer, we describe nucleotide binding and hydrolysis-driven conformational changes in atlastin and their sequence. Furthermore, we discovered a nucleotide exchange mechanism that is intrinsic to atlastin's N-terminal domains. Our results indicate that the cytoplasmic domain of atlastin acts as a tether and homotypic interactions are timed by GTP binding and hydrolysis. Perturbation of these mechanisms may be implicated in a group of atlastin-associated hereditary neurodegenerative diseases."

"To laugh often and much; To win the respect of intelligent people and the affection of children; To earn the appreciation of honest critics and endure the betrayal of false friends; To appreciate beauty, to find the best in others; To leave the world a bit better, whether by a healthy child, a garden patch, or a redeemed social condition; To know even one life has breathed easier because you have lived. This is to have succeeded." — This quote is often and erroneously attributed to Ralph Waldo Emerson. But whoever said it doesn't really matter. It's all true.

My Story about a Grandmother, Mother, Brother, and Child with HSP

by CandCsMom

My daughter who just turned four on January 1st was diagnosed with HSP back in January 2012. Just a little after her third birthday. From the time she was able to pull herself up to stand as an infant, she was up on her toes. (I think she'd make ballerinas jealous.) She was a very late walker. She would walk holding onto things a little while after she figured out how to pull herself up, but she didn't let go of things and walk until she was 18 months old.

I was hoping that her walking on her toes would be something she'd simply grow out of. My father told me that I too walked on my toes as a child and grew out of it. I took my child to several doctors. All of them told me she was okay and she would grow out of it.

It took the recommendation of one doctor to get her seen by a muscle specialist at the Children's Hospital here in Colorado at the North Campus. From there they referred me to the Muscle Clinic at the Children's Hospital main Campus in Colorado. That was where she was diagnosed.

At the age of four, my daughter is still a toe walker. She stands on her toes, even when she's not walking, but she can stand flat footed. It just takes some work to do it and is not too comfortable for her. That is her normal. It's normal for me to stand flat footed, while it's normal for her to be up on her toes.

She freaks out and cries when people other than myself mess with her feet. She cries when I cut her toe nails. When first diagnosed she went to physical therapy once a week and was taking Baclofen. She currently goes to physical therapy twice a month through February this year. Starting in March she will be going only once a month. She currently wears braces which helps her walk heel-to-toe.

When not in her braces she is up on her toes, stumbles, and drags her feet. If you let her go outside in just her socks and she runs on the sidewalk, within a few minutes there will be holes on the top of her socks.

Everything with my daughter is nothing new to me. (Well, the braces and Baclofen are.) My grandmother and mother had HSP. I say had because they are no longer with us. From as far back as I can remember as a child my mother would always walk on her toes. I thought it was because she had a job that required dress shoes and she wore high heels, so I thought maybe she was just used to walking in her heels even when she wasn't in them.

My mother didn't really show any other signs of HSP until a little after turning 50. Before she passed away, she was walking with a cane and sometimes a walker. More so a walker right before she passed. She had been diagnosed back in 2011. She did not die because of HSP. She passed away because of a blood clot.

My brother is 34 and also has HSP. As far back as I could remember as a child my brother has always stood with his knees bent, dragged his feet while walking, walked on his tip toes. Unlike my daughter who stands on her toes while standing, my brother stands flat footed with his knees bent. My mother stood flat footed as well.

My brother as a child was diagnosed with cerebral palsy. This was back in the late 70's early 80's where people were pretty unaware of HSP. My brother still hasn't officially diagnosed with HSP, but after my mother's diagnosis her doctor said that it was more than likely this is what he has, as well. He currently doesn't have any insurance and the state he lives in doesn't consider him to be disabled. As he is getting older his condition is getting worse. He walks more pigeon toed, has more pain in his feet and back, especially after standing or sitting for long periods of time.

So there are four generations of HSP that I know of in my family. My grandmother (my moms mother), my mother, my brother, and my daughter. When my daughter was first diagnosed I kicked myself for giving this to her. I know she didn't ask for it, and neither did any of my other family members. I just know first hand how hard it is for kids with disabilities in school.

