Synapse

Volume 20, Issue 3 Fall 2017

Newsletter of the Spastic Paraplegia Foundation

Letter from the President

Dear Friend,

Today, I want to talk to you about two very simple but giant topics - the Good and the Bad. I'm going to perhaps sound very immature and naive but I think it is something that, nevertheless, needs to be said. I will start with the Bad.

The Bad is exemplified by something that happened just the other day in Charlottesville VA. By the time this *Synapse* is printed it may be old news and maybe even forgotten. But, this is a story that should not be forgotten.

It scares me to think what is happening with political and almost any dialogue in this country. I am afraid when white supremacists waving Confederate flags, chanting Nazi-era slogans, wearing helmets and carrying shields chant phrases like "Jews will not replace us."

They came to protest the removal of a statue of Robert E Lee. I have read many biographies of Robert E Lee and I can promise you that he would be very disappointed in the lack of knowledge that people who want to remove his statue have. He would be utterly ashamed of the white supremacists who were protesting the removal of his statue. He would refuse to be on any part of their team.

There is every reason to be afraid but I don't want us to live in fear. The mission of SPF, as you know, is to find a cure for two rare upper motor neurological diseases, HSP & PLS. This country suffers from a heart disease. Charlottesville is a symptom of that heart disease. I think the Good each of us can do is to take heart. I want us to see the light that shines in the darkness. I want us to decide to not follow the closed-minded hateful perspectives that so many people follow, both left and right.

This does not begin in Charlottesville. It begins in our hearts where racism, violence, hatred and indifference often begins. It entails life giving courage which confronts evil head on but refuses to become what it hates. It seeks reconciliation and wellbeing of all people. It has no interest in destroying or humiliating the other. It is, in short - LOVE.

One of my favorite life mottos is one from Augustine (354-430) who said, "Love and do what you will." As naive as it may sound, if everyone could really, honestly try to make love the premise of all that we do and all that we think, then The Ku Klux Clan and the Nazi party, who survive on hate, would disappear. So much of the ugly rhetoric that we see on both sides of the political arena could subside and we could try constructively to work together and get things accomplished.

What's more, we in our Foundation can also work together right now (in love) toward a cure for HSP and PLS. This is the ideal time for each of us to be thinking about our peer-to-peer fundraising. I plan to write my annual letters to my friends and relatives to remind them of our battle to find a cure for HSP and PLS and let them know all the incredible progress that is taking place. You can get progress information from our 2016 Annual Report on our website. You can also find examples of good letters to write to your friends and family on our website.

I am excited about all the Good that is taking place with HSP and PLS research. I firmly believe that a cure is becoming more and more within reach but it is we who need to do that reaching. We will soon be doing our year end fundraising and I hope you will try your best to share in the Good that is taking place by donating generously.

All the Best,

Frank Davis, President

Frank Davis

President

Old Paraplegics Never Die…

They Just Walk Away

Almost all of us wonder, especially when older
Just what will be the final cause, the final event
There are enough fatal maladies to go around
Heart disease, cancer, strokes, accidents to name a few

But our lives are not only punctuated by
Infrequent, or sometimes frequent episodes
Of these well-worn maladies
They are continuously affected by our paraplegia
Because 24/7 every waking moment and even sleeping moments
We're on a roll…if we're lucky

There are a host of related maladies that we own
Predominant in us, but rare in others
Bladder and skin problems
Major risk of associated injuries
Side effects of treatment: medications, pumps
And being stationary/sitting almost all the time
Bone and muscle wasting
The emotional stress of "physical limitations"

We remember the day, the moment
When we encountered the true diagnosis
Realizing that our life was different, forever changed
No, it was not a dream, it was reality
New rules for living, and for loving
New friends who also were on a roll
Who truly understood us, the only ones who could, who did

So now I wonder just what is in store
Will the future be as warped as the present?
In the opera, *The Death of Klinghoffer*, he threw away
His wheelchair and crutches
After he passed away

Is that the ultimate salvation?
In other words, is my Soul also a paraplegic?
No, my soul remains able to walk

The Soul of paraplegics never dies
So Old Paraplegics Never Die…They Just Walk Away**

Malin Dollinger, M.D. 2017

***With great respect for and inspiration from
General of the Army Douglas MacArthur*

Synapse

Volume 20, Issue 3 - Fall 2017

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

Synapse Editors
John Staehle......................................Senior Editor
Malin Dollinger, M.D......... Medical/Research Editor

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

The SPF is a non-profit 501(c)3.
Tax ID # 04-3594491
Combined Federal Campaign CFC #12554

Send Correspondence & Donations to:
Spastic Paraplegia Foundation
1605 Goularte Place
Fremont, CA 94539-7241

Contact the SPF at:
(877) 773-4483
information@sp-foundation.org
or www.sp-foundation.org

SPF Board of Directors:
Frank Davis, President
Linda Gentner, Vice President
Jean Chambers, RN, Secretary
David Lewis, Treasurer
Members: Corey Braastad, PhD, John Cobb,
Tina Croghan, Laurie LeBlanc, David Ress, PhD,
Ben Robinson, Mark Weber, Esq., Jackie Wellman

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

Disclaimer: The Spastic Paraplegia Foundation does not endorse products, services or manufacturers. Those that are mentioned in Synapse are included for your information. The SPF assumes no liability whatsoever for the use or contents of any product or service mentioned in the newsletter.

