

Synapse

Volume 22, Issue 4 Fall 2019

Newsletter of the Spastic Paraplegia Foundation

Letter from the President



Dear Friend,

You know, since I work for the Spastic Paraplegia Foundation every day, it is sometimes hard for me to realize that we may have overlooked sharing very important information with you.

I do know that it is particularly important during this last quarter of the year that you be aware of the basics of all we do. So much depends on how well we communicate with you. You are our Rock of Gibraltar and I thank you so much for making all we do possible. It is you that has kept our all-volunteer, patient advocacy Foundation successful and growing for 17 years. Your thoughtfulness and generosity are making a huge difference in the lives of over 135,000 people worldwide suffering with the pain and agony of HSP and PLS. Thank You!

The Spastic Paraplegia Foundation (SPF) is a 501(c)(3) registered nonprofit charity organization founded in 2002 whose mission is to support the very best research possible to cure two very similar, rare, upper-motor, neurological diseases called Hereditary Spastic Paraplegia and Primary Lateral Sclerosis. SPF is the only foundation in the U.S. with this mission. Our Articles of Organization state that our focus is on Research, Education and Support. Therefore, practically all of our donation dollars are used to sponsor the very best research on this planet. We also use our website, Facebook Page, Synapse, Spastic World (our email blast newsletter) and our Annual Conference to help Educate and Support people suffering every day with HSP and PLS.

We have a very strong, specific focus. Our Articles of Organization were specifically written such that we can only use your donated funds for Research, Education and Support. We cannot use your donated funds to assist people with their medical bills or to help them buy medical or exercise equipment. Very fortunately, in the U.S., there are many charities that are built to help people in this way. There

is a page on our website (www.sp-foundation.org) with a list of many of these government and charity organizations, should you need to use them. On the site's first page menu, select "Resources" and then select "Government Aid and Charity Resources".

Our SPF Board of Directors are all very accomplished individuals but only a few have a medical background. You might wonder how we decide what research proposals to support. We are fortunate to have a great Scientific Advisory Board (SAB). You can read about them on our website. Only the most respected world-renowned experts on HSP and PLS are invited to be on our SAB. Fortunately, all members of our SAB work pro bono because they care about you and want our mission to be accomplished. Each research proposal that we receive is extensively studied and graded by at least two members of our SAB. They each turn in their reports to the chairman of our SAB, Dr. Martha Nance who sends us a report card, ranking every proposal from best to worst. The very best research proposals are ranked "Excellent and Very Promising" (EVP). Every year, half of our grant funding goes to EVP PLS research and the other half to EVP HSP Research. We support from \$600,000 to \$900,000 in EVP research proposals every year and each research proposal is funded for a 2 year period.

Dr. Martha Nance called us in tears last year saying that there were many more research proposals ranked as Excellent and Very Promising than we could afford to support. She said that it was so sad that we could not support more EVP Proposals as any one of those that we are forced to turn down could be a major breakthrough. Science is developing so fast today that it is an exciting time to be alive and there are so many possibilities for meaningful breakthroughs. Sadly, those scientists that we cannot afford to support have to find another project to work on and are forced to change their focus to curing another disease.

Oh, GREAT NEWS: our anonymous donor has again volunteered to match all donations received from 11/15/19 to 1/15/20, dollar-for-dollar, up to \$150,000. You will be

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

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getting my year-end, fund raising letter in early November and I hope you will be as generous as you possibly can with your fully tax-deductible donation. We need to finally discover those cures that are becoming more and more within reach every year.

You might be skeptical, thinking that HSP & PLS are such rare diseases that there are not enough people suffering with them to garner adequate financial support. What many people don't think about is that we are all surrounded by family, friends and associates that would love to help. They just need and want to be asked. About this time of the year, many of your fellow SPF members write an annual letter to their friends, family and associates explaining that they have a rare disease called either HSP or PLS, that a foundation has existed since 2002 called The Spastic Paraplegia Foundation with the mission of sponsoring research to cure HSP and PLS and they would appreciate a donation. If you should choose to do the same, just let your letter recipients know that all donations will be matched dollar for dollar, between 11/15/19 and 1/15/20, up to \$150,000. Please also inform them that our website

is www.sp-foundation.org where a credit card donation is easy to make. Enclosing an envelope with our address on it with your letter or mentioning our physical address if you prefer to email (Spastic Paraplegia Foundation, 1605 Goularte Place, Fremont CA 94539-7241) will also help.

Thank you again for all of your past support that has made cures for HSP and PLS seem so close that you can almost smell them in the air. Our programs and services wouldn't exist without friends like you who make the needs of others a priority in life. I hope this letter has helped answer most of your questions and explained how urgent, promising and exciting the current state of research is.

All the best,
Frank Davis

Frank Davis, President

PS: If you have any questions or concerns that I have overlooked, please do not hesitate to email them to frank.davis@sp-foundation.org. I'll respond as quickly as I can.

SPF Annual Conference 2020

By Norma Pruitt, Co-Executive Director, Annual Conference Coordinator

You may have already heard the SPF annual conference will not be held in Denver in 2020 as previously announced. It is regrettable, but the city is not so budget friendly. The Board is mindful of the cost of attending the annual conferences and the burden it places on some families. The annual conference is NOT a fundraiser for SPF, but an annual education and support expense. We scouted 20 hotels in the Denver area and only seven of those could meet our needs; however, the costs to provide meals and audio/video were almost fifty percent higher than the costs in previous years. The least expensive hotel guest room would have cost \$250 per night, not including parking or transportation costs to/from the airport. With all this in mind, the Board made a decision for the good of the SPF to choose a different city for the 2020 Annual Conference. As soon as the host city and a hotel are selected, more information will be made available.



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Awareness Activities – August 25-31, 2019

[The following are some of the HSP & PLS Awareness Week activities conducted by SPF members and reported to me in September. John Staehle, Senior Editor]

I handed out the attached fliers to (2) Neurologist, (2) Rehab hospitals, (4) Physical Therapists and (5) companies where I purchase equipment. I sent the email request to the members in Louisiana on September 9th. Stephanie Lindsly, Louisiana State Ambassador.

Some of the things I did for HSP & PLS Awareness Week are noted below:

- I wore one of the Awareness Bracelets that I purchased at the San Antonio Conference every day to bring attention to our situation. These bracelets were also given to family and friends.
- My Facebook page had a post about HSP Awareness week.
- I contacted WGN Radio in Chicago with the Press Release that was provided. They did not reply to my emails, this disappointed me. I will try a different approach next year to see if I can get their attention :-)
- I had a t-shirt on order that I planned to wear during Awareness week, but it didn't arrive till after the week was over. I will still wear the t-shirt to raise awareness.

Mary Levi, Illinois

I wear two rubber awareness bracelets. I talk to as many people as I can. I go to a gym (2) days a week and inspire from my wheelchair workouts. I do this from 9:30 am to noon then exercise in a pool for 1 1/2 hours. Tammy Nolan, Louisiana



Left: Jim Sheorn, Tennessee State Ambassador and SPF Board member, was interviewed by Ann Holt from the ABC news station WKRN in Nashville, TN. If you would like to watch, please use the following link. The segment is called Ann Holt's TN. It was [broadcast] on August 28th. <https://www.wkrn.com/news/middle-tenn-man-living-with-rare-genetic-disease-wants-to-help-others-live-their-best-life/>

I have forwarded your request to my 12 people with email addresses. As for me I have worn the HSP/PLS Bracelet 24/7 and told anyone who asked all about it. George Spowls, West Virginia State Ambassador

Tammy Booth Widseth did an interview on local news and also told her story in five video segments posted to Facebook. Tammy Widseth, Great Falls, Montana.

Adli's Awareness Hot Dog Fundraiser

We kicked off HSP Awareness Week with a Hot Dog Fundraiser. My 7-year-old daughter, Adli (SPG4), raised \$1,336.02 in 2 hours today. We served over 100 hot dogs in our little town in Iowa. We had such an awesome turnout for something we only published on Facebook a few days prior. We plan to make this a yearly event and the Fire Station has requested we hold it there in the future. We served hot dogs, chips, baked goods, and drinks. It was all a Free Will Donation. The weather was beautiful, we couldn't have asked for a better day! We can't wait to send the proceeds to the SP Foundation!



Left: Jeana Fraser walked 3 miles for HSP and PLS Awareness week August 25th-31st and posted several videos on Facebook.

Right: Greg Pruitt on the Air with Greg Dunker News Talk 94.3 Kentucky.