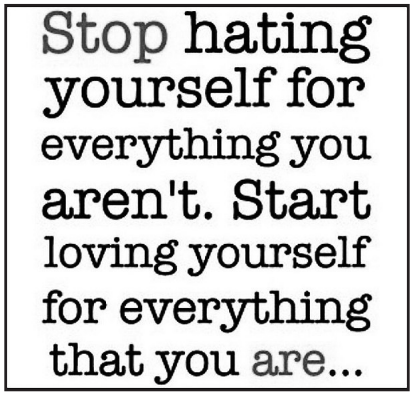
My brother and I are only three years apart, so throughout our school years, we were mainly in the same schools. It broke my heart seeing the way people treated my brother and they way they talked about him. My heart is going to break even more when it comes to my daughter. My brother is one thing to feel sad about, but when it comes to your own child, it's on a completely different level.

There is no way for me to know if later on in my life symptoms of HSP will show up or not. My mother's symptoms really didn't show up until a year or so after she turned 50. My mother and I are so much alike when it comes to kids. We both had two kids, one that has HSP and one that for the time being is showing no signs of HSP. Both of our kids that have HSP were first born. My brother is first born and my daughter is first born. We each had one boy and one girl. She had a boy and then girl and I had a girl and then boy. My mother had my brother at 27 and had me at 30. I had my daughter at 27 and my son at 29, four months shy of turning 30.

Not sure if this is going to help anyone, but that's my story about HSP. I don't mention really anything about my grandmother because I honestly didn't know she had HSP or any signs of HSP until after my mother passed away in 2009. My father said something about it after my daughter was diagnosed.

It Is What it Is

by Jackie Wellman



When my grandmother died on January 1, 2000 she was bitter and intensely unhappy. She was mad at the world for giving her HSP and some other bad luck but I learned a lot from her. I loved her and I wished I could have alleviated her misery.

I learned the only thing being angry and negative does is make those around you miserable. It certainly does not make the HSP mysteriously vanish. Unfortunately, we are stuck with this genetic mishap until one of the researchers comes through for us. I am counting on that being sooner rather than later. Why would you not? We have nothing if we do not have hope.

There are several things that I believe are critical in dealing with our neurological disasters. Number one is to keep moving. Now I mean that several ways. First, exercise as much as you can. It is so important to keep what strength we have. Dr. Fink described the muscle to me once as a mosaic. HSP or PLS weakens small parts and the key is to strengthen the other parts. Stretching also makes life with HSP much easier. Stretching before bed will lessen night spasms.

Another way to interpret the keep moving statement is that we need to just "do" as much as possible. We can do almost as much as people with healthy neurons; just maybe slower or with a cane or walker. Maybe even a chair.

Exercise your brain also. Many of the drugs to relive spasms make us a little foggy. Read books, do puzzles, play scrabble, etc. It really helps.

We all have our days of self-pity. I think we would be really abnormal if having a slowly degenerating faulty neurons never bothered us. The hardest thing is just as you get used to some limitation, then another comes along. The key is to make those pity parties few and far between. Being social helps with that. Join the e-mail support group on the SPF page or the SPF's open or closed *(if you would rather just share with a few folks instead of the whole world)* Facebook pages.

I cannot get out as much as I would like so these contacts with other living beings are very important to me. Because of Facebook, I've had lunch and dinner with so many friends that I have not seen since high school, which was 30 years ago. It is easy to keep in contact with others in this day and age with texting and e-mail, so take advantage of that. We all need to be active participants in life. See as many friends as you can.

Another way of keeping positive that helps me is to learn as much about HSP and treatment for my particular symptoms as I can. Knowledge is power. Seek out the best doctors in your area. Ask questions. Read. Get involved with research studies. The ways of joining research studies are listed on the SPF website (www.sp-foundation.org). Research studies bring us closer to a cure.