Board Business

April 2017 through June 2017

APRIL: Norma Pruitt, Atlanta Conference Coordinator reported everything was progressing a planned. She asked all SPF members to register as soon as possible so she can work with the hotel to plan meals and breaks. John Staehle agreed to work with Mari White to secure Austin Plain of Challenge Air, as the after-dinner speaker for Friday.

On April 2, Frank McKeown, HSP Research Foundation, Australia, Frank Davis, SPF, and Dr. Corey Braastad, Covance Drug Discovery held a conference call to discuss the progress made in Australia's clinical drug trials and the path ahead.

The board decided to build a relationship with Invitae and are in the very early stages of getting started.

MAY: Frank Davis reported that the Foundation's annual report was delivered to the printer early this month; however, he does not yet have a mailing date.

Norma Pruitt reported video recordings of presentations will only made on Saturday. She and Mari White have been unsuccessful, so far, contacting Austin Plain to be Friday's speaker. Norma said she has been bombarded with questions about the AC. She will copy and paste the material from *Synapse* to go onto the website and onto the Facebook page.

JUNE 23, 2017 (in Atlanta): Frank Davis called the meeting to order at 8:17 AM (EDT). Ten board members attended the meeting including three by a telephone conference call. There were two guests. Norma Pruitt, Conference Coordinator; attended partly on behalf of her husband, Greg, who was called home at the last minute by family matters. Also in attendance was SPF's Medical Advisor, John K. Fink, M.D., University of Michigan.

The approved agenda for the meeting covered:

- Fundraising: what has been successful, what hasn't and what are some possible new fundraising initiatives.
- Improved Planning and Scheduling of Annual Conferences and monthly Board of Directors; meetings (start planning earlier, link ACs to another professional meeting and utilize video conferencing).
- Three committees formed: Grant Committee - members David Ress, Jim Sheorn, Tina Croghan, Jackie Wellman, Norma Pruitt; Information Committee - members Tina Croghan, Laurie LeBlanc, John Cobb, Dr. Fink, Jean Chambers; Conference Committee – members Norma Pruitt, Tina Croghan, Kris Brocchini.
- Elections of Officers and Board Members for the 2017-2019 term: President, Frank Davis; Vice President, Linda Gentner, Secretary, Jean Chambers, R.N., Treasurer, David Lewis, and Board Members Corey Braastad, PhD, John Cobb, Tina Croghan, Laurie LeBlanc, Greg Pruitt, David Ress, PhD, Ben Robinson, Mark Weber, Esq., Jackie Wellman.
    - ○ John Staehle chose not to serve another 2-year term at this time, but agreed to continue serving as Editor of *Synapse*.
- Hire an Executive Director at an average cost of $61,800/yr. Greg Pruitt was prepared to present the alternate proposal he and Norma prepared and distributed to the Board prior to the meeting, but due to his absence, this agenda item was tabled until Greg was available.
- Proposal to hire a webmaster to take the burden off Frank Davis. John Cobb volunteered to take on this responsibility. He and Frank will work out the details to make an orderly hand-off of webmaster duties.
- SPF State Ambassadors: Current system is ineffective. Though we have ambassadors for many of the states, many of them essentially do nothing. Jackie Wellman sent an email to all the State Ambassadors of record at the time requesting a reply. Those that did not were "fired" and replaced. There were discussions about better preparing ambassadors to perform their duties, organizing ambassadors into regions, conducting webinars for training and video conferences between ambassadors for Q&A, drop "State" from job title, to name a few.
- Review Current System for Funding Researchers: Frank Davis ask Mark Weber if he could be assisted by someone with this task. He replied that it would require an enormous amount of time and effort to give the necessary information to anyone who offered to assist. It is easier for him to do it himself. Mark also reported the Requests for Proposals have been sent to the HSP and PLS research community.
- Research Australia: Frank Davis read a letter he just received from Frank McKeown, HSP Research Foundation, Australia, updating the status of their clinical trials.
- Dr. Fink reported he has submitted a grant application to NIH that involves one form of HSP that has biomarkers. A similar application was previously denied, but this application has changes that Dr. Fink is confident will result in an acceptance this time. He expects a decision yet this Summer.

2017 Annual Conference Recap

Atlanta, Georgia
June 23-25, 2017

Facts:

- The total number of registered participants was 140, of which 8 were under the age of 18.
- Dinner on Friday evening was served to 140 people.
- Saturday's General Session was attended by 145.
- Twelve people took the Saturday excursion to the Atlanta Zoo.
- Forty people took the Sunday excursion to the Atlanta Aquarium.