Austin Patient Connection 2019 -- August 31. I let attendees with HSP know the most important thing



right now is getting blood test to determine the HSP gene. More with the same gene could get research started. Attendees received two places to go for genetic testing. “How to Help” information and information to talk with Dr. Fink each month was given. Marlene Doolen, SPF State Ambassador, Central Texas

Emails have gone out to the Nebraska members with working emails. I sent the press release with a bit more information regarding the diseases to the Omaha media (3 TV stations/newspaper), but unfortunately, received no responses. I forwarded the press release to members outstate with a request that they provide it to their local media. Since I can no longer walk more than about 60 yards even with a walker, I did my 5 K on the NuStep exercise machine at my local YMCA. Kirk Winkler, Nebraska State Ambassador

I told my neurologist about the awareness and his interest peeked and he said he was going to tweet it out! Donna Dugas



I attached some photos from my ride (recumbent tadpole trike). Friends, family, co-workers engaged in a wide variety of activities — walking, soccer, elliptical machine, yoga & biking (no running). On the final day of Awareness Week, I rode my recumbent tadpole trike on Missouri’s Katy Trail. I gave away many of those rubber bracelets (thanks to Tina & Tim Croghan), talked up research needed for rare diseases like HSP & PLS, and gave “test drives” on my trike (popular with kids). Mary B. Schultz, Missouri

I participated in it by posting something about HSP on my Facebook page every day. I also used the #HSPandPLS to try to promote it. I did the virtual 5k by doing my exercise bike 5 days that week. Julie

Ann Samson, SPF Provincial Ambassador, Ontario, Canada



Annual Conference Presentation Summaries

There is no written summary of the presentation by Dr. Craig Blackstone. To view the complete video of his presentation go to https://sp-foundation.org/what_we_do/annual-conference-recap.html.

PLS Functional Rating Scale & Report on 2nd International PLS Medical Conference

Presented by Hiroshi Mitsumoto, MD, DSc, Wesley J. Howe Professor of Neurology, Columbia University Irvine Medical Center



Dr. Mitsumoto

Dr. Mitsumoto presented a specific clinimetric scale on behalf of the PLS Functional Rating Scale (PLSFRS) Study Group, a large number of neurology/PLS researchers from across the country. PLS is studied because it affects only upper motor neurons and is the rarest form of motor neuron disease. PLS causes life-long progressive severe motor dysfunction, but unlike ALS, survival is not an issue. Since

PLS has never been included in any clinical trials, perhaps because it is so rare, there is an unmet need. We need to study PLS to understand its cause and to develop treatment.

He presented the differences between PLS and ALS: for example, the average disease duration in PLS is 88 months compared to 11½ months for ALS. Dr. Mitsumoto presented the ALS Functional Rating Scale, published in 1996. He then presented a comprehensive newly-developed PLS rating scale, including comparison with ALS patients. This rating scale will be extremely important and useful to scientists and physicians studying treatments for PLS patients. The PLSFRS was determined to be a valid rating scale when PLS patients are in clinical trials. This will enable physician-researchers in different institutions to compare groups of patients that have similar stages and symptoms, analogous to other clinical staging systems in more common medical conditions, such as cancer and heart disease. For example, he determined that 12 months’ time

Continued on next page

is sufficient to test the efficacy of potential medications in future clinical trials in PLS.

It will also be possible to use the more sensitive PLSFRS in clinical trials of new treatments in ALS. Further understanding of PLS is necessary and helpful for developing future clinical trials in PLS.

The Second International PLS Conference was held May 3rd and 4th in Philadelphia. He presented a brief history of PLS, beginning in 1865, and ending with the above conference. PLS patients have never been included in ALS clinical trials. The need for a three-year unsettling waiting period before making a diagnosis of PLS (as compared with ALS) generates great frustration for patients and their physicians.

What we know about PLS is: (1) There are approximately 75 to 125 new PLS cases per year; (2) The cause of PLS is unknown; and (3) clinical features are variable. Various standard medical pathways for study in PLS, such as biomarkers, genetics and clinical trials, have mostly not occurred.

The goals of the 2nd International PLS Conference were to increase our awareness and knowledge of PLS, increase networking and expand PLS research. Various speakers discussed topics including electrophysiology, neuroimaging, PLS progression, biomarkers, genetics, and comparisons between PLS, ALS, and FTD (frontotemporal dementia). The outcome of this conference included the plan to have a PLS conference every two years, update the PLS diagnostic criteria, publish a supplement to an ALS-related journal, form an International PLS Study Group, and determine if there is NIH funding available to support a basic science study in PLS.

The video of Dr. Mitsumoto's complete presentation may be viewed on the SPF website at, https://sp-foundation.org/what_we_do/annual-conference-recap.html.

[This presentation summary was prepared by Malin Dollinger, M.D., Synapse Medical & Research Editor.]

Cellular and Molecular Basis of Upper Motor Neuron Vulnerability

Presented by P. Hande Ozdinler, PhD, Associate Professor, Department of Neurology Northwestern University, Feinberg School of Medicine

Dr. Ozdinler opened her presentation by acknowledging the many PhDs, MD PhDs and PhD candidates in her lab and a long list of the collaborators



Dr. Ozdinler

without whom we would not be able to achieve what we have in science. She also acknowledged the sources of funding for her research: the Les Turner ALS Foundation, the ALS Association, and the National Institutes of Health. Although her presentation focused on her ALS research on upper motor neurons, she reminded the audience that HSP and PLS are also upper motor neuron diseases and the results of her research benefits them as well. Dr. Ozdinler told the audience that she will go fast over some parts of her presentation because they are published data and are very scientific.

The Importance of Upper Motor Neurons – Upper motor neurons (UMNs) play a very important role in voluntary movement. UMNs serve as “the ‘spokesperson’ of the cerebral cortex for motor function. They have long axons that go from the brain to a very specific area of the spinal cord. The axons are 20 to 30 microns in diameter and up to a 1.5 m long to reach the sacral area. All cortical neurons feed the UMNs with information on what they want to do. The UMN then integrates all the instructions it receives and determines the message to be sent. Then it transmits an instruction to a very specific area of the spinal cord. Upper motor neurons progressively degenerate in ALS/Lou Gehrig's disease and in related motor neuron diseases like Hereditary Spastic Paraplegia and Primary Lateral Sclerosis. When the UMN is damaged, the connection to the specific area of the spinal cord is broken and the instruction for motor function is never received. That is why people with UMN diseases cannot move the way they used to.

To study UMNs, Dr. Ozdinler's team needed to be able to identify them, to label them. One way is to implant a fluorescent bead into the corticospinal tract of the mouse. This older method is known as retrograde labeling and requires a difficult and delicate surgery to implant the bead. Also, labeling efficiency may vary from experiment to experiment and it cannot reveal the details of cell morphology. The most appropriate method is genetic labeling, so Dr. Ozdinler's team engineered a mouse model in which the UMNs were fluorescent. The UMNs remained fluorescent from the mouse's birth to old age. That led to the development of the first reporter line for upper motor neurons which allowed her team to understand why UMNs become diseased.

Why Upper Motor Neurons Become Vulnerable – Thanks to the development of the reporter lines for UMNs and the technologies in the lab, Dr. Ozdinler's team is beginning to understand why UMNS become vulnerable. There are different underlying causes:

- Cytoarchitectural integrity (the arrangement of nerve-cell bodies in the brain, especially the cerebral cortex¹).
- Endoplasmic Reticulum Stress (ER Stress) – Under various conditions, protein folding in the ER is impaired

leading to the accumulation of misfolded proteins²; and misfolded proteins cause unexpected troubles.

- Mitochondrial dysfunction – the inability of the mitochondria to generate energy due to deterioration from disease
- Golgi apparatus defects - The Golgi apparatus, sometimes called the Golgi complex or Golgi body, is responsible for manufacturing, warehousing, and shipping certain cellular products, particularly those from the endoplasmic reticulum (ER).³

Some of these causes may be more prevalent in some human patients and other causes may be more prevalent in other patients. The team's next task is to identify which patients are more profoundly affected by which cause.

Upper Motor Neurons Mode of Deterioration – UMNs in the mouse models are fluorescent. A technique that was used in cancer research for years, Fluorescence Activated Cell Sorting (FACS), was used by Dr. Ozdinler's team to separate the corticospinal motor neurons that show primary vulnerability from all the other neurons in the cortex. P15, P30, P60 and P90 are mouse models at various stages of the disease. P15, the "child," is at a very early stage; P30, the "teenager," is still at an early, but noticeable stage of damage to the UMN; P60, the "adult," is at a stage where the damage is very apparent; P90, the "mature adult," is at a stage where the damage is significant. At P30, the neurons know there is something wrong and are trying to fix it. At P60, the neurons can't do anything about the damage. At P90 there's nothing that can be done.