Get out of yourself and do something for others. There a hundreds of ways to volunteer. Doing something for someone else can take the focus off your problems. Volunteering will feel incredibly good. Like you always hear, giving is better for the giver than the receiver.

Giving does not have to cost money either. You could go to a local school and read books to children or help them with their math. Write letters to the troops. You can give blood. You can volunteer at a political campaign. Good Search volunteering. Do some fundraising for SPF.

The bottom line is keep as active as you can. Keep as positive as you can. It will get better.

Researchers turn one form of neuron into another in the brain

by BD Colen

A new finding by Harvard stem cell biologists turns one of the basics of neurobiology on its head by demonstrating that it is possible to turn one type of already differentiated neuron into another within the brain.

The discovery by Paola Arlotta and Caroline Rouaux "tells you that maybe the brain is not as immutable as we always thought, because at least during an early window of time one can reprogram the identity of one neuronal class into another," said Arlotta, an associate professor in Harvard's Department of Stem Cell and Regenerative Biology (SCRB).

The principle of direct lineage reprogramming of differentiated cells within the body was first proven by SCRB co-chair and Harvard Stem Cell Institute (HSCI) co-director Doug Melton and colleagues five years ago, when they reprogrammed exocrine pancreatic cells directly into insulin-producing beta cells.

Arlotta and Rouaux now have proven that neurons too can change. The work is being published online today by the journal Nature Cell Biology.

Their experiments targeted callosal projection neurons, which connect the two hemispheres of the brain, and turned them into neurons similar tocorticospinal motor neurons, one of two populations of neurons destroyed in Amyotrophic lateral sclerosis (ALS), also known as Lou Gehrig's disease. To achieve such reprogramming of neuronal identity, the researchers used a transcription factor called Fezf2, which long has been known for playing a central role in the development of corticospinal neurons in the embryo.

What makes the findings even more significant is that the work was done in the brains of living mice rather than in collections of cells in laboratory dishes. The mice were young, so researchers still do not know if neuronal reprogramming will be possible in older laboratory animals, or in humans. If that proves possible, the implications for the treatment of neurodegenerative diseases could be enormous.

"Neurodegenerative diseases typically affect a specific population of neurons, leaving many others untouched. For example, in ALS it is corticospinal motor neurons in the brain and motor neurons in the spinal cord, among the many neurons of the nervous system, that selectively die," Arlotta said. "What if one could take neurons that are spared in a given disease and turn them directly into the neurons that die off? In ALS, if you could generate even a small percentage of corticospinal motor neurons, it would likely be sufficient to recover basic functioning."

The experiments that led to the new finding began five years ago when, "We wondered: In nature, you never see a neuron change identity," she said. "Are we just not seeing it, or is this the reality? Can we take one type of neuron and turn it into another?"

The researchers analyzed "thousands and thousands of neurons, looking for many molecular markers, as well as new connectivity that would indicate that reprogramming was occurring," Arlotta said. "We could have had this two years ago, but while this was a conceptually very simple set of experiments, it was technically difficult. The work was meant to test important dogmas on the irreversible nature of neurons *in vivo*. We had to prove, without a shadow of a doubt, that this was happening."

The work in Arlotta's lab is focused on the cerebral cortex, but "it opens the door to reprogramming in other areas of the central nervous system," she said.

Arlotta, an HSCI principal faculty member, is now working with colleagueTakao Hensch of Harvard's Department of Molecular and Cellular Biologyto explicate the physiology of the reprogrammed neurons, and learn how they communicate within pre-existing neuronal networks.

"My hope is that this will facilitate work in a new field of neurobiology that explores the boundaries and power of neuronal reprogramming to re-engineer circuits relevant to disease," Arlotta said.

The work was financed by a seed grant from HSCI, and by support from the National Institutes of Health and the Spastic Paraplegia Foundation.

Please Consider SPF in Your Planned Giving

Planned Giving provides donors with the ability to support nonprofit associations and charities by arranging for gifts to be made through financial or estate planning. This practice enables donors to make charitable gifts now and establishes provisions for gifts that will continue after their lifetimes. Planned Giving also provides an avenue for supporters to make even larger donations than they could during their lifetimes. This approach provides financial and tax benefits to both donors and their heirs.