CONFERENCE SPONSORS

Our Heartfelt Thanks to This Year's Sponsors

KRIS BROCCHINI

Kris Brocchini

Kris was diagnosed with PLS at UCSF in June 2003. After looking up PLS on the internet, she found the SPF and made plans to attend the Lexington, MA, Conference and Team*Walk* in late September 2003 where she and her two daughters and a brand-new granddaughter were welcomed with open arms. Questions were answered, stories were compared and a whole new, frightening world to think about was discovered. Seeing all those people using wheelchairs and other mechanical devices was frightening indeed. But informative. She thinks everyone needs to attend that first conference.

Kris was invited to join the SPF Board of Directors after the 2005 Columbus, OH, Conference and her first board meeting was in March 2007 in Nashville, TN. She served on the Board until 2016.

Her personal support of the SPF Annual Conference for the past 8 Conferences is the result of remembering how she felt when she found the SPF community.

MNG LABORATORIES
Neurogenetic Answers™

MNG Laboratories™ provides expert diagnostics through clinical services, complex biochemical testing and sequencing. Our Next Generation Sequencing panels are the most cost effective and comprehensive available, particularly for cellular energetics defects, muscular dystrophies, epilepsy, cardiomyopathy and intellectual disabilities.

INVITAE

Invitae, a genetic information company, is bringing genetic testing into mainstream medical practice by providing high-quality, affordable genetic testing.

CHRISTOPHER & DANA REEVE FOUNDATION
PARALYSIS RESOURCE CENTER®

The Christopher & Dana Reeve Foundation provides a Paralysis Resource Center that educates and empowers those with paralysis with a host of FREE programs that include information services, peer mentoring, community education program, grants for non-profits that serve individuals with paralysis and a lending library. All of our services can be accessed through christopherreeve.org or by calling 800-539-7309.

SAOL THERAPEUTICS

Each patient with a rare disease is a patient who needs to be heard and who deserves to be treated. And that is why Saol Therapeutics exists. We bring therapies for these serious and often life-threatening conditions to market and to the life of the patient who desperately needs it.

Addressing the needs of patients with rare diseases and under-served neurological conditions are our passion and focus at Saol. Supporting patients with high un-met needs by offering promising treatment options is what drives us.

INHERITANCE PATTERNS

Presented by Corey Braastad, PhD

Vice President and General Manager of Genomics, Covance Drug Discovery

Dr. Braastad opened his presentation stating this year's subject will be about how HSP is inherited from your parents rather than about genetics, which had been his subject in recent years.

He began by stating that virtually all diseases have a genetic component. HSP falls into a category called Mendelian Diseases which are the rarest of all medical conditions and have a large genetic component with little impact from environment and exposures. A genetic change in <u>one</u> gene <u>causes</u> disease. The inheritance patterns of Mendelian Diseases are well-known and easily predicted: autosomal dominant; autosomal recessive; X-linked dominant; and X-linked recessive.

Dr. Braastad explained the all of us have 23 pairs of chromosomes. Each parent provides 23 chromosomes and when they combine, the 23 pairs define every physical characteristic about us. Twenty-two of the pairs are autosomal which means they are not linked to a person's sex and the 23rd pair defines their sex, an X chromosome from both parents, XX, is female and an X from the mother and a Y from the father, XY, is male. Dominant means only one copy of a gene is necessary to cause phenotype while recessive requires two.

The Autosomal Recessive inheritance pattern requires both copies of a gene pair to contain the pathogenic mutation. Individuals with one mutated gene are healthy carriers and are not affected by the condition. There is a 24% recurrence risk for offspring when both parents are carriers and males and females are equally affected. HSP examples are SPG5A, SPG7 (Paraplegin), SPG11 (Spatacsin), SPG15 and SPG35 (FA2H). One surprising fact presented by Dr. Braastad was, "Everyone is a carrier autosomal recessive conditions."

The Autosomal Dominant pattern requires one mutated gene in the gene pair to contain a polymorphism. All individuals with the mutation are affected by or predisposed to develop the condition. Mutations in one or both genes can cause the disease. There is a 50% risk of recurrence for all offspring and males and females are equally affected. HSP examples are SPG4 (Spastin), SPG3A (Atlastin). SPG6 (NIPA1) and SPG31 (REEP1).

The X-Linked Recessive pattern is the most common of the X-linked patterns while the X-linked dominant pattern is very rare. It is a subtype of autosomal recessive pattern in which the mutated gene is on the X-chromosome. The disorder is expressed in males because they only have one X-chromosome while females have a spare X-chromosome to compensate for the mutated gene. HSP examples are SPG1, SPG2, SPG16, SPG22 and SPG34.

Dr. Braastad's complete presentation may be viewed on the SPF website at https://sp-foundationorg.presencehost.net/what_we_do/annual-conference-recap.html.

● ● ● ● ● ● ● ● ● ● ● ● ● ● ●

HSP AND PLS UPDATE

Transforming Remarkable Genetic Advances into A Path Toward Treatment

John K. Fink, M.D., University of Michigan Department of Neurology
<u>jkfink@umich.edu</u>

Dr. Fink opened his presentation by stating he is not an expert on HSP - the people in the audience with HSP are the experts. He then said that in his opinion it is unacceptable to talk just about research advances without talking about how to use those advances to make the transition to therapy.

