

# Synapse

Volume 21, Issue 1

Winter 2018

*Newsletter of the Spastic Paraplegia Foundation*

## Letter from the President



Dear Friend,

I want to take this opportunity to say a **HUGE THANKS** to you for your generosity during our recent year-end fundraising event. We surpassed our year-end goal of \$150,000 by 69% and with the amount being matched by our anonymous donor, the total raised is over **\$403,000**. Congratulations!!!

Since being founded in 2002, the vision of our Foundation is the day when everyone with Hereditary Spastic Paraplegia and Primary Lateral Sclerosis will be diagnosed, treated and cured. Our mission is to support research that will reach that day as quickly as possible. Our very talented and caring Board of Directors work hard with our expert, internationally-renowned Scientific Advisory Board every year to know for sure that we are supporting the very best research and so not waste a penny of your donations.

You know, as your president, I try every day to do my job a little bit better. My former career as a businessman did not fully prepare me for this job. For one thing, I have never fully understood how to best communicate about giving to the people that are a part of this community.

I've been reading a lot about psychology lately and it is interesting that the latest research shows that your decision to give to SPF is much more than just a rational decision. People used to think of reason as Plato did when he described it as a charioteer controlling the horses (emotions) of his chariot to reach his destination.

Modern psychology is determining that the 18th century Scottish philosopher, David Hume, was right when he said that human reason is "the slave of the passions." Whether we are male or female, we make such decisions as whether or not to donate to such an incredibly good cause as The Spastic Paraplegia Foundation based primarily on our feelings.

We want to make the best decision possible, mind you, so we inform ourselves about all of the great progress that SPF is accomplishing toward that goal of curing HSP and PLS. This information was part of our last Annual Report and can be found on our website, [www.sp-foundation.org](http://www.sp-foundation.org). This information is learned and kept in the prefrontal cortex of our brains.

But there is more to the game than just information. Today, psychologists are coming to realize we have multiple "sub-selves" or modules in our brain, each competing for attention at the same time and it is our feelings that control which of the modules we are more prone to pay attention to. Thoughts are said to "think themselves" guided primarily by what feelings we are currently feeling.

Psychologists, Kendrick & Griskevics, divided the mind neatly into seven sub-selves with the following missions: self-protection, mate attraction, mate retention, affiliation (making and keeping friends), kin care, social status and disease avoidance. Each sub-self gives us important motivations to donate to SPF, but there are other factors in our lives trying to compete both for our attention and our dollars.

If you are feeling afraid for your financial future or are wanting to impress friends with a new car more than you are feeling charity toward your close family and friends with HSP or PLS, you will be more inclined to decide not to donate. So, your decision involves many parts of you, all playing a part toward reaching it. You are more like the chairman of the House of Representatives of your mind, listening to the votes from each subself or module rather than the president or CEO making the decisions.

So, I want to again thank you for your donations that make everything possible for The Spastic Paraplegia Foundation and I also want to especially thank you for the deep personal feelings or love that make those decisions possible

Sincerely,

*Frank Davis*

Frank Davis, President

# Synapse

**Volume 21, Issue 1 - Winter 2018**

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

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## Board Business

*Highlights of SPF Board of Directors meetings held between August and November 2017*

**August Annual Conference:** Tina reported the Conference Team was concentrating on the 2018 conference in Pittsburgh. Look-ahead plans are to have the 2019 conference in San Antonio and a location on the west coast in 2020.

The State Ambassadors Team reported there is now a Facebook page for all SA's, "SPF, Inc State Ambassadors" (a closed group).

**September: Kentucky Golf Tournament:** Greg Pruitt reported terrific weather, a good turnout and new sponsors led to a "Great day!" Participants quickly signed on and quickly sent in their checks. The tournament grossed about \$21,000 and after all the expenses are paid, we will net between \$17,000 and \$18,000 for SPF.

**November: Research Report:** Mark Weber reported the receipt of 15 grant proposals – 1 for PLS, 1 for combined and 13 for HSP. The list has been sent to Martha Nance, PhD, chair of the Scientific Advisory Board. We should have the SAB evaluations back by January. Mark also reported the SAB will stay as it was last year.

**Synapse:** John Staehle provided a written report to the Board. The Fall *Synapse* was printed and mailed to approximately 2000 recipients the end of September. It was also posted on the website. A call for Winter issue articles was posted on Facebook, the HSP ListServ and two PLS Yahoo Groups on November 3rd. The deadline for receiving material for the issue is December 1st.

## New Board Member – Jim Sheorn



Jim graduated with a BS in Business Marketing. He has over 20 years of experience in sales and over 15 years of experience with non-profits, including the American Heart Association, Spastic Paraplegia Foundation and Achilles International. Jim previously served on the SPF Board and was President in 2009. He lives in Brentwood, TN with his wife Melissa and two teenagers. Jim was diagnosed with HSP in 1994. Later it was confirmed with genetic testing that he has SPG4. His main focus will be trying to identify organizations and individuals that can help financially support HSP/PLS medical research.

# Fundraisers

## Golfing Fundraiser in KY 2017

By Norma and Greg Pruitt

Spastic Paraplegia Foundation in Kentucky raised over \$21,000 with a golfing fundraiser on September 9, 2017 at Drake Creek, to help fund scientific research for rare diseases HSP and PLS. Beautiful weather, and many thanks to fifteen golfing teams and more than fifty sponsors that raised funds in helping to find a cure and treatments. These two diseases, Hereditary Spastic Paraplegia and Primary Lateral Sclerosis, result in weakness and spasticity in the lower body and arms, slurred speech and difficulty swallowing.