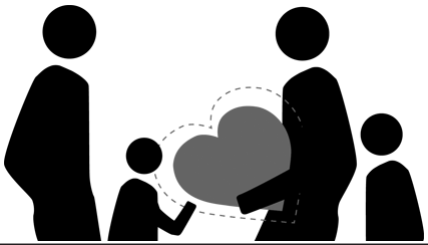
Our Endowment Fund

The Spastic Paraplegia Foundation benefits from Planned Giving through our endowment fund. This fund was established in 2013 with a gift of $25,000, and through additional and generous contributions over the years to come stands to reach to ever larger amounts. Only the interest income from the fund is used to support Foundation programs, while the principal remains untouched, growing ever larger as new donations are made.

There are a number of ways to contribute to the Foundation's endowment fund – bequests, gifts of stock, ownership of property, direct donations, and more. Each of these vehicles can offer substantial tax deductions to the donor, and each helps to guarantee the future of the Foundation's work.

Bequests

Bequests are made through a will or a trust, and represent the most common type of planned gift. Remembering the Spastic Paraplegia Foundation in your will is an important and personal way of providing hope to children and adults with Hereditary Spastic Paraplegia and Primary Lateral Sclerosis until the happy day when a cure will be announced. Bequests are fully deductible for estate tax purposes.



To make a bequest of cash or other property to the Foundation, your will *(or supplemental codicil, if you do not wish to write a new will)* should state: "I give and bequeath to the Spastic Paraplegia Foundation, a nonprofit corporation, organized under the laws of the State of Virginia, and having its office at 7700 Leesburg Pike, Falls Church, VA 22043 the sum of $____ or ____ percent of the rest, residue, and remainder of my estate to be used for general purposes of the Foundation."

It is strongly recommended that you seek advice from your personal tax or legal advisor when making a bequest. These professionals can provide important guidance in setting up a bequest that is best suited to your individual tax situation.

Other Planned Gifts

Planned gifts can take several different forms. Funds can be contributed from:

Trusts, such as charitable lead trusts and charitable remainder trusts

- Charitable gift annuities
- Life insurance policy assets
- Retirement plan assets
- Endowment funds

An Extraordinary Impact

Please know that your planned gift can make a significant difference in the lives of patients and families fighting the rare upper motor neurological disorders of HSP and PLS, and will aid in funding real life changing research toward a cure. The recent PLS $200,000 fellowship that allows a researcher to work full time on finding the causes and a treatment for PLS was the result of a very significant gift from Virginia Freer-Sweeney's estate.

If you would like more information about presenting a gift to the Foundation's endowment fund or making a bequest, please contact the Spastic Paraplegia Foundation at 1 (877) 773-4483.

SYNAPSE APPEAL

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

Reflections from Boston

by Allen Bernard

As you may have seen from the website my brother-in-law Jack Prior ran the Boston marathon on April 15 as part of the Genzyme running team. The team members each ran for a patient partner. In our case it was my daughter Brianna, who was diagnosed with SPG3A three years ago at age two.

It was such a wonderful day. The sun was shining. It was perfect running and watching weather: a rarity in New England during early Spring. Spirits were high and the excitement was palatable as 25,000 or so runners took to the streets.

On the Saturday night prior to the race, Genzyme hosted a dinner for the 17 runners their patient-partner and the families at the companies Allston facility just across the Charles River from downtown Boston. It was a great evening where we all got a chance to meet and share our stories and gain inspiration from one another.

To cheer on Jack and all the runners my wife, Karen, and I headed out Monday morning to the Genzyme team cheering area in Wellesley. Many of the folks from Saturday night were there and the sense of camaraderie was high. Everyone was feeling good and happy to be part of such a great event. In case you've never been around Boston for the marathon I can tell you it's a BIG deal. The state basically stops and about 500,000 people come out to watch and cheer along the 26 mile route.