He then presented various paths to therapeutic development, the first being the mechanistic approach. A limited number of HSP autopsies, 15 or so, and clinical observations of the symptoms expressed by people with different variations of HSP, showed a correlation between pathologic nerve degeneration and those clinical features. From that, a hypothesis was

Continued on next page

drawn that upper motor neurons and dorsal columns (the path of long nerves from the brain to the lower spine) have selective neuronal vulnerability. Then began the process to postulate the molecular mechanisms that affect those long nerves' vulnerability.

Gene discovery is exploding and "continues to explode." Dr. Fink said he made a list of the 60 known HSP genes 2 weeks prior to the conference and it is already out of date. He also noted that there are as many as 8 PLS genes that have been identified. These genes, when disturbed, cause the degeneration of neurons in a particular way. This fact allows researchers to develop in vitro and in vivo models that lead toward the establishment of treatment. The mechanistic approach has been successful in treatment development, but it is not the only approach.

Serendipitous, or accidental, drug discovery is the discovery of a treatment originally developed for a specific condition that has a side effect that makes it an effective treatment for a totally different condition. He gave the example of a drug used in the early 1950's as an antibiotic for tuberculosis that also produced a feeling of euphoria was given to subjects suffering from depression. Began the revolution of treatment developments for depression.

Another approach is empiric trial of re-purposing treatments used for a disorder with similar symptoms or similar mechanisms. Examples given are levodopa in dopa-responsive dystonia and anticonvulsants for migraine and bipolar disease.

Dr. Fink believes we need to move towards an approach that uses more "crowd sourced clinical trials" especially for rare diseases like PLS that have such a small population that it is difficult to conduct meaningful clinical trials. Worldwide participation by physicians, clinicians and investigators, each using standardized collection, evaluation and analysis methods on a small number of patients and sending their results in a standardized format to centralized data collection. This approach would yield a larger population of results that would have a much higher probability of attracting the funds needed to develop treatments for rare diseases like PLS.

Which approach is best? We should use all of them.

Dr. Fink then began a discussion on the biases that investigators bring to research. Historical generalizations about HSP and PLS mechanisms become accepted dogma. This dogma can bias research hypotheses, but some of this dogma is false. An example is the dogma that PLS, unlike HSP, is a non-genetic condition based on observations that individuals with PLS very rarely have similarly affected relatives. We now know that is not true. Gene mutations are found in some subjects with PLS. PLS is a "syndrome," a common group of symptoms and signs, and not one single disorder. There are multiple forms of PLS.

After discussing whole exome sequencing versus whole genome sequencing as methods to identify gene mutations and addressing HSP's central dogma that progressive lower extremity weakness is due to the progressive degeneration of long axons, Dr. Fink identified four potential treatment targets:

1. Treat the disease process. This may vary with each HSP/PLS type and sometimes within the same HSP type.
2. Treat the degenerative process or its consequences. The concept of neurodegeneration is the sequence of cell disassembly, initiated by gene mutations followed by a regulated disassembly process. Treatment to stop the neurodegenerative process is one approach. Another is to facilitate the damaged nerve transmission or nerve growth even without treating the underlying abnormality.
3. Treat the symptoms. To accomplish this, we need a better understanding of the neurochemistry of spasticity. We also need to identify what causes weakness in HSP and PLS.
4. Understand and facilitate neuro-compensation (neuroplasticity). Need to understand the basis for "apparent functional plateau" or "slower rate of worsening." Does the degenerative process slow down or stop? Is the apparent slowing of the rate of functional worsening due to compensatory neuro-reorganization? If we knew what caused the disease progression to slow down, could this process be induced to stop the disorder in its earliest stages?

Dr. Fink closed his presentation with a “to do” list:

- Better animal models
- Natural history studies
- Crowd-sourced repurposing trials using standardized evaluation and centralized data
- Rehabilitation strategies
- Biomarkers
- Clinically available biochemical assays of HSP protein function.

Dr. Finks’s complete presentation may be viewed on the SPF website at https://sp-foundationorg.presencehost.net/what_we_do/annual-conference-recap.html.

* * *

CREATE

Jeffrey Statland, M.D., Assistant Professor of Neurology, University of Kansas Medical Center

CReATe, the acronym for Clinical Research in ALS and Related Disorders for Therapeutic Development, is a member of the Rare Diseases Clinical Research Networks (RDCRN) and is dedicated to studying motor neuron diseases including HSP and PLS. It is a good example of the government funding, through the National Institutes of Health, of areas of underfunded research like rare diseases.

It is estimated there are around 7,000 rare diseases. Rare diseases are complex and often have inadequate or no treatment. Patients are frequently misdiagnosed or are undiagnosed. It’s hard to study rare disease without collaborations, which can be costly.

The RDCRN’s mission is to advance medical research on rare diseases by providing support for clinical studies and facilitating collaboration, study enrollment and data sharing. Its mission also is to gain a better understanding of the natural history, to improve diagnosis and to develop therapies. The RDCRN does this by providing money for collaborative studies and the infrastructure to support the studies with coordinators, evaluators and investigators. It provides central data coordination by making sure data is collected in the same way and is complete. It also provides central ethical oversight. Collectively, the RDCRN is studying more than 200 rare diseases at more than 400 active sites in more than 20 countries with 40,000+ people enrolled in clinical studies. There are currently 22 separate consortiums supported by the RDCRN, one of which is CReATe.