Dr. Ozdinler corroborated with colleagues in Turkey and Italy to get samples from patients that show UMN involvement and progress of UMN loss. They tested a small sample, 5 or 6 patients at different stages of the disease, and discovered that 5 out of 5 had the same six proteins trending upward. She is trying to get funding to expand this study to a larger population of patients to see if the results of the small sample are duplicated in the large sample. If true, then they may be moving toward finding a biomarker for measuring the progression of UMN deterioration and loss.

RNA-Seq and proteomic studies begin to reveal the molecular basis of UMN vulnerability and progressive degeneration. Biomarkers, early detection markers, are beginning to emerge.

Drug Discovery Efforts – To begin the process of drug discovery there first needs to be a proven translation of what works on a mouse model will work on a human patient. Obviously, a mouse and a human are markedly different at the species level, but at the cellular level, the UMNs of mice are almost identical to human UMNs. The examination of the CSMN dendrite of the control mouse to a mouse model and the dendrite of a human control sample and a human with ALS looks the same. The same

comparison is true with the cell nuclei, mitochondria and endoplasmic reticulum (ER). Translation exists at the level of cells and neurons.

When you disassociate the mouse model cortex and put the cells in culture, the UMNs retain their fluorescence. It is easy to identify the UMNs from the other cortical cells. This is exactly what the drug companies need to move the discovery phase of drug forward. They can easily determine if a specific drug has a positive impact on the UMNs. Dr. Ozdinler is working with Dr. Silverman at Northwestern University to develop drug treatments that target UMNs. Her collaboration with Dr. Silverman has identified one compound that improves the health of upper motor neurons.

Summary

Upper motor neurons are not in the dark any more.

We begin to understand why they degenerate (more work is needed).

We are identifying biomarkers for upper motor neuron degeneration.

We developed a novel drug discovery/verification platform using upper motor neuron survival.

We are beginning to discover novel compounds that improve upper motor neuron health.

References and sources used by preparer

1. Source: Medical Dictionary for the Health Professions and Nursing © Farlex 2012
2. Source: Millipore Sigma, Endoplasmic Reticulum Stress (ER Stress)
3. Bailey, Regina, "Golgi Apparatus", ThoughtCo., March 11, 2019

A video of Dr. Ozdinler's presentation may be viewed in its entirety on the SPF website, https://sp-foundation.org/what_we_do/annual-conference-recap.html

[This presentation summary was prepared by John Staehle, Synapse Senior Editor.]

QR Code for Ease of Donations!

Using your smart-phone, scan this quick response (QR) code as a convenient tool for submitting donations to the Spastic Paraplegia Foundation. Scan it with your smartphone and a QR reader app and it will go straight to our donate page.

Your contribution allows for medical research of Hereditary Spastic Paraplegia (HSP) and Primary Lateral Sclerosis (PLS), together we are #hspandpls.



General Interest

The Alinker

Together we move differently & change the world for all who want to stay active, regardless of physical challenges.

(Source: Instagram, @the_alinker_world)

By Lori Renna Linton, HSP

The picture below is from a screenshot of the Instagram post that started the adventure that is retraining my brain. On March 5th, Selma Blair, a famous actress and mom and someone battling MS, posted herself on the Alinker with the caption: **I got places to go!**



Source: Instagram

awesome, yellow, mobility device. The director of my school, Hemma Poledna, took care of that. She helped finance the bike.

Cut to now. I have had the Alinker since August 15th when I flew from Vienna to Zurich to pick it up and to meet BE Alink, the inventor of the device that has literally changed my life. My first try: I could barely ride it. I had no business saying, “Yes, I’ll take it back with me.” But, I knew, that it was the right decision. I knew that I was going to have to practice every day to make it work for me.

Ideally, you should put your full weight on the seat and walk. I don’t think I need to say that having HSP (or PLS ... but I don’t have that), makes that idea very challenging. In my case, my hamstrings are so weak, that I don’t have enough force to push the ground in order to propel myself forward. Therefore, I must come off the seat, in order to have more force with which to push off.

On August 30th, I walked with the Alinker 4 times around a track (1600 meters/ 1 mile). It took me 107 minutes. Two weeks later, I walked that same distance with the Alinker but it only took me 85 min. I shaved 22 minutes off my time. My hamstrings are stronger: I can do one complete

I responded, thinking it was super cool and that it could possibly be what I had been looking for...something to help me exercise and strengthen my nearly non-existent hamstring muscles.

That was March. It stayed on my mind. However, the price was the main factor between me and the



Lori and BE Alink

standing hamstring curl (couldn’t get it off the ground weeks ago) and I am closer to sitting comfortably on the seat. I use the Alinker at school to get from room to room, I use it in the classroom so I can teach and I use it at the track so I can exercise. The most important thing though, is that it has given me legs. It has given me the freedom to imagine that an experience is not off limits because of my limited mobility. To facilitate those experiences, the Alinker is foldable for easy transport. Visit the Alinker website, www.thealinker.com, for details.

It has only been a little over 4 weeks since riding the Alinker and I’ve already noticed a difference. Let’s see what difference a year will make!

Lori is from New York and has lived in Austria for 17 years where she lives with her 2 children and teaches English to “fantastic students in grades 5 through 12.” She was a featured presenter at the 2016 Annual Conference in Chicago and then spoke at one of the break-out sessions at the 2017 Annual Conference in Atlanta. Both times her subject was Potato Pants.

WHEELCHAIR BOWLING

Everybody Struggled, Nobody Judged

By Frank W. Goebel II, HSP SPG4, Bowler; Secretary Rensselaer County Men (a competitive league); Secretary/Treasurer Mixed Nuts (a social league)

I’ve always been an avid bowler and until my mid-30s, I could walk to the line in a “normal” (mainstream) manner. I had a 300 back in 1999 with multiple 700+ series along the way. At the time, I hadn’t even thought of HSP or that I would have such a condition affecting my every day.

As HSP symptoms set in, I eventually had to modify my approach, first by compressing my steps to combat the drag. I later went to a one-step delivery that I took directly from a coaching drill I’ve used to teach others. I managed to do pretty well, all things considered.

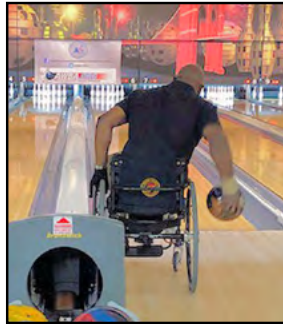
I got my first wheelchair around mid-May of this year and started bowling in it by the end of the month. I also joined the American Wheelchair Bowling Association (AWBA). Their membership requirements are quite broad in terms of types of disabilities. I then bowled two summer leagues to work on my wheelchair game and the AWBA invited me to Iowa



2019 Doubles Champions
Frank Goebel (L) and
James Tipton

for their recent tournament in Council Bluffs. Like most other AWBA events, it was run in conjunction with the local Great Plains chapter of the Paralyzed Veterans of America (PVA). Being a veteran, even though HSP isn't service-connected, I am eligible to join the PVA.

I went to that event mostly to meet other people, to hear their stories and experiences, from dealing with whatever caused their disability to how much they've bowled and to a few military stories. They were as interested in my background as well. I believe that I may have been the only person there whose disability had a genetic origin. They were as interested in my situation as I was in theirs.



Again, it was the experience that I sought and craved...and I certainly got it there. And then there was the bowling...so, competition was to be had. Thursday started with Doubles. I was paired randomly with an Air Force Veteran, James Tipton, "Tip," and we won that Doubles event.

Friday commenced a three-day Singles, which had six games bowled that day as well as Saturday, and three more on Sunday. In my division, I wound up in the top 5 and in a Stepladder Final, I managed a fourth-place finish. I had to file a W-9 as a result.

During a lull in the events, they held a bowling clinic for those who were new to either bowling and/or wheelchair use. A Friday night event included the Up/Down Tournament, a fundraiser where each lane had a team of one wheelchair bowler with four standing bowlers, many of them volunteers who had helped the event as a whole as a thank-you gesture. It was noisy, organized, and FUN.

Strikes and high scores don't come as often for wheelchair bowlers, so spares are paramount. I watched even the high average bowlers throw a frustrating "off" delivery, and I saw those with lower averages managed a few strikes along the way. At the same time, even though it was competitive, everyone rejoiced at every little success along the way, even if it was for a head to head opponent at the time. Based on my observations, I arrived at a four-word descriptor: "Everybody struggled; nobody judged."