Team SPF Kentucky



Tournament Graphic

We invite everyone to participate in next year's fundraiser on September 8, 2018. This fundraising event is planned and worked annually by SPF board member and his wife, Greg (HSP) and Norma Pruitt; SPF Kentucky State Ambassador, Kari Averill (HSP); Tim Vaughan (HSP); Mike and Joann (HSP) Faiht; and many family members and friends.

Anyone interested in learning more or matching donations, please contact us at [SPFKentucky@gmail.com](mailto:SPFKentucky@gmail.com) or visit [www.sp-foundation.org](http://www.sp-foundation.org). Follow us at #HSPandPLS or <https://www.facebook.com/SpasticParaplegia/>.



## Annual New York Fundraiser

By Ann Marie Lakin



Stevie B Shows Off His Acoustic Guitar Skills

On September 29, 2017 I held my annual NY fundraiser and, as last year, I dedicated it to my sister Helen who passed away very suddenly 2 years ago. Approximately 75 people attended the potluck event. We enjoyed a variety of delicious homemade food. Entertainment was provided by Stevie B on acoustic guitar and Nannyhagen Creek who sang a mix of bluegrass and other songs. Both entertainers had us singing and dancing along to some great music.

To help raise money I also had a silent auction. This year I was able to have 25 items to bid on. Local vendors were very generous - I'm always amazed at people's willingness to help. I had a lot of gift cards from restaurants as well as jewelry and other items donated by friends. Starbucks had donated a mug and a pound of coffee - people got into a real bidding war over that one.



Nannyhagen Creek Performs

At the door I asked for a \$10 suggested donation and with additional cash donations by attendees and the proceeds from the Silent Auction, that night I raised about \$4,000. Before the event I sent out invitations and I received about \$1,000 in donations from people

who couldn't attend. Then after the event someone made a very generous donation of \$5,000, bringing the total raised to about \$10,000. Since the meeting place, the Silent Auction items and the paper goods were all donated, the entire amount received will go to the Spastic Paraplegia Foundation.



The Silent Auction Table

My fundraiser this year was a HUGE success and I plan to do it again in 2018. I hope to see some new faces from the NY area or maybe you can do your own event. I was pleasantly surprised at how easy it was to do when I first started doing this 10 years ago.

# Living with HPS / PLS

## Get Rid of Those Sacks of Potatoes

By Christopher Falconer

Calf compression sleeves or socks is an option I highly recommend those with HSP consider to help alleviate the feeling of carrying a sack of potatoes on their legs. My wife, Jacqueline, bought me two pairs of the calf compression sleeves and socks which I just kept in my drawer.



After ignoring them for months and her insistence I try them, I gave in to her wishes. I had the most pleasant surprise sleeping in the compression sleeves and socks over the past month. The first night I wore them I woke up in amazement at how lighter my legs felt and how much easier it was to move my legs under the sheet and comforter. Also, my walking is much improved because I don't have that customary "sack of potatoes tied to my leg" experience where I struggle to lift my legs. I'm not sure exactly why they produced the physical benefits I have experienced, but these calf compression sleeves and socks are now a constant in my life – thank you Jacqueline. They also provide the added benefit of keeping my cold legs warm. Give them a try.

You can find these compression socks and sleeves at Bed Bath & Beyond or on Amazon. Amazon sells the compression socks based on your shoe size at prices between \$10 and \$35, and you are provided with customer review ratings. I love the Copperfit brand compression socks which sell for \$20. I also wear the Blitzu compression sleeves (size L-XL) which cost about \$10.00. These are available in a range of sizes from small to XXL. Amazon offers a useful guide to help you determine the size and fit that are best for you. Because of their benefits, I now wear compression socks day and night.

[Use AmazonSmile and select Spastic Paraplegia Foundation, Inc. to earn a no-cost-to-you donation to SPF with your purchase. Ed.]

## The HSP Bladder

By Jackie Wellman, SPF Board Member and SPF State Ambassador for Iowa

In my case it started with years of always having to go. I knew where every public bathroom in Des Moines was. I eventually tried many types of medication to control the bladder spasms and retention. Those meds helped for years but it got to the point where I needed a little more help.

I have had three family members with HSP die of bladder infections that were chronic and went systemic. The idea of an infection really bothered me. At one point I was taking a daily maintenance dose of an antibiotics for the bladder. Who wants that though?

The next step was to talk to the urologist about self-cathing. It sounded disgusting but if it prevented infections...? I learned how from the nurse. It is easy and painless. I did it every morning and night. The doctor told me to think of the bladder as a pond full of water that just sits there. If it never empties then some nasty stuff grows in it. HSP bladders do not empty. They just do not. Plus doing it before bed makes you get to stay in your warm bed all night! During the day things are much easier to deal with if you are starting with an empty bladder.

That controlled things for a while. After several years I decided to try bladder Botox. A catheter is used and the bladder is filled with Lidocaine. After about half an hour the doctor comes in and through a cystoscope she gives you about thirty shots in the bladder. Painless and in 10 minutes you get dressed and drive home. The first time lasted about nine months. Now I am doing it every four months. Your body builds up antibodies and it does not last as long. Some people get much longer effectiveness out of a treatment than I do. We are all so different. I do self-cath more using Botox than I used to. I have been doing this for years. Now I actually self-cath six times a day. It is just routine now. I would much rather do that than get an infection that kills me or wet my pants!

## The Chair That Gives You Freedom

By Kenny Hersh

My life has changed since my diagnosis with PLS some 15 years ago. As I reflect on it, some of the changes have been truly positive and the others range from annoying to more formidable. My inner drive and things that I enjoy have not changed all that much. I was a runner in my pre-neuro world. I enjoyed running a 5K and an occasional 10K three times a week, even during the brutal New England winters.