CReATe is run from the University of Miami by Michael Benatar, CReATe founder and Principle Investigator, Joanne Wuu, Co-Investigator, a project and data management team and a biorepository. It is a consortium of physicians and scientists studying ALS (Amyotrophic Lateral Sclerosis), ALS-FTD (frontotemporal dementia), PLS (Primary Lateral Sclerosis), HSP (Hereditary Spastic Paraplegia) and PMA (Progressive Muscular Atrophy). The current network of clinical sites includes 2 coordinating sites for data in Florida, 10 active sites and 5 pending sites (2 of which are outside the U.S.). There also are 14 partner organizations, one of which is the Spastic Paraplegia Foundation. CReATe’s overarching goal is to better understand the natural history, relationship of genotype to the clinical expression, develop biomarkers and tools to help facilitate drug discovery.

There are 6 active CReATe Consortium Protocols, of which the largest is 8001 – Phenotype, Genotype, Biomarkers (PGB). The PGB study is a longitudinal study with an initial onsite baseline visit followed by onsite visits at months 6, 12, 18 and 24. After that there will be annual phone calls as long as the subject chooses to continue in the study. Inclusion criteria for affected individuals include a clinical diagnosis or strong clinical suspicion for a diagnosis of ALS or a related disorder and the subject’s ability and willingness to comply with study procedures. Family members of genetically affected individuals may also enter a limited study.

Expected outcomes of the PGB study are:

- Biomarkers: blood, serum, urine, CSF;
- Genetic testing;
- History: detailed symptom history, family pedigree;
- Detailed exam that assesses both upper and lower motor neuron involvement and respiratory function;
- Functional assessments using the ALS Functional Rating Scale – Revised (ALSFRS-R) and the Spastic Paraplegia Rating Scale (SPRS).

Continued on next page

ALSFRS-R rates from 4, no involvement, to 0, significant involvement, 12 domains which represent basic daily activities. SPRS rates from 0, no involvement, to 4, significant involvement, 13 functional domains.

Protocol 8003 is A Patient-Centric Motor Neuron Disease Activities of Daily Living Scale (PADL-ALS). The goals of this study are:

- To determine the feasibility, ease of use and rates of ALS and related disease progression using a patient-reported activities of daily living scale;
- To determine the patient-reported clinical determinates of disease progression;
- To compare previously reported disease progression models.

Participants need to be enrolled in CReATe Connect, an RDCRN contact registry. They can self-report a diagnosis of motor neuron disease (ALS, ALS-FTD, PLS, PMA, HSP) and need to be able to fill out the survey in English. An introductory letter will be sent to all eligible Connect Registry participants followed by an initial survey that includes the PADL-ALS. Twenty-seven percent of participants in the CReATe Connect Registry have HSP or PLS.

CReATe is still enrolling participants for this study. If you are interested in participating, contact:

Sumaira Hussain
CReATe Project Manager
Phone: 844-837-1031
Email: ProjectCReATe@miami.edu

Dr. Statland's complete presentation may be viewed on the SPF website at https://sp-foundationorg.presencehost.net/what_we_do/annual-conference-recap.html.

Primary Lateral Sclerosis (PLS) and Hereditary Spastic Paraplegia (HSP) Research Challenges

Christina Fournier, M.D., MSc, Emory University School of Medicine

Dr. Fournier opened her presentation describing two significant problems developing treatments for rare diseases like PLS and HSP while research continues to search for cures. The first problem is how to identify treatments that slow progression when the diseases themselves progress slowly and when the diseases are variable. The second problem is getting enough participants to conduct meaningful clinical studies on rare diseases like PLS and HSP.

Issues related to the first problem that must be addressed are having reproducible, responsive measures of disease status and having biomarkers that identify effective treatments before clinical changes are obvious. ALS progression is measured by a 48-point self-reported scale, the Revised ALS Functional Rating Scale (ALSFRS-R), that shows ALS declines by an average of 1 point per month. If ALSFRS-R is used to measure disease progression for PLS, the average decline in the rating is 0.2 points per month meaning PLS trials would have to be much longer and bigger to detect a treatment effect. PLS scores using the ALSFRS-R over time seem to go up and down making its use less reliable. The rarity of PLS makes bigger trials extremely difficult to conduct. A PLS-specific rating scale or objective measures (e.g., biomarkers) would facilitate more effective trials, but neither currently exits.

HSP has similar problems even though there is an HSP-specific functional rating scale, the Spastic Paraplegia Rating Scale (SPRS). The ratings are based on a subjective evaluation and subsequent scale grading performed by a neurologist. Thirteen functional characteristics are graded, each scored from 0, no involvement, to 4, significant involvement or unable to perform. A total score of 0 is normal (no involvement of the disease) and a 52 indicates the highest level of disability. The average increase in the rating (decline in abilities) is 0.08 per month or 1 point per year. HSP's slow progression requires long clinical studies to determine the effectiveness of treatments.