I had such a great time, prize money notwithstanding, that I wanted to go to another tournament in Short Pump, Virginia, commencing on September 13th. I already expect to see many that I met in Iowa and I'll be bowling the Doubles event with "Tip" again. No matter what the outcome, I already know that I will have a great time.

Additional information: AWBA website: <https://awba.org/>

The Pros and Cons of a Diagnosis

By Michael Isber

A Geneticist diagnosed me with a recessive condition of HSP SPG 7 in early 2013. I was 57 at the time. It immediately followed my brother's SPG7 diagnosis. I have a spastic gait and weakness in the hips (as did my brother). I have two sisters with no symptoms of HSP. No one else on either my mother's or father's side of the family have symptoms.

There have been a lot of advantages to having the SPG7 diagnosis. I can see a neurologist with a diagnosis in hand and I also have accommodated parking at work which is a huge advantage.

However, there is one big disadvantage. Since my diagnosis, I tend to explain the worsening of any symptom on HSP. I believe in my circumstance, the defective gene causes symptoms of spasticity and weakness in the hips and also weakness in the back/spine, particularly in the lumbar region. This weakness can amplify my other symptoms.

In 2013 my neurologist recommended I see a particular Athletic Therapist. The therapist put me through a one-hour evaluation and stated, "the signal is there but it's not getting through". I was extremely happy as my assumption had been the signal wasn't there.

Almost everyone will suggest specific exercises. The big problem with an unfocused exercise regimen for me was there may be no benefit to an exercise and some of the exercises I was doing were bad for me. I showed a marked improvement when I left that up to the professional who in this case was the Athletic Therapist. I have regained a portion of the mobility which I had lost since my back injury in 2012.

I still have wonky hips but I am hoping for improvement. I believe it will take time to retrain my brain/body on moving and walking now that I am getting a bit stronger as I use muscles properly instead of having my body optimize movement, either to avoid pain or allow movement. Although there have been many strength improvements, my walk is still more labored than it was.

I am finding that a lot of what I am dealing with is due to weak glutes. I think as I have become less active, my glutes have become weaker and hamstrings tighter causing more back and foot problems. As I work on gaining strength in my glutes, I also have to stretch my hamstrings as they are so tight my legs don't straighten when walking. If I land on my toe instead of my heel when walking, my leg hyperextends and locks which is really not good.

Everyday activity can be better for me than my exercise regimen. I will forego some of my exercises one day if I can use that time to be more active. I am very careful with my back. No repetitive bending like picking up leaves. Now that I know more

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about the existence of bulging discs, I can avoid the things and exercises I was doing that were harmful. Examples are the rowing machine, some yoga positions, and sit ups.

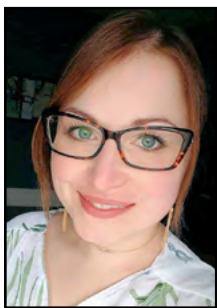
There is no doubt that my back and core are getting stronger and the strength and improved walking have given me good days which are better than before, but I still have bad days, especially if I am tired. To help avoid fatigue I am careful with my diet which includes a daily complement of vitamins/minerals. I often use the walking stick as this helps and does let people know to give me more space when I am walking.

It seems that once I had the HSP diagnosis, any decrease in ability is expected and I can incorrectly explain away on HSP. That is one big disadvantage to having a diagnosis!

I would say for me, that my condition of HSP has not changed much, but my ability to walk has gotten worse. Over 20 years I have gone from fairly athletic to moving like Frankenstein and at times, I have difficulty just stepping over a crack in the sidewalk. But in the last 6 years after my back injury, I have lost much general movement. I tend to think of my symptoms as either directly or indirectly from HSP. A weak spine from the defective gene I might consider as direct and therefore, more difficult to address, but a weak spine/back may cause many other issues (weak core, stiff ankles and weak glutes) which I might be able to address through exercise.

I am also aware that with a defective gene, the whole thing can go South. I guess that helps drive me to continue. Another diagnosis advantage?

How My Disability Changed My Perspective on Job Seeking



By Rebecca Porter, SPF Provincial Ambassador for the Atlantic Region of Canada

After she was diagnosed with a progressive condition, Rebecca Porter feared the time and effort she had put into building a nursing career would go to waste.

Job seeking can be difficult in the best of circumstances. Browsing through postings, hoping to see a position that will not only meet your financial needs but also allow you to build a long and meaningful career, can be frustrating. When living with a disability, you share the same insecurities as other jobseekers, but you are also flooded with emotion and concerns, wondering if your limitations (perceived or otherwise) will eliminate you from even being considered.

My name is Rebecca Porter, and I live with a condition called Hereditary Spastic Paraparesis.

I have been trained in and worked as a Human Services Worker, Pharmacy Technician and most recently became a Licensed Practical Nurse specializing in Alzheimer and dementia care. I live in Saint John, New Brunswick, work for a non-profit organization and volunteer with a local nursing home. I have a passion for caregiving and educating others on various health topics as well as advocating for those who do not have a voice.

Finding a job was never very difficult for me. I had a great resume and a lot of experience and education. This all changed after I was diagnosed with a progressive condition and my mobility began to deteriorate. I still had the same qualifications and experience, but now I felt burdened with a label and new restrictions. I was a nurse, but now a nurse with a disability. This newly attached addendum to my life felt like an apology with a “but” attached to it. I felt my skills and experience would mean less and by that, I would mean less as a person in the health-care field. How could I compete with those who had the same qualifications and knowledge, but were able to stand for hours on end, when I could not?

Being a nurse is an extremely physical job, but it was my passion. I was left struggling to reassure myself that I was still viable in my chosen field and that not only could I continue and flourish in my current vocation, but also all the time, effort and study I had put into my career was not without merit. I began to feel as though my life, which was tethered to my career in many facets, was now as limited as I had begun to feel physically. My mental and physical health began to suffer as I pushed myself more and more to show not only those I worked with, but also myself, that I could still be a viable member of a care team. I pushed myself so hard, in fact, that I now require crutches and, in many instances, a wheelchair to complete even the simplest of tasks that I once took for granted. As time passed, I began to feel more isolated and ineffectual.

I reached out to a friend to talk about my situation. He told me about an organization called the Canadian Council on Rehabilitation and Work (CCRW) and how they not only assisted him with finding a meaningful and appropriate career, but even more importantly, reminded him of his worth as a productive member of society. I took his advice to reach out and, after only three months, I found a position that suited my needs and accommodations, and also allowed me to support a population of people – often neglected and under-served – living with Alzheimer’s and dementia.

Advocacy organizations help people who may have lost their drive, or even their ability to feel productive and of worth, to see themselves as equal to their peers. This is no small task, as physical ailments can create or exacerbate mental ailments if a person is feeling undervalued at work. Much work is still to be done, but with the increasing number of inclusive employers and organizations such as

the CCRW working together, we are now making what was once impossible, possible.

Rebecca Porter is 27 years old and lives in Saint John, NB. In her spare time, she enjoys volunteering at her church, exploring New Brunswick's beaches and spending time with her husband, their Boston terrier and their cat.

Freedom Is a Lift Named Joey

By Debbie Davis, HSP SPAST (SPG4)

I have HSP. I am now at the stage of my disease that makes it very difficult to walk. I have used many mobility aids and I am at the place where I need to use a scooter or a wheelchair when I go outside my home. I originally bought a small 3-wheeled scooter, great for airports. The advantage of this scooter is it comes apart into pieces, the heaviest being approximately 25 pounds. This was great when I was with my husband as he could lift these pieces in and out of the trunks of our cars, something I could not manage by myself. It is impossible for me to use two hands to lift those parts when I have little balance and am very unsteady on my feet. I continued to be trapped in my home as I needed the scooter to ambulate.

The other problem with the 3-wheeled scooter is its instability from not having a fourth wheel; I managed to tip it over and fall a few times. I needed to get a larger more stable aid. So, I got an electric wheelchair.

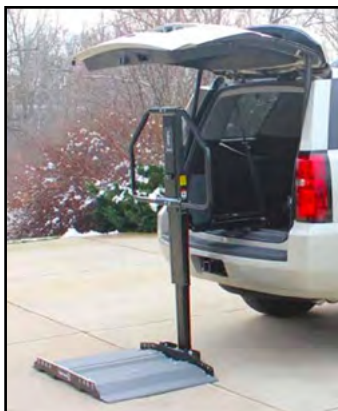
My local mobility specialist suggested a company that modifies vehicles to be able to transport scooters and wheelchairs. After examining all the choices and their pros and cons, I decided on a lift that lifts either my scooter or my wheelchair into and out of the back of my van.