About a year ago, one of my kids saw an article in the Boston Globe that discussed the Freedom Chair and the small company that invented and manufactures it, GRIT (their website is [www.gogrit.us](http://www.gogrit.us)). I was quite intrigued by it and after some looking at their excellent website that has videos and details about the chair, I contacted them for an appointment to check it out myself and ask the million questions I always have. I live in Providence, RI and the GRIT office is in Boston, so it was an hour trip.



I usually use a walker to get around safely, but I found that I was missing out on walks with my wife, kids, grandkids, and friends. Going on a hike in the woods was no longer feasible. Walking around cities and strolling on bike paths were also difficult if not impossible activities to achieve until now.

The best way I can describe a Freedom Chair is that it is a hybrid of a wheel chair and a recumbent bike with hand controls. It uses a clever lever system that is powered by your arms. My first time in the chair was a bit like riding a new bike for the first time. There is a learning curve. However, once you practice a bit and watch videos that the GRIT team and users of the chair have posted, it is fun and easy to ride.

The GRIT team is remarkable. They are a very small group of dedicated people who truly believe in their mission to make the impossible possible for those of us who have some challenges. They are eager for feedback from users and have made many changes to the chair from suggestions by users. I have made some comments to them which they are always happy

to receive. One of the staff said to me that it is just the engineering thing to always try to improve the mousetrap. I have even read on the Freedom Chair Users Facebook page of modifications they did for one person who needed them due to his situation. That sense of caring and openness to suggestions is not common in today's world. With over 500 unit sold worldwide, they are truly making an impact.

We traveled to Toronto in July for a family wedding. While there, we met up with some friends that we have not seen in quite a few years. We wanted to "walk" around Toronto and luckily, I had brought my Freedom Chair with me. One of our friends happens to be an occupational therapist and she literally went crazy watching me get around Toronto in it. We walked around for five hours and I did get many positive comments and thumbs up from strangers as I was keeping pace with the group. There is not a chance I could have done that using my walker.

This past August, we did a family vacation on Cape Cod with our three kids, their 9 kids [a 10th grandchild was born in October!] and a few nieces and nephews. We went on a hike in the White Cedar Swamp walk in the Marconi Station National Park area. This was the hardest trail I have attempted and, except for some railroad tie steps that my sons and son-in-law gave me an assist, I was able to get through it without difficulty. We like to be "memory makers" for our kids and grandkids and being able to do this with them rather than sitting back at the house was fabulous. My grandkids argue as to which one will get to sit on my lap as I go on it. How great is that? On the same vacation, we spent many hours walking around Provincetown as well, which was a treat.



While not on vacation, I use it for exercise and am hoping to do the Providence Downtown 5K race that I used to run in my pre-PLS days. That race is in September, so I have time to train for it. I am able to walk with my wife and friends around the neighborhood and on the many bike paths in the area. Using it just keeps you in the game of life!

*Continued on next page*

The Freedom Chair does not come cheap. The cost is around \$3,000.00. However, if you are a veteran, it is approved by the VA at no cost to you. GRIT has no-interest financing options available. They often have promos that will reduce the cost a bit. It does come apart and can easily fit into the back of my Honda CRV. However, I find it a bit difficult to get it in and out of the car on my own and do need some assistance. I usually keep it assembled in the garage so I can use it easily around the neighborhood.

If this is something that is of interest to you, please go to the [gogrit.us](http://gogrit.us) website for more info and lots of pictures and videos. Feel free to contact me at: [kennethhersh@gmail.com](mailto:kennethhersh@gmail.com) if you would like to ask a question of me. Finally, please email me if you think you are going to purchase one as I can provide you a code for a discount.

Go for it...you will be so happy that you did!

## A Name You Need to Know

By Lori Renna-Linton

**Lily Rice**, a 13-year-old with **HSP**, is the **first** girl in Europe to land a wheelchair **backflip**.

So far, she's raised £7,161 (\$9,802) from a goal of £6,000 in order to go to the **WCMX** (*wheel chair motocross*) **World Championships** in California.



Quoted from Lily's GoFundMe page: "Skating helps me feel free and forget about my problems and I want to show others that you **can do anything you want to, you just have**

**to put your mind to it or do it differently.** Lily works out at skateboard parks to perfect her WCMX moves.

My motto is: **#puttingtheabilityintodisability**

You can either google Lily Rice or search for the following article written for the BBC in September: <http://www.bbc.com/news/uk-wales-41397992>

## TAKE AN EXTRA STEP!

By Tina Croghan, State Ambassador for Missouri

I have found—mostly by accident—that if I would slow down and not over-reach or multi-task, I would avoid a fall. "Just pause, take an extra step and simplify the task," says Ingrid Hurley of Hurley Pilates iStudio. She goes on to encourage, "Center your weight over your feet."

We all have discovered little shortcuts that save us time, but what I'm saying is try to avoid rushing—that's when accidents (i.e. falling) happen.

I have incorporated—Pause...take a deep breath...stand up straight...bring my shoulders down and elbows to my waist. Now—look at my feet. Make sure my weight is over both feet. I'm not trying to do something acrobatic on one foot. I'm not reaching or leaning. My hands aren't wet or trying to hold something!

You would think this adds time to my already rushed day - but NO! All of the above, "...just takes an extra second to assess the surroundings," says Ingrid. Pretty soon this became second nature to me. With a couple of nasty falls under my belt, I knew that I needed to change something before I hurt myself to a point where the injury dominated my already awkward movement.