In summary, the outcome scales for PLS and HSP are easy to administer, reproducible and clinically relevant but they are not responsive to short term changes in status the scores bounce around.

Solutions to problem one are (1) develop more responsive disease specific outcome measures and (2) identify the biomarkers of disease progression. Non-interventional studies – the study of natural histories, biomarkers and clinical scales – are an important step toward developing these solutions. They allow for the development of better outcome measures which can accelerate drug development by improving the ability to detect treatment response and the development of biomarkers needed to determine if drugs hit their target by allowing faster measurement of treatment response.

You can find information on research studies by going to NIH's site, ClinicalTrials.gov, and searching for Hereditary Spastic Paraplegia or Primary Lateral Sclerosis studies (PLS patients should also check for ALS studies). You can also check with your neurologist and your local academic institution.

Problem two is the rarity of PLS and HSP makes conducting clinical trials difficult. The prevalence of PLS in the United States is estimated between 1,000 and 2,000 and HSP is estimated between 10,000 and 20,000. Trials for specific HSP gene mutations further reduce the pool of patients eligible to participate in that trial. Even though a single site may have all the resources needed to conduct a trial, it may find it difficult to find enough participants to make the outcomes of the study statistically accurate. The solution is to establish multi-center, collaborative research efforts. The Northeast ALS Consortium, NEALS, is an example of such a collaborative effort. Patients with upper motor neuron disease are seen at one of the consortium's 20 sites, 233 in the past year. The study has provided detailed clinical information, common symptoms, level of disability and test results. A new longitudinal registry is being analyzed now with 281 patients.

A recent Canadian observational study of 526 HSP patients and a German observational study of 608 HSP patients provided detailed information about clinics, complications, level of disability and genetic testing.

In summary, overcoming the challenges of rare disease research may be accomplished by:

- Conducting observational, non-treatment trials which will allow the development of better outcome measures, allow for biomarker development and establish the feasibility and patient base for future clinical trials; and
- Conducting multi-center collaborative research studies with motivated patients.

Dr. Fournier's complete presentation may be viewed on the SPF website at https://sp-foundationorg.presencehost.net/what_we_do/annual-conference-recap.html.

ADVANCES IN PLS RESEARCH

Sabrina Paganoni, MD, PhD, Assistant Professor, Harvard Medical School, Massachusetts General Hospital, Neurological Clinical Research Institute

Dr. Paganoni introduced herself to the conference attendees and explained she was the second recipient of the Virginia Freer-Sweeney Fellowship granted through the Spastic Paraplegia Foundation. She is in the second year of the 2-year fellowship. The goals of her fellowship are:

- Improve our knowledge of PLS
- Discover PLS disease mechanisms
- Promote PLS clinical trials
- Create a community of PLS researchers.

PLS is rare…an estimate of 1,000 cases in the United States. Previous PLS studies would involve only 10 to 50 patients. NEALS, the Northeast ALS Consortium, has 120 sites throughout the U.S. to register ALS patients. They also have registered 275 PLS patients and therefore have a large data base of PLS natural histories.

Since Dr. Paganoni's fellowship was limited to 2 years, she concentrated on a 15-year retrospective review of nearly 300 NEALS files on PLS patients. Data collection is complete and she now will spend the remainder of her fellowship analyzing that data. Collaborative NEALS efforts include a prospective collection of PLS data by Mary Kay Floeter, MD, PhD, using the Revised ALS Functional Rating System (ALSFRS-R) on 80 PLS patients that she has been following annually since 2000. The next step is to develop a PLS-specific rating system, something currently being addressed by Hiroshi Mitsumoto, MD (partially funded by the Spastic Paraplegia Foundation).

Dr. Paganoni's second fellowship objective is to discover PLS disease mechanisms. In a normal brain, the upper motor neurons (UMNs) connect to the long axons that carry the brain's signals to the lower limbs. In cases of PLS, the UMNs are affected and do not function properly leading to difficulty walking. To

Continued on next page

determine what is happening, a scanner that combines Magnetic Resonance Imaging (MRI) with Positron Emission Tomography (PET) is used to look at the structure of the UMN and the functional integrity of the axons. PET scans require a radioactive tracer to be injected to show affected areas of the brain. The results of these scans show an increase inflammation of the UMN area of the PLS brain. Further analysis shows a relation between the amount of inflammation and the loss of structural integrity and nerve cells. The challenge now is to find ways to mitigate the inflammation.

The third goal of her fellowship is to promote PLS clinical trials. Small trials to determine proof of mechanism will be used to determine whether or not a drug affects the inflammation of the upper motor neurons. Such trials are not being done for PLS but are being started for ALS. As we learn more about PLS, we will be able to initiate similar trials for PLS. There is one drug trial in process for PLS. The drug Ampyra is given to patients with MS to improve gait. The PLS trial is to determine if a similar result is possible with PLS patients.

The more we learn about PLS and the more clinical trials that are conducted, the more interest there will be in doing PLS research. It is important to create a community of PLS researchers in order to continue the development of new treatments for the disease.

Dr. Paganoni's complete presentation may be viewed on the SPF website at https://sp-foundationorg.presencehost.net/what_we_do/annual-conference-recap.html.