This one is easy to use. It has no ramps that need extra room. A push of a button moves it up and down and into my van. I still have second row seating in the van with the lift only using the area of the third-row seating.

I keep a pair of walking sticks in the van to help me get from the lift to the driver's seat and back. I now feel so free!

The lift that I got is the Bruno Joey VSL-4400. This photo from the Bruno Independent Living Aids company website, www.bruno.com, shows the lift in the down position.

There are many other types of lifts out there, but this one was well suited to my needs.



I Went to the Beach

By Carol Parris, HSP

A trip to Hilton Head was mandatory to attend my daughter's destination wedding. I was happy to learn that my four children and eleven grandchildren would also be in attendance. Having HSP with an unknown gene deficiency and no family with the same disease, my disability keeps me from travel, but I was determined to attend.



My disease began when I was in my late 50's. It was a slow progression. I had recently become widowed and my disease was in the early stages. I just knew that the occasional tripping and noticeable walking differences were caused by something. My diagnosis took five years of tests: a spinal tap, MRI, body scans, and still no answer. After five years of these tests, a physiatrist in Pittsburgh told me I had the classic symptoms of HSP. I follow a baclofen regimen and go to PT, as my insurance allows. I exercise at home and walk with the aid of a rollator. I also have an electric scooter (paid by Medicare after an evaluation) in my car that is operated by a hydraulic lift, which I purchased. Also, last summer I had hand controls installed in my car and continue to drive. That expense was not covered by insurance. I replaced my tub with a walk-in shower and use a shower chair. All items necessary for me to continue living alone and take care of myself. The loss of bladder control is an added problem that is hard to accept. I receive Botox twice a year for that and purchase products for constant wear.

My disability is difficult to accept. I am sure everyone feels this way. For sixty years I had a normal lifestyle. Today, I get accolades for just venturing out. How sad is that? I always say what is the alternative, being an invalid or housebound? It is hard to get to my car and hard to unload the scooter, but I make myself do it. I try to go somewhere every day, even if it is just to the shopping mall or to the park with a takeout meal.

I do like my own company and have developed several hobbies. I am determined to keep living in my little first floor apartment, with no steps. I do rely on some social services, such as an aide who comes twice a week to do light housework, and meals on wheels. My daughter sends me home cooked meals and a granddaughter will sometimes accompany me to appointments to help with the scooter. I also rely on a van service to travel to the other side of the city for doctor appointments. My adult children are very busy with their own families and I try to attend their events where ramps are present.

Continued on next page

Until you need a ramp for access it is surprising how hard it is to find one. I belong to a fraternal organization and have accepted a board position that I can do from home.

So back to the wedding. I rode in my car with my son-in-law driving. We also had another car driven by my oldest daughter and two of her daughters. My car had the electric scooter which was used each time we made a “pit stop.” The family helped me unload the scooter in the interest of time during the 10-hour drive. Once we arrived at our destination, I ordered a beach wheelchair. The rental was \$195.00, for the week. It is a big chair with large tires and can maneuver easily on sand and in the ocean surf. It is ugly but does the trick. My son would push the chair to the edge of the surf, and I could dangle my feet and enjoy the ocean with the rest of my family. Mostly, I enjoyed being oceanside. You can also rent an umbrella, which is a must for this redhead. It worked great and I encourage anyone who is going on a beach vacation to use it.

My Search for a Diagnosis

By Tracy Hood, HSP SPG5A

My symptoms started in the spring of 2015. At first, it seemed to be a balance issue and then I started limping. After a diagnosis of a labral tear in my left hip, I went to see a physical therapist, who noticed neurological responses in my evaluation and urged me to see a neurologist, specifically for a movement disorder. This started a long journey and series of tests. MRIs were ordered of my brain and full spine, tons of blood work looking for autoimmune issues, vitamin deficiencies, toxicity, you name it. Everything came back normal, but it was obvious to doctors that I was in rapid decline without an answer. The next round of tests included an electromyography and a nerve conduction study, both of which came back normal. The neurologist who performed these last sets of tests came back with a diagnosis of Hereditary Spastic Paraparesis or Primary Lateral Sclerosis. I collapsed in devastation leaving the doctor’s office that day.

A couple of months passed and I decided to seek out a second opinion, as it seemed the diagnosis was one of elimination, and it seemed odd to me to get such a rare diagnosis in that way. Surely, other tests could be performed to get answers. I sought out another neurology team, and at the same time, developed a few more issues. Late fall of 2015, I was diagnosed with Acute Onset High Blood Pressure, Hypothyroidism and kidney stones. I was 44 years old and completely falling apart. I was spiraling and that is when my primary care physician put a package together to submit my case to Mayo Clinic in Jacksonville, Florida.

I went to Mayo a total of four times – I saw a neurologist, who ordered a lumbar puncture. In the meantime, I also

saw a new neurology team in my hometown, who ordered another round of MRIs. All tests, once again, came back normal. I was referred to a geneticist at Mayo, who confirmed a diagnosis of HSP, specifically SPG5A. Mine is recessive – both of my parents were symptomless carriers. I won the unlucky genetic lottery. I am relieved my kids won’t be affected. A month later, I was diagnosed with Asthma, attributed to a weakened diaphragm, brought on by HSP. So, I actually have four major health issues that all hit within one year.

Improving My Quality of Life

Fast forward to today and I am actively managing my health on multiple levels to fight this disease with everything I have. I have completely changed my diet – I eat clean, whole foods and I have cut out gluten and meat. I do eat fish. I have sought out the help of a personal trainer because insurance only covers physical therapy for 60 visits a year. She has been a lifeline – I have seen her twice a week for about two and a half years. She’s a COTA (Certified Occupational Therapist Assistant) and has had patients with parallel symptoms, so she can work with me therapeutically and she challenges me to increase my strength. The goal is to keep moving! I also invested in a recumbent Catrike, which enables me to bike safely. I live in Florida, so when weather cooperates, I’m in the pool for daily laps. Swimming each morning before work gets the body moving. Lastly, I get a therapeutic massage every three weeks to help relieve the tightness in my back and legs.

In addition to all of my meds (Baclofen, Gabapentin and Ampyra), I have added plenty of vitamin supplements, and I have added CBD Oil. The CBD oil makes a noticeable difference in reducing spasticity and improving movement. This is a MUST for me – my therapists, my trainers, and my neurologist can physically see improvements in the fluidity of my gait as well as reduction in clonus.

My quest is to stay active and to move as much as possible. The support I have from my family and friends, the guidance I get from my team of healthcare providers – my Neurologist, my Massage Therapist, my Physical Therapists, my Personal Trainers – are what drive me forward, one day at a time. My husband is my rock, my biggest advocate, and an active participant and supporter in my quest to manage my health and my physical wellbeing.

I still have dark days; I still get frustrated with my limitations, and how much function I have lost. I hate the feeling of being a burden on anyone. I choose to focus on controlling what I can and believing that one day, a cure will come. And until that day comes, I will keep fighting.

A Good Day

By Kasey Edwards

I was born happy. Growing up, my family was known for our happiness. But when we were told that my daughter, Robbie, had a rare neurodegenerative disorder, HSP SPG47, and, at the time, the 9 cases reported reflect severely intellectually impaired children who have regressed to paraplegia and quadriplegia, I admitted to my husband I would never be happy again. My joy in life had been depleted. He quickly told me, with a confidence I thought foolish, this was not true. To me, it seemed he didn't grasp the gravity of the situation. It turns out I was wrong and I have never been so pleased to admit he was right.



Photo: Brooke Whicher
Photography

After a short grieving period, I found myself soaking up every second of delight my children could bring to me. The turning point occurred when I asked myself two questions: Do I want to live a life where I am anxious and terrified daily of a disorder that, being so rare, has been deemed impossible to predict?

No. Should the day come when my daughter's muscles betray her, and we realize words may never become abundant, do I want to spend what may be her, and our family's "best" years with half of my mind already consumed by her "worst" years? No. I realized I needed to live for the day I was in. I became determined to see more smiles, happiness, and laughter out of my children at the end of the day than tears or disappointment. It became clear that, regardless of what happens in the future, I want our children to remember their early years as flashbacks of silly dance parties, constructing elaborate costumes from cardboard and decorative tape, indulgent smiles, and sticky hands from a coveted ice-cream treat.