I'm not  
**CLUMSY**

**It's just the floor hates me,  
the tables and chairs are bullies  
and the wall gets in my way.**

## My Radio-Controlled SPF Race Car

By Dan Cook

I am 53 years old and I was diagnosed in 2009 with HSP. I had always walked with a bit of a limp and remember stubbing my toes a lot growing up. But I played softball, basketball and bowled until I was almost 40 when things started to go a little different. My paternal grandfather had the gait that many with HSP get over time and he eventually was in a wheelchair. My mother, her twin sister and her brother also have the same problems that I eventually found out to be HSP. I talked my mother into seeing the same neurologist and she was finally diagnosed in 2014. They never had doctors that would tell them what they had and they never asked.

I got my wheelchair in 2009, but had always loved being outdoors and missed one of the things I really loved and that was working on race cars, particularly my cousin's car. I missed the comradery and family relationships the most, I guess. In 2012 I saw my first radio-controlled (r/c) race car and decided I had to have one. It offered so much of what I missed about auto racing. So, on and off since then, I have run dirt oval r/c cars. The bodies of the r/c cars have increasingly mimicked real race cars with every passing year. Other racers' cars had sponsor decals on them so, on one of mine, I decided I would sport the Spastic Paraplegia Foundation logo and contact information on my car, not as a sponsor, but to draw awareness to SPF and Hereditary Spastic Paraplegia. I ran this car around central and upstate New York in a

class called MudBoss. It was a bit of a conversation piece. When people asked about it, I made sure they also knew about SPF and HSP.

I don't get to compete in many things these days and the competitiveness of r/c racing has helped to fill that part of me that playing sports filled when I was younger. Working on the car, always trying to find the combination of adjustments that make it perform just a little better, is what really draws me to it. I also tend to be a bit anxious and working on the car or building a new car is one of the most relaxing things I can think of.

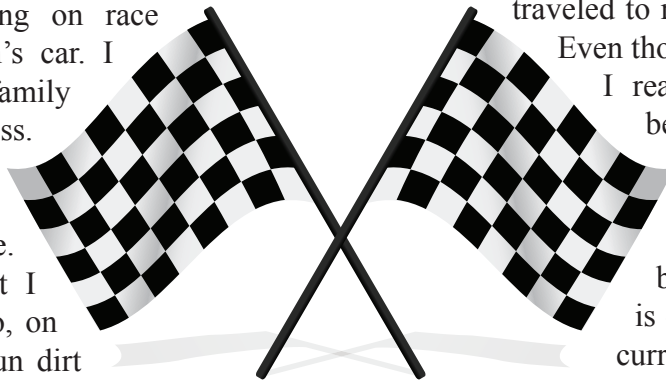
I try to make my cars eye popping enough to draw attention to them. These pictures show my latest design. I hope I have done the Foundation proud. I have not run this car yet, but I hope to get out in December. I like to get away once in a while and I have traveled to race in a couple of big events.

Even though I was not very successful,

I really enjoyed racing with the best. I raced in a class where the amount of money invested in a car made a difference in its performance. In the class I'll be racing this new car, everyone is equal so it should be fun. I'm currently waiting to receive the type of tires that all entrants must

have on their cars. I will be racing in the Syracuse, NY area mainly, but will be travelling to some big races as well, the first one being in Harrisburg, Pennsylvania in February at the Motorama event there.

Later in 2018, I hope to organize a fundraiser using a special race day to raise money for SPF and create awareness for this disease.



## Two Great Mobility Aids

By Craig Leda

SPG78 prevents me from walking. I am an outdoorsman and mobility devices are not built for rugged terrain. I found an adaptable device that works well in the woods. It is made in



Colorado for hunters to move long distances efficiently. It is called a QuietKat Trike ([www.quietkat.com](http://www.quietkat.com)) and retails for \$5000 to \$6,000. I replaced the stock seat with a seat for a mobility scooter that I bought on eBay. The scooter seat makes the QuietKat look more like a mobility device, which it now is for me. It is easily cleaned after use by a good hose down.

I stopped hunting this year due to my arms becoming more affected, but I am still able to get out and enjoy nature using my QuietKat.



Also, I bought a lightweight power chair, an Air Hawk, that weighs just 41 pounds with batteries. This saved me \$20,000 by not needing an accessible van. The chair folds down to the size of piece of luggage on wheels.

My wife is able to put it into our minivan without much effort. If or when the time comes that I can't easily transfer, then we will do the van conversion. Insurance didn't cover the cost of this chair, but it was less expensive in the long run because I did not need additional equipment to travel. Retail price was \$2080.

I'm still searching for a way to deal with snow and ice...



## Go Bag Checklist

By Sid Clark, Illinois State Co-Ambassador

Not too long ago I was rushed to the hospital; with HSP/PLS, it happens. A little bit of planning can save you time and aggravation when you have more than enough on your plate. I was happy I had most of what I listed below in a "Go Bag."

You should have these things at the ready when you need to go to an Emergency Room (ER) or to check in for a hospital stay.



### For the ER:

- Insurance Card & photo ID – usually the first thing they ask for.
- Prescription list – paper copy of what prescriptions you take, when, how often, who prescribed, including over the counter medications.
- Any special instructions and results of recent tests- include HSP/PLS.
- Your doctors' names, phone numbers and addresses.
- Your friends' and contacts' phone numbers.
- Your drug store, address and phone number.