Pumped Up Again

My Baclofen pump, to help with my walking
So people wouldn't stare, no more of that gawking
Eight years ago, the first pump went in
But along about now, the pump's a has-been

So another one replaced, old battery almost gone
Arrived at surgery at the crack of dawn
Awake during the time the new pump implanted
Rather interesting and painless, old one explanted

So souvenir now, that gadget that was inside
New one now working, I'm ready to confide
That my operative fears are now no big deal
My brand-new pump is all mine for real

Medtronic rep was there for me too
Company sure the pump said, "I do"
So now I'm back home, large dressing in place
I'm now getting ready my work schedule to face

So thanks to the surgeon and all the kind staff
For getting me through, I'm surprised I could laugh
At how easy it was, how satisfied I could be
I reassure all of you, it was easy, you'll see!

On behalf of Cynthia Wagner
by her father, Malin Dollinger, 2017

The CFC or Combined Federal Campaign is a fundraising campaign the Federal Government offers its employees to participate with each year. It begins Sept 1st and goes through Dec 15th. Federal employees are allowed to pick from over 200 registered non-profits to contribute to. Many CFC fairs will be held at Federal facilities throughout the campaign. This allows employees to learn about the non-profits and make their selections.

Please let friends and family members who are Federal employees know they can choose the Spastic Paraplegia Foundation to donate to. The SPF CFC number is **12554**.

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

If you have any questions or suggestions, please contact Jim Sheorn at jimsheorn@gmail.com or 615-479-7369.

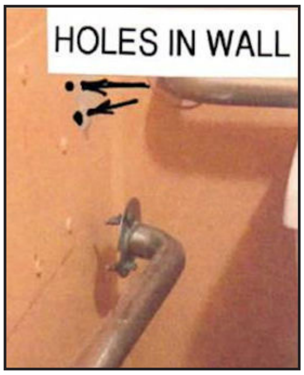
Please help us generate more financial resources for research.

HANDICAP AIDS: HOME, OFFICE, CAR, AND GARAGE

by Malin Dollinger, M.D., SPG4

This article mostly pertains to those who are able to stand, either by themselves or by "holding on" to furniture or a grab bar. There are two kinds of handicap aids: 1) commercially available handicap aids, and 2) handicap aids that you or a craftsman can create yourselves. I will not discuss commercial aids for those who *cannot* stand, such as lift devices, e.g. Hoyer lifts, and various specialized items such as transfer boards, methods of getting in and out of bathtubs, and complex aids for daily living.

COMMERCIALLY AVAILABLE HANDICAP AIDS:



There are many items specifically manufactured to assist handicap folks. Universal handicap aids are those that can be used in different kinds of places, such as handicap grab bars. You can use them in various rooms in your home, the bedroom, bathrooms, dressing areas, the office, the kitchen, on stairways and in the garage. These have mounting plates at each end, usually with three holes for mounting screws. Many are mounted vertically, especially in places you need to stand, while holding onto something with one hand. These are placed on cabinets, book shelves, and in your garage or shop, wherever you need to "hold on" while standing. Some are mounted horizontally, particularly near the bathroom sink and on both sides of the toilet, to hold onto when getting up and sitting down. Grab bars come in different lengths, including 12", 18", 24", and longer like the ADA-required lengths of 36" behind a public toilet and 48" on the side of a public toilet. The wall stud spacing is every 16 inches, so often I placed other sizes of grab bars, e.g. 18 inches, at a slight angle so both ends are over studs. Commercial grab bars are white or metallic, but other colors are available by special order. They are sold in most large hardware stores and drugstores, as well as medical supply houses, the same outlets where we buy walking aids and handicap items in general. The most important rule is that they are screwed in place into either solid wood studs behind the wall or by using some type of effective and safe mounting system, such as a Molly or similar anchor, that "grabs the wall" from both sides. If they are simply screwed into wallboard, not the studs, and/or the screws are too short, they may come off the wall when you grab them.

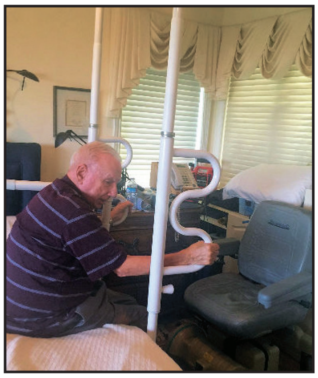
Remember not to use a *towel bar* to grab onto. As installed by contractors or workers, they are not secure enough to hold your weight, and the size of the bar itself is too small for a secure handhold. The photo shows the grab bars around a toilet, including one on the back wall, for men to hold when standing. Note the chrome metal *towel bar*, not be used to hold onto. Also note the "slip-on" toilet paper holder and the nearby spare roll *(you really don't want to have to get up at that moment or to call for help on the wall phone)*. Yes, I do have an emergency *("I've fallen")* pendant, but I always have my cell phone with me wherever I go.