For our part, this was a great day because Jack took it upon himself to run for Brianna. He trained hard and got the whole family involved and excited. Jack's energy, enthusiasm, and dedication were infectious lifting everyone's spirits and bringing the family together around Brianna and HSP.

Courtesy Genzyme

Of course, that excitement evaporated when news of the bombings came in. The whole family was at Jack's house in Newton, just a few miles from Copley Square and the finish line, eagerly awaiting news of his crossing when we first heard. I think I was heading the kitchen when I overheard the words "bombing."

We knew Jack was close to finishing so where was he? Was he okay? What was going on? Thankfully, Jack was about a mile away just passing through Kenmore Square when "about 15 cops with walkie-talkies pressed to their ears came out onto the course and stopped everyone," as Jack described it.

We were very relieved to hear this news but also very concerned about his condition and what kind of help was available to all the runners who had to suddenly pull up short with no medical treatment or blankets to keep them warm as their bodies cooled off or anyway to contact loved ones. These were tense moments for everyone that changed the day from one of triumph to one of sadness and reflection.

It's still hard to reconcile the feelings of being relieved for my family, sad for those who lost everything and disappointed for the loss of what was supposed to be a day of overcoming obstacles, of persevering, of hope … not coping with sadness and anger. Nothing was accomplished by this cowardly and shameful act. Nothing. There is no good that comes out of such actions. Only heartache for what could have been and for those that were killed and injured.

And yet this does not take away from what was accomplished by my brother-in-law and the other Genzyme team members. I have to remember that. It's hard but it is important to do so. Even though Jack didn't get to cross the finish line, he's definitely a winner in my book. Certainly, running a marathon is a most appropriate metahpor for what it is like to live with a rare disease. I guess you just have to decide to focus on the positive if you want to move forward regardless of the many obstacles this disease — or anyone else — puts in your way.

So it is this I will take with me. Not the disappointment, sadness and anger caused by two mindless shameful cowards.

Genzyme's Boston Marathon team raised $65,000 for rare diseases. Pictured are, **Front row:** Kristin Rapp, Kai Simon, Jen Tedstone, Jessi Colund. **Second row:** Colleen Dalton-Petillo, Andrew Scholte, Sharon Cotnam, Phil Maderia, Sean McShera **Third row:** Dan Wilkens, James Drob, Lisa Valaika, John Koltvedt, Kyle Kellinghaus. **Back row:** Shane James, Cian O'Brian, David Cunningham. **Not pictured:** Jack Prior

SPF SPASTIC PARAPLEGIA FOUNDATION, INC.
Research / Education / Support

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SPF and NEALS Announce Two Year, $200,000 Research Grant

SPF and the Northeast Amyotrophic Lateral Sclerosis Consortium (NEALS) announced the first Virginia Freer-Sweeney Clinical Research Training Fellowship in primary lateral sclerosis (PLS) in February. The fellowship highlights another successful collaboration between the NEALS Upper Motor Neuron Taskforce and the SPF.

The training fellowship offers two years of salary support of $90,000 per year plus $10,000 per year for educational activities. The objective of this fellowship is to train outstanding junior clinical researchers to rapidly and efficiently translate advances in neuroscience into treatments for people with PLS.

ALS, PLS, and HSP are very closely related from a clinical point of view because they share common outward symptoms, or phenotypes. Often doctors have to wait for symptoms to progress significantly before finalizing a diagnosis of ALS, for example. They also share a common mechanism of pathology that makes them very interesting to researchers since discoveries in one yield benefits and insights into the others.

There are currently no treatments for PLS or HSP outside of some mild symptom relief. But, because these diseases open a window into the inner workings of some of the most interesting and mysterious cells in nature, upper motor neurons, they are attracting researchers from around the world who want to understand not only UMNs but how all cells function.

These discoveries stand to aid clinicians in the treatment all diseases, not just those associated with the central nervous system (CNS). It is for this reason, the SPF is proud to be partnering with NEALS in this important endeavor.