I have never more understood the value in counting your blessings. While Robbie's disease trajectory is unpredictable, we have been told that her case is presenting mildly at this point. That is a blessing. The support through friends, family, and our amazing Early Intervention team has been phenomenal. My husband is my rock: consistent in his strength, security, and sense of humor. My son is patient and gentle with Robbie, a kind soul who genuinely enjoys his sister. And then there is Robbie. She is, quite frankly, the essence of joy. Her smile is utterly irresistible, with laughter surprisingly robust for such a quiet little girl. To meet her really is to adore her. It amazes me daily how so much personality can be projected out of a child with so few words and who faces so many challenges. On countless occasions, medical professionals have said that her personality will help her go far, and of this I am certain. These are just a few of my many blessings.

Since my decision to live for the day and to appreciate the smaller moments, every day has been a good day. There have been sad days and hectic days, busy days and long days. But I have not had a bad day. Every day my daughter shows progress is celebrated. Each milestone is marked with giddiness and elation, a happiness my children relish and, in turn, exude in their daily life. Should the time come when our worst fears become a reality, I will take a moment to grieve. And then I will re-evaluate what I consider to bring me joy and strive to attain it. I may have been born happy, but circumstances can defeat even the sunniest of dispositions. In these moments, when my pleasure in life does not come innately, I know that I have a choice. And I choose happiness.

*You can learn more about Robbie's diagnosis and progress by joining the "Cure AP-4" group on Facebook. SPG47 is one of four AP-4-associated HSP variants supported by Cure AP-4, the others are SPG50, SPG51 and SPG52. [Many people with the AP-4-associated HSP variants may have been originally diagnosed with cerebral palsy. With the advent of lower cost genetic testing, the current total number of people with AP-4-associated HSP, according to Dr. Ebrahimi-Fakhari, Boston Children's Hospital, is 156 with 53 being SPG47. Ed.]

Resilience Matters – Part One What is Resilience?

By Kathi Geisler, President of Life's Still Good



Resilience is commonly described as the ability to bounce back positively from setbacks and difficult circumstances – to find satisfaction and happiness despite change, traumas and challenges. It's a factor that allows us to thrive in the face of stress and loss.

Some people have a natural ability to be resilient – it's part of their makeup to be optimistic and look for solutions. That characteristic is not thought to be primarily due to genetics or things we were taught by our parents. Rather, it's generally thought to be a set of coping mechanisms we develop over time. Other people tend to naturally have low levels of resistance – they struggle to find their way and the physical, psychological and spiritual challenges can be overwhelming.

Researchers say there are six Roots of Resilience, all contributing to your ability to "bounce back" from life's set-backs. These are having:

1. **Meaning in your life** – This can be one's personal goals, religious faith, family or service to others that enables us to persevere when challenged

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2. **Confidence** – A belief and trust in one’s own abilities to positively manage whatever comes our way
3. **Support** – Feeling listened to, cared for and being part of a supportive social network, giving us greater resolve to persist
4. **Flexibility and Adaptability** – The ability to think, feel and behave in manners that allow us to see opportunities, find solutions and move forward
5. **A Growth Mind Set** – The belief that our abilities can be developed with effort, allowing us to learn and grow from mistakes and be proactive
6. **Emotional Fortitude** – The ability to manage our thoughts and emotions positively, allowing us to retain perspective and see problems proportionately

It can be helpful to take the time to consider these six Roots in understanding how you react to and manage challenges in your lives. Another helpful thing is to take the online Resiliency Quiz, by Al Siebert, PhD. Dr. Siebert is the author of the books *The Resiliency Advantage* and *The Survivor Personality*. Quiz: <https://www.resiliencyquiz.com/index.shtml>

For a little pick-me up fun, enjoy The Bounce Back Song: <https://youtu.be/5NpFgYfvA18>

Look for Part Two of the Resilience Matters Column, “Why Resilience is Important,” in the Winter issue of *Synapse*.



Kathi Geisler is a Co-Founder of the Spastic Paraplegia Foundation and a former Vice President of the SPF. She has HSP and lives in Massachusetts.

BILLY Shoes

By Jean Chambers, RN, HSP, SPF Secretary

This is about my new pair of shoes from www.billyfootwear.com. They were designed by Billy Price and his friends. Billy fell 3 stories as a younger person, broke his neck and is paralyzed from the chest down. He cannot move his fingers. He has managed to adapt a lot of items for his condition as a ‘low quad.’ But until recently, he had not been able to find shoes that he could put on without assistance.

That all changed with the development of the shoes pictured. They actually zip open and closed. The laces can be adjusted during fitting to accommodate high arches. Initially the shoes were made only for children, both in high tops and low-cut styles, and they have a terrific assortment of colors and designs. The children’s shoes are available on Amazon (don’t forget to use Amazon



Smile to have a donation made to SPF). Adult sizes, also in high tops and low cuts, are now available, but are not on Amazon yet. I purchased this pair online through Nordstroms. The insoles are removable. The first photo shows the right shoe zipped up and the left shoe unzipped. The second photo shows one of my shoes on its side to show the zipper. If you open the BILLY Footwear link (see above), you can see all the styles and colors available in both children’s and adult sizes.



My Journey from CP to HSP

By (Name Withheld by Request)

Recently, in my early fifties, I received the results of a genetic test confirming that I have Hereditary Spastic Paraplegia SPAST (SPG4). Given that my age of onset was infancy, it seems like it took a long time to reach that diagnosis. There were a number of factors that caused the delay. As a toddler, I was incorrectly diagnosed as having cerebral palsy (CP). Furthermore, my father’s age of onset was not until he was in his seventies and he was not diagnosed until his eighties. Given that he was an only child, I had no uncles, aunts or cousins that may have had the gene mutation. In addition, none of my siblings showed any symptoms of HSP.

Though my father’s physical diagnosis didn’t come until I was in my fifties, once it did come, it was much easier to speculate the real cause of my mobility issues. A closer review of my CP diagnosis report revealed that it was based more on speculation than conclusive evidence.

I feel like I have had a mid-life disability swap. I have traded my CP for HSP. On the downside, my “new” disability is degenerative. On the upside, even though a gene therapy solution has yet to arrive for my particular gene mutation, there is at least a hope of some kind of treatment for the problem that has been with me my entire life.

Broken Barn

By Max Reddick, 2019

Fresh pain peels back,
Revealing older cracks
And faded hues.

Useless painting this wreck,
Better to let me weather,
Give me time to breathe.

Sun, rain, ice, snow, and moonlight
Witness the calico barn’s deterioration,
Which is a form of healing.

[Poetry helps Max cope with his HSP. Ed.]

You Got Game?

Some inspirational statements below may help get you started or help keep you motivated to press on. No one said it would be easy, but no one said it might be this challenging either. Since life is not a dress rehearsal, keep in mind no one, even you, fails by trying. Gene Levin, 8/31/2019

Our greatest fear should not be of failure ... but of succeeding at things in life that don't really matter.

D.J. Moody

**Each one of us can make a difference.
Together we can make a change.**

Barbara Mikulski

It's better to be at the bottom of the ladder you want to climb than at top of the one you don't.

Stephen Kellogg

True life is lived when tiny changes occur.

Leo Tolstoy

Imperfections are not inadequacies, they're reminders that we're all in this together.

Brene Brown

Editor's Note: Gene Levin was diagnosed with PLS in 1980. Since 2013, he has served as the SPF State Ambassador for Western Florida. He explains the title of this series of notable quotes with: "I see life as a game. This caption [title] asks if 'ya wanna play.'"

Living with HSP/PLS

Incontinence, A Medical Perspective

By Malin Dollinger, M.D., HSP SPG4, Synapse Medical and Research Editor

Synapse's Senior Editor, John Staehle, wrote a very useful article on urinary incontinence, published in the Summer issue of *Synapse*, pages 14 and 15 (available online at <https://sp-foundation.org/news-resources/newsletter.html>). He very accurately and interestingly described his own experience and solutions. His incontinence was accentuated by having, at first, undiagnosed diabetes. The cardinal symptoms of diabetes are excessive thirst, excessive appetite, and excessive urination. That of course made the incontinence much more difficult to control. So those with excessive urination, especially with incontinence, should be tested for diabetes. My daughter had consultations with urologists before it was realized that some with HSP have bladder symptoms. There are a variety of local and practical measures that John has well-described, such as the timing of urination and especially self-catheterization, if needed. Wearing some type of pad or protective garment may be very useful. Stress incontinence refers to loss of urine when "bearing down," such as when coughing or moving your bowels, and for some, it may be the only time when they lose some urine.