### Additionally for a Hospital Stay:

- Medical Power of attorney – for some reason in the last stay each Department wanted one, so I take several copies.
- Advance directive form - the hospital may want its own form.
- Cell phone & charger – get the nurses station phone number. I have had a call bell that did not work. I had to use my cell to call to get help!
- Pens and blank paper (or small notebook) – make a list of questions you have for doctors and hospital staff; when they rush in I can forget what I wanted to know.
- Hand sanitizers – it may be a hospital but wipe everything.
- Mints, cough drops, or lip balm – hospitals are dry places.
- A small flashlight –I like to check things out if I wake at night.
- Small amount of cash - \$10 to \$20. I got a haircut for \$10 and vending machines and newspaper may require some coins. No jewelry.
- Finally mark your name and number on all your personal items.



## DIS-ABILITY LAW: ADA Requirements for Restrooms

By Mary B. Schultz



The ADA (Americans with Disabilities Act, Pub. L. N. 101-336 26 July 1990, 104 Stat. 328 (1990), 42 U.S.C. §§12101-12213) is landmark legislation that was passed on July 26, 1990, and became effective on January 26, 1992.

The ADA requires that all bathrooms, whether newly constructed or renovated, must be usable by people with disabilities. Title III of the ADA is the portion of the law that established structural requirements for the design and construction, whether new or modified, of ADA compliant bathrooms. The 2010 Guidelines specify requirements by application of the ADA for (among other things) bathrooms (like minimum size, turning space, door swing, stalls, toilets, sinks, and counters) and bathroom products (like grab bars, bath and shower seats, mirrors, and dispensers). The USAB (United States Access Board) ([www.access-board.gov](http://www.access-board.gov)) is an independent Federal agency devoted to accessibility for individuals with disabilities. The USAB promulgated guidelines for accessible design and construction or renovation called “ADAAG” (ADA Accessibility Guidelines).

On September 15, 2010, the DOJ (U.S. Department of Justice) published “2010 ADA Standards for Accessible Design” in the Federal Register. The 2010 Standards specified minimum requirements for newly designed and constructed or renovated state and local government facilities, public accommodations, and commercial facilities to be readily accessible to and usable by persons with disabilities. The DOJ also published “2010 Standards for Public Accommodations and Commercial Facilities”. The DOJ guidelines may be downloaded (for free) at [www.ada.gov](http://www.ada.gov). There is also a toll-free ADA Information Line at the DOJ at (800) 514-0301.

Because many of the requirements for bathrooms are difficult to visualize, diagrams are available in DOJ and other referenced publications. An excellent guide for accessible bathrooms is by the Bobrick Supply Company called the “Planning Guide for Accessible Restrooms”. The guide may be downloaded (for free) at [www.bobrick.com/Documents/PlanningGuide.pdf](http://www.bobrick.com/Documents/PlanningGuide.pdf).

Mary Schultz is a partner in the law firm of Schultz & Associates LLP, [www.sl-lawyers.com](http://www.sl-lawyers.com), 640 Cepi Dr., Suite A; Chesterfield (St. Louis), Missouri 63005, (636) 537-4645. Mary B. Schultz graduated from Northwestern University Law School more than 30 years ago, in 1985, and has been practicing primarily in Missouri ever since. Mary B. Schultz is admitted to practice in Missouri and Illinois. Mary Schultz was diagnosed with HSP in 2012, and through genetic testing has learned that her gene mutation is in SPG7. Mary has been a member of SPF since 2012.

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## Ongoing Collaboration between Foundations

Frank Davis, President Spastic Paraplegia Foundation and Frank McKeown, President HSP Research Foundation

People often wonder what HSP research is going on, where it is being done and who is doing it. They also wonder, “Do the researchers talk with each other? Are there results out there that we may not know about?”

For several years now, there has been a fruitful collaboration between the Spastic Paraplegia Foundation in the USA and the HSP Research Foundation in Australia. Successive presidents of the two Foundations have kept each other up-to-date with informal phone calls over the years. The first meeting of global support groups was held in Madrid, Spain in 2015. Among those attending were current presidents Frank Davis (SP Foundation) and Frank McKeown (HSP Research Foundation). A major topic of discussion was the need for, and value of, increased and more substantial collaboration. Following that meeting, the level and extent of collaboration ramped up substantially. A major reflection of this was the joint dollar-for-dollar funding of a drug validation study that was successfully completed in December 2016.

The relationship and collaboration has developed where the two Franks, Frank D from the USA and Frank M from Australia, regularly talk with each other, share progress and challenges, seek opportunities to speed up research to find an effective treatment, to maximize research impact and to multiply the efforts of researchers in different places through communication,

*Continued on next page*

collaboration and joint funding. Frank Davis commented, “I know what they are doing there and they know what we are doing here – and why. We have very open communications and trust and freely share our thoughts, because our fundamental interests are the same. We serve different communities of people, but especially when we are talking about diseases as rare as these, really we are just talking about one global community.” Frank McKeown added, “Anything that is good for people with these conditions anywhere, is good for people with these conditions everywhere. A rising tide lifts all boats and there are all sorts of advantages in close collaboration between our two Foundations. After all, it is only support groups like ours whose primary aim is to serve the needs and interests of the communities of people with these conditions. No other organizations have that as their primary aim – not medical professionals, not research institutes, not government. Of course, these bodies care about people with these conditions, but medical professionals and researchers cover various diseases. Their interests are much broader, as are those of government bodies and healthcare organizations.”

Both Franks emphasized that people should not fear missing out on news or information because of where they live. As well as the collaboration between the two Foundations, they are also seeking to facilitate the sharing of information by researchers working on these conditions. Whilst duplication of effort and results in research can be reinforcing and confirming, it is sometimes just double work that can waste time, effort and money. There is also a role for the Foundations in preventing needless duplication of research effort through information sharing and funding processes. Frank McKeown commented, “If we were a commercial organization, we would almost certainly be a single multinational with branch offices in different countries. That doesn’t make sense in our case because we are constituted and funded by donations from our respective communities. What does make sense is that we act like a multinational with largely the same goals and aspirations and with a compelling case to work closely together.” Frank Davis added, “This is the direction we are headed in, where there will be increasing joint effort as time goes on, and a better, brighter future for people with these conditions as a result.”



*Frank McKeown (l.) and Frank Davis at Madrid Conference*

## Gene Therapy – Our Latest Potential Treatment

*By Jim Sheorn and Malin Dollinger, M.D.*

Gene therapy is a new way to treat, cure and possibly even prevent genetic diseases. This is done by changing or replacing a defective gene. We have 23 pairs of chromosomes in the nucleus of each cell in our bodies and the DNA of each one containing hundreds to thousands of genes. DNA is the blueprint for how the body is formed and how it works. It is the same in every cell in our body and contains instructions for telling RNA which protein to make, depending on which kind of protein is needed in that part of the body. In the case of HSP and PLS there are genetically determined abnormal nerve proteins and structure.

An abnormal area of DNA may cause a genetic disease. Some genetic diseases have a single wrong place in the DNA chain; others may have several. Gene therapy works by correcting the abnormal DNA back to normal. HSP and inherited PLS likely have more than one area of abnormal DNA. Gene therapy uses corrected genes to prevent or treat a disease. If it is successful, it inserts or modifies a gene instead of using other standard disease treatments such as surgery, radiation therapy or medications. The pathways include a) replacing a mutated gene that causes disease with a normal healthy copy of the same gene, b) inactivating a “bad” gene that is causing a disease, or c) adding a new gene to the body. Gene therapy is now being tested only for diseases that have no other cures.

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Gene therapy is typically injected into the body and is often attached to a “carrier,” like a harmless virus that enables the new gene to go to the right place. But one of the risks and concerns of gene therapy is the possibility that the “new gene” can be taken up by some other unintended place in the DNA of that chromosome, or to another one, and cause a new disease or undesirable abnormality. One important problem is delivery of the new gene to the correct place in the body. A carrier known as a vector is genetically engineered to deliver the gene. For example, certain viruses are used as vectors, usually modified so they can’t cause disease themselves. A couple of gene therapies have been approved in Europe. So far none have been approved in the US. It is expected that the first of about a dozen gene therapies for very rare diseases may be approved within the next two to three years.

There are two kinds of gene therapy. *Somatic cell gene therapy* changes the abnormal gene in the specifically selected tissues/cells of the treated person with the intention of curing or helping a disease in that person. In the case of HSP and PLS, the target of therapy would be nerve cells. The other kind of gene therapy involves the attempt to change the DNA in the “germ cells”, the eggs and sperm that come together to form an embryo and then a new person. If successful, all future offspring of that affected person, and their children, and their children’s children, and so on, would no longer have the abnormal gene. This is called *germline gene therapy* and that process and its ethics are much more complicated. There might be other undesired mutations and adverse effects in later offspring, who could not possibly consent to the treatment since they haven’t been born or even conceived yet. There is currently great reluctance to try germline gene therapy and the government will not fund germline gene therapy research. A concern about treating someone with germline gene therapy is what will happen with that person’s children. Will they be born without disease or could gene therapy cause other health related problems? This is one of the significant ethical problems which occupy major discussions among scientists and the regulatory bodies/government agencies.

We now know that in the most common form of HSP, SPG4, and now called SPAST, there are mutations in the SPAST gene which encodes the microtubule-severing-protein spastin (which is inside our nerves). New evidence suggests that this one abnormal gene does not fully explain SPG4 HSP. HSP is likely not simply one abnormal gene, which produces one abnormal

protein, which interferes with how our nerves function. Gene therapy will not likely be simply the repair or replacement of a single abnormal gene, but would be much more complicated if it is necessary to “repair” several abnormal genes, instead of only one. There is an ample list of genetic diseases that we would like to see corrected and cured by means of gene therapy. There is still the scientific and ethical concern about the creation of deleterious off-target mutations. As is common nowadays, the ethical and legal principles and decisions have not kept up with the recent remarkable scientific discoveries and progress.

Is gene therapy safe? Current research looks at this important question; some of the possible risks include toxic side effects, inflammation and cancer. The National Institutes of Health (NIH) controls this research by providing requirements and guidelines when investigators are conducting clinical trials. An Institutional Review Board and an Institutional Biosafety Committee must approve every gene therapy clinical trial. So, gene therapy is not yet ready for “prime time” – used to treat almost all genetic diseases. It has been used for a few very rare terrible genetic diseases that now have no current treatment with a very small number of remarkable results.

Then there is the cost factor. Gene therapy is very expensive, for example because in addition to very high development costs – millions of dollars – often each patient requires a new specially formulated gene treatment that can be used only in that person. There is an ethical problem and concern about allocation of costs and funding for such treatment. Gene therapy is specific and will not affect all genes. Since there are now close to 80 different types of HSP, each one would need its own gene therapy to treat or cure. That of course entails significantly more money, more resources and most difficult, more time.

In developing new medicines and therapies, medical companies can spend millions of dollars in research that sometimes takes many years to produce a therapy that works. If there are not enough affected patients to make it profitable, the therapy may never begin the research phase. During the research phases, side effects may be found which could delay approval. Small patient populations, diseases with life-threatening issues and availability of resources can delay or prevent a therapy from coming to market. The FDA is in place to make sure pharmaceutical companies produce safe and effective medications. Many times, the rules and

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regulations create hurdles for medical companies to overcome. In the case of gene therapy, these hurdles will be tremendous. We will need a lot of time in addition to extensive research.