His article is a very useful summary. Fact is, I could have written much the same article about myself, an elderly man with HSP and diabetes. Yes, age of course may bring on incontinence anyway. I carry with me at all times a collection of paper towels or pads to fold up inside my underwear. I have special underwear that can be put on and

removed from above, "over my head," like an undershirt, so in the middle of the day, if the problem is more severe, I can change underwear as well, without needing to get undressed/ remove my pants, shoes and socks. All I need is a bathroom, and in half a minute I'm dry again and ready to go. I keep underwear and towels in a bathroom drawer at home, so I can routinely solve the problem during a regular bathroom trip.

Before discussing the causes of incontinence, I'll make some practical suggestions for control. One simple aid, for men who routinely get up at night one or more times to pass urine is to not drink lots of water after dinner, and to empty your bladder before you go to bed. One kind of incontinence, called "overflow incontinence," can be diagnosed by a urologist using ultrasound imaging or by passing a catheter to measure bladder urine residual after you "think" you are done urinating. In overflow incontinence, you always have some urine remaining in the bladder, and when it reaches a certain amount, say 300cc or 400cc, you have an urgency to urinate. But when you do "empty" your bladder, you removed only the amount of urine over that 300 to 400cc amount, and then very soon thereafter, the additional smaller amount of urine, added to what remained there, again gives you the urgency to urinate. This results in frequent urination of relatively small amounts of urine -- and a lot of frustration, especially at nighttime.

As men grow older, their prostate glands, which surround the urination tube, the urethra, enlarge and restrict outgoing urine flow. This may produce frequent urination, and if muscle weakness or lack of muscle control occurs, there may be incontinence. Getting older increases the chance of developing incontinence, but it is not inevitable.

Continued on next page

There are various mechanical devices to assist with control of incontinence, such as permanent catheters. These have the disadvantage of being awkward and somewhat cumbersome and there is a significant chance of developing a chronic bladder infection. In his article, John Staehle has well-described intermittent self-catheterization. There are physical devices available which directly compress the urethral tube. This problem of incontinence is best discussed with a urologist when it first happens.

There are many causes and kinds of incontinence and the degree of incontinence ranges from a loss of only a few drops or small amount of urine, for example when coughing or straining in the bathroom, solved by such measures as a pad, to more serious amounts of urine requiring professional diagnosis and solutions.

The solutions to help with incontinence may depend on the cause. Urinary incontinence is nicely described and summarized in the *Mayo Clinic articles on Patient Care and Health Information*. According to the Mayo Clinic, causes of urinary incontinence include:

- Stress -- when you cough, sneeze, exercise, lift something heavy, or laugh;
- Urgency -- a sudden desire to urinate, which can be caused by an infection, a neurologic disorder or diabetes;
- Overflow -- when your bladder does not completely empty, described above;
- Functional -- when you cannot get to the toilet in time or cannot get unbuttoned, for example from arthritis

Temporary incontinence can result from alcohol, caffeine, carbonated drinks, artificial sweeteners, chocolate, chili peppers, certain foods such as citrus, some heart and blood pressure medications, sedatives and muscle relaxants and large doses of vitamin C. Medical causes include bladder infections, constipation, pregnancy and childbirth, menopause, hysterectomy, enlarged prostate, whether benign or cancer, stones in the urinary tract, and various neurologic disorders, including spinal injury, MS, Parkinson's and stroke.

Thus, if incontinence is a significant problem, medical evaluation is indicated, especially if it is in relation to a particular food, activity, medication or other symptoms such as those of diabetes. Or if your life is significantly affected, even if only mild.

Remaining Positive in the Midst of Uncertainty

By John Hayes, HSP SPG7

With so many challenges, and new emerging symptoms and symptoms that won't get better, it's understandable

why someone would succumb to a sense of inevitable decline, and the subsequent inactivity and pessimism. It stinks that we have the condition we have, but we can't allow ourselves to accept that we are victims with no hope things can get better.

While it varies overall and by geographic area, it's true what a Facebook poster wrote recently: "... just learn how your body works, this is a rare disease, doctors don't have the answers yet." This comment was written about a specific issue, but the gist is true: 1) Each person has issues that affect the body, some are somewhat common, but each of us is unique in the way we are affected and many have symptoms that are specific to just that person; 2) Doctors don't have all the answers, yet the medical knowledge on HSP, and the specific genetic types, advances all the time. Please don't fall into the trap I did for a while of just living with the condition and relying on what my doctors told me. Take charge of your health. Take the initiative to facilitate your knowledge of the condition and your own body and be an advocate for your medical care.

The significance of being positive for yourself and those around you is enormous. Not only will you make yourself open to try new things, you're more likely to accomplish them. It will also show others that you are perseverant. You can be a model for your significant other, your children, extended family, and friends. Sometimes you may need to take a step back, and rethink your position. Rather than dwell on what you think/know you can't do, think about what you might be able to do. On a side note, I admire my father. Sure, I reaped the rewards of his recessive gene, but he has, and continues to have, a positive outlook despite his own considerable health issues related to Parkinson's. He is an inspiration to me, yet unfortunately I didn't inherit his ability to think positively on a consistent basis. It's sometimes a challenge for me, and I need to remind myself of the model he set.

The reverse of being positive is also extremely significant. The impact of persistent skepticism on others and yourself is draining. Being positive doesn't suggest you are saying your condition isn't a major issue. Regardless of the level of understanding and support you may feel is afforded you, don't let yourself be a downer. People don't want to be around a person who has that attitude.

Here are a few strategies I employ.

Find and share some humor about your condition. This doesn't at all suggest you should practice self-deprecating humor or that your ability to find humor in your circumstance somehow shows that you're weak. For example, I've indicated that I was disappointed to be passed over again in the National Football League Draft, but I still hope that next year my name will be called and my picture will be put up on the big screen (I know it may not be considered

“real” football where you are). Another message I’ve used in describing this condition to others is how I’m a member of an exclusive group! I boast that it’s very hard to get into this group, but I did.

Think about, and write down, goals for things you want to accomplish. Research shows we’re more likely to accomplish tasks/goals when we write them down. It could involve visiting a specific place, interacting more, an exercise regimen, or whatever you desire. Unlike my humorous example above, goals need to be realistic, or frustration will ensue. Perhaps you’ve heard of SMART goals? While doing a study on SMART goals is not the point here, nonetheless, the “S” is for Specific, the “M” is for Measurable, the “A” is for Achievable, the “R” is for Relevant and the “T” is for Time-Bound. Each of these is important, but what’s more important is that you take the initiative to come up with goals. Some examples of good goals that adhere to the criteria mentioned above include:

- I will do 25 stand-up exercises from my chair every day before noon.
- I will attend weekly (your area) support group meetings by October 1, 2019.
- I will literally visit Zurich, Switzerland, by February 1, 2020.

Never give in to the condition you have, and never give up. It wasn’t fair, but we need to find a way to do something positive about it. Find something you can improve or even maintain. Maintenance of what you have now is a goal, too. Find a way to take “Baby Steps” to make continual progress (I remember this from the movie *What About Bob*). Developing a sense of accomplishment, finding support from family members, friends, counselors, and HSP Facebook Groups, and discovering some degree of humor about your condition can help build a more positive approach to your challenges.

I absolutely hate that I think I sound preachy here. I just want people to make more of their circumstances. I’m also motivated by my own mistakes. You are not alone!

Flora Brand’s Story: Living with PLS

Interview by Mary Ann Inman

Flora’s described her illness onset, “It all started with a fall when I was 48 years young. At the time I worked at the college bookstore as an accounting clerk. When I returned home it was raining. My feet and shoes were wet. I slipped, fell, and broke my wrist. During my wrist recovery physical therapy, I noticed that my balance was off. I chalked it up to nerves. Then my primary physician had me see a neurologist because my balance issues became worse. After numerous tests, it was thought to be either ALS or

PLS. Only time could tell. After about a year and no other symptoms appeared, the neurologist thought it was PLS.”

In the beginning Flora’s voice sounded strained so her primary doctor referred her to an Ear, Nose, and Throat (ENT) doctor. The specialist saw that one of her vocal cords was slapping into the other. She was given Botox injections in one cord to relax it. It worked. Her voice was back to normal. But Botox wears off and had to be repeated every 4 to 6 weeks. After a year of treatment, she stopped and to her surprise, her voice stayed normal. Life has been good for Flora!