With a therapy that may only treat a rare disease, the cost of providing the therapy will need to be looked at and compared to current therapy. If a new therapy is available but will only offer a slight improvement and will cost more than what is already available, then likely insurance will not cover the cost of the therapy and thus this new therapy will not make it to market. If that new therapy has a risk of major side effects, that complicates things.

It is estimated that about 10% of Americans have a rare disease that is caused by a genetic malfunction. Much has been talked about the cost of insurance and trying to find a way to make it affordable for all those that would like to have it. It is estimated that if only 1% of rare diseases are able to be treated with gene therapy, the cost of care could rise to about \$3 trillion. That is about what is currently spent in the US on healthcare per year. Many people who take medicine can save money by taking generic equivalents. Since gene therapy is new, it will be many years before generic gene therapies can be produced (if that is even possible) to make this treatment less expensive.

Manufacturing and distribution will likely be expensive for gene therapy. Because rare diseases are associated with small patient populations, the therapies will not be able to be produced in mass quantities. Mass production typically reduces cost. Shelf life, or how long the prepared medication will remain effective, can cause the price to increase as well.

The number of gene therapies in development has increased rapidly in the past several years to over 1600. Of those, almost 700 are for cancer. Rare diseases accounted for just over 500. Rare neurological disease therapy is about 150. About 23 of these potential new therapies are in Phase III trials, the last phase before coming to market. They are not all guaranteed to be available. For example, it may be determined that the therapy caused other problems, is too risky or too costly.

We must distinguish between fixing somatic cells, the cells all over our bodies that do all these tasks, and fixing germ cells, the ones in eggs and sperm that combine to make new people. For living people, fixing their germ cells is a worthwhile goal, which might thus prevent their offspring from inheriting the HSP gene. Our abnormal “gene pool” would end with that type of

treatment. However, fixing our own germ cells to help our offspring would not help us personally, the suffering living. The other pathway, somatic cell gene therapy, would fix the abnormal nerve cells we ourselves have, so we, the sufferers, could be helped. It’s still very early, and we need a lot of research to discover just what role these new gene repair and replacement techniques may have for affected people. A very few persons, children with neuromuscular conditions in particular, have had such treatment with amazing results. We are anxiously awaiting the research progress to include HSP in the group of potentially treatable patients. There is exciting new research that enables gene therapy without cutting the DNA, thus making unwanted mutations much less likely. Other new research gets the new genes into our cells by attaching them to nanoparticles [very tiny carriers], then making a temporary triple DNA helix, instead of double. The body selects and keeps only the two normal DNA helix chains.

Gene therapy is a new and exciting opportunity to treat rare diseases. But it comes with many uncertainties and obstacles that may delay therapies for years to come. Stay up to date with the latest HSP and PLS medical updates by logging onto the Spastic Paraplegia Foundation website: [www.sp-foundation.org](http://www.sp-foundation.org). Look at News and Announcements towards the bottom of the main page.

To more fully explain the research, ethics, and legal aspect of gene therapy, there will appear in two issues of Synapse in 2018, a much more complete discussion and description of gene therapy, including the science, the results so far, a discussion of the ethics involved, as well as a section by attorney Mary Schultz on the legal aspects of gene therapy. As a basis, in part, for this article, we have abstracted an important reference source, the ICER (Institute for Clinical and Economic Review) report entitled, “Gene Therapy: Understanding the Science, Assessing the Evidence, and Paying for Value,” March 2017.

How can you help? Make annual financial contributions to the Spastic Paraplegia Foundation. The SPF’s main focus is funding medical research in hope of finding breakthroughs that lead to cures for HSP and PLS. Make sure that politicians in your area know that you support medical research, especially rare disease medical research. Be aware of medical research in your area. Offer to participate. Often researchers have a difficult time finding enough participants to perform their research project.

## Connections

### Austin Patient Connection – September 23, 2017

By Marlene Doolen, SPF Ambassador  
Central Texas Region

There were 17 people at the Austin Patient Connection on Saturday, September 23, 2017. We met in a separate, enclosed room at the Texas Land & Cattle Restaurant in Austin, TX. The separate room made it easy to talk with each other. We had lunch and went around the room introducing ourselves and asked questions that were replied to by others in the room.



One of the members of the Spastic Paraplegia Foundation (SPF) is Board Member, David Ress, Houston, TX, who talked about the SPF.

Half an hour before the end of the Connection, we gathered to talk and separated into two groups, one being those with HSP (Hereditary Spastic Paraplegia or Spastic Paraplegia) and the other with PLS (Primary Lateral Sclerosis).

A few of the attendees said they wanted to have a couple of Patient Connections each year. I told them this is possible now that we have a new South Texas State Ambassador, Janet Woodham, who lives in Houston. She will be planning a Houston Patient Connection in 2018.



### 17th California TeamWalk for Our Cures and Connection

By Linda Gentner, SPF VP

On September 23<sup>rd</sup>, about 40 people joined together at St. James Episcopal Church in Fremont, California, for our annual Connection and “Share and Compare Discussion” hosted by Linda Gentner and her family. This year, we welcomed 8 first-time attendees — 4 HSPers plus their friends/family. Everyone present enjoyed our annual “compare and share” discussion where we talked about how we progressed since last year’s connection. It was also a time for us to share tips and suggestions that we found to help us with our daily activities while living with HSP or PLS. As in past years, our raffle was fun for all and a big hit. The event raised approximately \$30,000 for research.



2017 CA TeamWalk & Connection Attendees



Host Linda Gentner (l.) with Jean Chambers

As people were leaving, they were already talking about and looking forward to next year’s event.