Fast forward 25 years. Today, Flora is age 72. Although her balance issues remain, nothing has changed. Flora was able to care for her infant grandson during the week. Her husband Douglas mounted a baby carrier onto a second walker and she was able to move him all through the house. Flora adapted her moves. She sat to remove him from the carrier and put him on the bed to change him. This method worked and allowed Flora to care for his little sister, too.

Flora summed up her experience with PLS, “I know that I’m very lucky to be this mobile still. I clean house, other than my husband runs the vacuum and mops every week. I wash clothes, cook and do laundry. Like I said, after almost 25 years I can still do most things. That is my story.”

Lock Laces

By James Miller

I bought these a while ago from Amazon and haven’t had to mess with tying shoelaces since. Sometimes I adjust them for a looser or a tighter feel, all depending on my mood or if edema sets in. I hope these can help others. They come in a variety of colors.

[Search Amazon for “lock laces”, single pair for \$7.99, 2 pair for \$14.99; Amazon Prime members get free 1-day delivery. Use Amazon Smile to earn a small donation to SPF. Ed.]





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Fundraisers

The 2019 Combined Federal Campaign



The Combined Federal Campaign or CFC is a fundraising campaign the Federal Government offers its employees. It begins September 1st and goes through mid December. Federal employees are

allowed to pick from over 200 registered nonprofits to contribute to. Many CFC fairs will be held at Federal facilities throughout the US. This allows employees to learn about the nonprofits and make their selections.

The Spastic Paraplegia Foundation CFC number is **12554**.

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

Millions of dollars are raised each year through the CFC. The amount the SPF receives is somewhat small since little is known about HSP and PLS. Please let your federally employed friends and family know about the SPF and educate them about how you are affected so that we can take advantage of this great opportunity.

Golfing for Rare Diseases

By Kari Averill, SPF Kentucky State Ambassador

SPFKentucky raised \$23,720 with our 5th Annual Golfing for Rare Disease fundraiser on September 7, 2019 at Drake Creek Golf Club to help raise awareness and fund medical research for HSP and PLS.

We couldn't have asked for a better day. The weather was perfect! We had 25 teams this year; our best yet. We also want to say a great big thank you to Rickie and Denise Goode and all their family. They are a new family this year and a partner in this effort. They worked hard recruiting teams and sponsors and helped big time in helping us reach a new level of fundraising! We couldn't have done it without them. Also helping Golfing for Rare Diseases was Kira Przybylski and Tina and Tim Croghan. Thank you to all the golfers, sponsors, those who donated and of course,



the hands and feet that came together to make this event such a success!

We invite everyone to participate in next year's fundraiser on September 12, 2020. Save the Date and start raising funds now! Follow us on social media at #HSPandPLS and SpasticWorld #hspandpls. You can also contact us at SPFKentucky@gmail.com.



The Hands and Feet

California Virtual Walk 'n' Roll-a-thon October 5, 2019

By Linda Gentner, SPF Vice President

Wish we weren't under the overhang, but inside 42 enjoyed good food, great raffle items and lively conversation. This year we had 7 new attendees - both PLS and HSP and caregivers. We had an informative SHARE AND COMPARE discussion. We were fortunate to have Jody Westbrook, geneticist from Invitae, to answer interesting questions regarding genetic testing. Everyone had an enjoyable time! SPF was well represented having three board members (Jean Chambers, Linda Gentner and John Cobb) and one former board member (Kris Brocchini). Initial amount raised for SPF is around \$20,000; however, more donations are expected.



SPF Illinois Connection July 20, 2019

By Sid Clark, HSP and Hank Chiuppi, PLS; Illinois SPF Co-Ambassadors

With outdoor temperatures in the 90's we had our 21st connection since we started in 2011! With a number of new members, we had 17 in attendance. At a connection we work together and share with each other what works and what does not work. We talked about the need to join and update SP-foundation records. When shopping Amazon register for "Amazon Smile" with the credit going to Spastic Paraplegia Foundation, Inc.

From the Spring Synapse we highlighted Malin Dollinger's article on "HSP/PLS Surgical Anesthesia". The references were printed and all have a copy they can use when the need arise. Thank you Dr. Dollinger. Several had been at the national convention and they gave their thoughts and impressions. You can see the presentations on the SP-Foundation site. As with our local connection it is good to meet other HSPers/PLSers face to face. We covered individual stories of doctors, therapies, drugs, restless leg syndrome and alternatives to Baclofen. A question on the effectiveness of the pump was brought up and the majority of those with the pump did not like it.

Sharing with others with the same condition is rewarding. Many thoughts were shared during the socializing after the meeting. Thank you for the treats. To allow for some pre-planning our fall connection is slated for Oct 26.



Pictured back L to R: Leena Armin, Frank Madrigali, Paulette Chiuppi, Carol Bagal, Byron Capersen, Tom Inman, Mary Ann Inman (WI Ambassador), Mary Eileen Levi, Teresa Flemming, Carol Clark, Barbara Capersen, Shay Amin. In the center: Sid Clark, Phyllis Madrigali. Inserts Top to Bottom: Stewart Flemming, Richard Fairbairn, and Hank Chiuppi

Central Texas Patient Connection August 31, 2019

By Marlene Doolen, HSP, SPF State Ambassador – Central Texas Region

The 2019 Central Texas Patient Connection was held at La Margarita Restaurant, Round Rock, Texas, with 12 attending.

Mr. Gary Cohen, United Access, Pflugerville, TX, spoke about driving a car with hand controls and/or side or back of car ramp. One attendee uses hand controls and a back of car ramp. The other attendee has no hand controls but does use side of car ramp.

"How to Help" information and information to talk with Dr. Fink the first Tuesday of each month was given to each attendee.

The Spastic Paraplegia Foundation (SPF), says the most important thing right now is getting a blood test to determine your specific HSP gene. More test subjects with the same HSP gene could get research and clinical studies started sooner. The two companies that have genetic testing are: **Athena Diagnostics**, 200 Forest Street, 2nd Floor, Marlborough, MA 01752, (508) 756-2886; and **Invitae**, 1400 16th Street, San Francisco, CA 94103.

SYNAPSE APPEAL

Synapse costs lots of money to print and mail, and we need your help to keep it going. If you or a loved one enjoyed this issue and would like to support it, please use the enclosed response envelope to make a donation.

Every little bit helps.

1605 Goularte Place
Fremont, CA 94539-7241

Inaugural Wisconsin Connection October 19, 2019

By Mary Ann Inman, SPF Wisconsin State Ambassador

The Wisconsin Connection was held at Doolittles Woodfire Grill on Madison's east side on Saturday, October 19th from 11 am to 2 pm. Nineteen attended including HSPers, PLSers, spouses and caregivers.

A second meeting is planned at the same place and time on Saturday, April 25, 2020. For more information, contact Wisconsin SPF Ambassador, Mary Ann Inman at inman_ma@yahoo.com.

Highlights of the meeting were:

- Tim Bryden (Ripon) showed his wood turned holiday ornaments and talked about the value of staying active with hobbies and water exercise.
- Charlie Squires (Madison) and Brad Schlosser (Muskego) showed features about their portable wheelchairs.
- Dick Mace (Brookfield) and Craig Bunkoske (Beaver Dam) set the mood with humorous comments. Carl Reiche (Wild Rose) and Jeff Louk (West Bend) helped keep the meeting moving.
- Diane Stahl (Hudson) traveled the furthest to attend the connection (about 240 miles each way).
- The group talked about airport accessibility and free use of mobility aides and services such as Katie's Closet and ALS clinics.

- Lisa Hillis (Evansville), Nell Mace (Brookfield), Tim Bryden, Charlie Squires, Carl Rieche, Jayne Romano (Waukesha), Craig Bunkoske, Jeff Reid (Madison) and Bradley Schlosser (Muskego) shared family HSP issues including: family history, mobility aids, genetic testing, and experiences with the baclofen pump, batteries, and refills.
- Diane Stahl, Jeff Louk and Mary Ann Inman (Clinton) noted issues with speech related to their PLS.
- Shared topics discussed were the value of humor, exercise, diet, fall prevention, safety communication, finding a good neurologist, the National SPF Conference, Dr Fink podcasts, Facebook support groups and Synapse articles.
- Last but not least, spouses and caregivers were recognized for being there for us!

Clockwise from left: Jeff Reid, Bonnie Reid, Carl Reiche, Craig Bunkoske, Nell Mace, Richard Mace, Tim Bryden, Brenda Bryden, Jayne Romano, Tom Romano, Bradley Schlosser, Diane Schlosser, Charlie Squires, Tom Inman, Mary Ann Inman, Jeff Louk, Kelly Louk, Lisa Hillis, and Diane Stahl.