## SPFillinois Connection September 9, 2017

*By Sid Clark, HSP and Hank Chiuppi, PLS,  
Illinois State Co-Ambassadors*

On a beautiful September Saturday, we had 14 in attendance at our 15<sup>th</sup> connection. The great thing about our get togethers is we get to see each other and meet new HSPers and PLSers. We shared ideas about “go bags”. A Go Bag is a folder or bag containing the paper work and things you should have available if you need to go to an emergency room or hospital [See the *Go Bag Checklist article elsewhere in this issue*]. We discussed Terry Welch’s diet and protocols. Among other subject reviewed were vacations and travel considerations, WalkAide, neurologist appointments, handicap vans and baclofen pump problems.

We also want to thank those who brought treats for the meeting and even a door prize. On October 14th, we will have a presentation on motion therapeutics, a demonstration on how it works and its benefits.



*Group Photo includes L to R: BJ & Greg Irwin, Back Row- Paulette Chiuppi, Carol Clark, Sara Wright, Joan Morris, Debbi Sexton, Right Side Debbie Forsythe, Chris Sexton, Center clock wise Steve Beutelspacher, Carolyn Wright, Rich Fairairn. Sid Clark, and Hank Chiuppi.*



*Don't follow  
my footsteps.  
I run into walls.*

## SPFillinois Connection Follow-up October 14, 2017

*By Sid Clark, HSP and Hank Chiuppi, PLS, Illinois  
State Co-Ambassadors*

As a follow-up to our Fall connection in September, we had a therapist demonstrate BalanceWare. It is a non-invasive vest worn to help with walking balance problems. The therapist strategically placed light weights at one or more locations in the vest to enhance the stability of the trunk’s ability to counter-balance the body. Clockwise trying the vest are Lynn Staudacher, Joan Morris, and Hank Chiuppi.

For those who tried BalanceWare, there were mixed results. Since I [Sid] do not walk, I got the chance to watch and it did seem to help everyone, but to varying degrees. The help it provided was not black and white. It was not like you put on the vest and then took up hiking as a sport. When the vest was on, the wearers did not lean as much and I noticed there was much less scissoring. It also helped with alignment and control. Besides improving balance, their gait was noticeably faster. So, in the end, they did walk better.

Insurance as a rule does not pay for the vest. The therapist said the cost was around \$795. For MS patients they have a special price of \$425! It must be ordered and fitted by a trained therapist. Both of the therapists at our meeting were trained.



## Tina's "Cane" Tips

By Tina Croghan State Ambassador of Missouri  
and SPF Board Member

Recently, I came across a discussion in Facebook about tips for cane users in winter weather. I found everyone's ideas very helpful and thought that all of our community would benefit.

**David Pineault** asks, "What type of canes should I use in the winter? I found that it's not secure to use my cane when there is ice or snow. Any tips or ideas?"

**Vicki Gomes Petilli** replies, "Look online for an attachment for the cane—it is a little cleat that flip up and down. This will help in the snow itself. Carry an [unbended], sic large paperclip to scratch out the ice [or snow] once you get back inside."

She gives this website to find the Prong Ice Grip. <https://www.4mdmedical.com>

**Abdullah Meryem Oguz** says that he had, "... bought these tips (Ergocap) about 8 months ago," on Amazon. "However," he warns, "I can say that it is not [as] flexible as it [is] sic claimed. It is heavier, and grips better, but still slides on a wet surface. <https://www.amazon.com>

**Stephanie Bridgeman** adds, "They also have things that look like springs that grab the snow and ice. The claw or the pic can slide easier. I lived in MN and IL for much of my life."

**Michael Allan** gives an important tip. "Have those on my crutches. They are great, however for ice and snow, there is an upgraded version that has a set of teeth that you can pop in and out when needed to assure better grip." (For Icy and Snowy Terrain – [Ergoactives'] Ergocap X-Treme Terrain, [<https://www.ergoactives.com/collections/accessories/products/ergocapwinter>].)

Michael goes on to say, "I have been making and selling canes for 20 some years now. Have researched and tried many different tips. For canes in dry months, I would recommend Keen [Quest with] Aventure [Pivoting Tips], <https://keenhealthcare.com/keen-aventure-pivoting-tips.html>. However, for icy winter weather the Ergocap Winter tip(s) are tops in my book."

Whatever you choose—try it out carefully first before you end up in some dark parking lot and it starts to freezing rain and you're caught!



# INVITAE

## SPF Partners with Invitae

By John Cobb, SPF Board Member

SPF is excited to announce a new partnership with Invitae and their Patients Insights Network (PIN) program. Invitae is one of the leading genetic testing companies and their PIN will allow SPF to build a centralized network of de-identified patient information to help researchers and biopharmaceutical companies find treatments for HSP and PLS. As our network grows, the Foundation will be able to attract more research opportunities, so it is important that we maximize our participation. In this Information Age, data is in high demand, so it is important that we can provide researchers what they need. Participants of the PIN will also be able to access the de-identified information, giving them additional insights into how we are managing the challenges we face. Your de-identified information is only shared with your permission. You own it so you can decide at any point to be removed from the Network. Additionally, all personal identifiers will be removed. Only trained PIN managers will have access to that information, so you can rest assured it is safe.

We are very excited about this program and will provide more information on our website, [www.spf-foundation.org](http://www.spf-foundation.org), as it becomes available!



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# Spastic Paraplegia Foundation invites you to **PITTSBURGH** Annual Conference June 22-23, 2018

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**\*To receive discount, payment must be received by SPF.**

**Registration Begins January 15, 2018**