Paraplegics should avoid doing anything that might injure a shoulder. With severe leg weakness, you use your arms/hands many times throughout the day to pull yourself up and lower yourself down, change positions, get on/off your walker or scooter, and "balance yourself" when moving from place to place. If you injure one shoulder, very often when you have to depend only on the other "good" arm/shoulder for these functions, the sudden, intense, and repeated strain on that "good" shoulder may injure it as well. The result is two bad shoulders, as well as two bad legs, and effectively you have become a quadriplegic. I know this story well; I grabbed a towel bar in a hotel – that was the only thing available to grab — and my hand slipped off, but not before injuring one shoulder. Later the other one was injured from all the extra work it had to do while resting the first shoulder. This is why you should have grab bars installed in your shower and next to your toilet and also on or near the edge of your

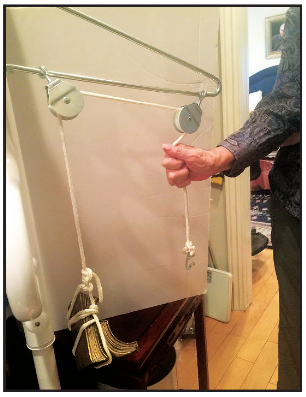
Continued on next page

bed. The important principle is to hold on, support your weight as necessary, *using both arms and hands*, whenever you transfer positions. When transferring, I often push up with one hand using the armrest of my scooter while pulling up using a grab bar with the other hand.

There are commercially available, specialized grab bars for specific places and uses. In the bedroom photo you can see the black horizontal grab bar next to the bed. When I travel, I take a portable grab bar, in the shape of a large "L," which comes disassembled flat in a carrying case. One side slides between the hotel bed's mattress and the box spring and the other projects upwards, to hold onto when getting in and out of the bed. This type of grab bar can easily be used "permanently" at home, and these "for home" grab bars have a very long portion that slides under the mattress – two or three feet long – so they are stable, safe, and will not move around when you pull on them. Some commercial ones can hinge out of the way, like the ones in hospital beds. You can strap a plastic cup onto a bed grab bar, to hold a bottle of water for nighttime use, and also, for men, a hook to "park" a urinal to use in the middle of the night to save a trip to the bathroom – a complicated event for us. Just be careful which one you reach for in the dark! Handicap aids companies make many different styles of grab bars and similar things to hold onto.



One such clever grab bar device is called a Security Pole and Curve, which runs floor to ceiling, and can be mounted using spring-loaded spindles, like a car jack, or with long screws into the floor and ceiling. Note the curved hand grabs halfway up, the location of which is suitable for most everyone. I use them to help me transfer from my bed to my scooter or from one scooter to another. One that I installed where I get dressed has an additional use. My homemade "pants up" device uses a commercial neck-stretching assembly, which comes with a pulley up top (mounted over a door). One rope end has a weight and the other a fabric strap assembly that holds your head/chin to be stretched by the weight. My adaptation has the pulley mounted at eye-level on the curved middle part of the floor-to-ceiling grab bar. It has a heavy weight on one end of the rope, and the other end has a large hook *(I used a large picture-hanging hook)*. To pull my pants up in the morning, while sitting in my scooter next to this homemade device, I put both legs into the pants, as far as I can while sitting, then attach the hook to the first belt strap on one side of the pants. Then while holding onto the grab bar, I stand up. The weight/pulley grabs the pants and pulls them up. I then sit down while I pull up the other side of the pants, and then I sit down with the pants up all the way. This gadget is very efficient and practical.



The same idea, using a weight attached to a pants belt loop to move your pants can be used in the evening, when you need to pull them down. Mine get caught on the front of my scooter seat. So I took another large picture hanger hook, attached a short chain (the hardware store sells keychain extension chains), and a small weight at the other end of the chain. While sitting in your scooter at bedtime, place the hook, with weight attached, inside a belt hook at the front of your pants, then stand up. After the weight pulls your pants down, take the hook out of your pants on the floor and put it in a handy place for tomorrow night.

Consider obtaining sets of "offset" door hinges, that add 2" to the door width. This may make the difference in being able to get a scooter or wheelchair through a doorway "the easy way," rather than having to do a lot of carpentry to rebuild or widen the doorway.

I assume you can get on and off your toilet easily and safely. Hopefully the seat is high enough. Some commercial toilet seats, especially the low-profile ones, are "way down there," which makes getting onto the seat an aiming-target experience, and getting up an awkward and strenuous experience. You can buy a seat riser that adds up to three or four inches to the height of the "sitting surface." Some risers are built into the seat itself and others fit between the toilet rim and the seat. There are even some that are installed between the floor and the base of the toilet *(it's recommended you have a plumber install this one)*. In the scooter articles, I described the use of your scooter arm as an important aid in pulling yourself up, something else to hold onto. This is especially important and useful in bathrooms away from your home, which usually have only a single wall grab bar. Very considerate commercial places have bathrooms with an extra arm rest/grab bar that is attached to the wall on the other side of the toilet, and folds down when needed. That way you can rest each arm on its own armrest.

● ● ● ● ● ● ● ● ● ● ● ● ● ● ●

THE KITCHEN: Here are a few practical tips and ideas. If you use a microwave oven, they are usually mounted "high-up," like over the stove. It's difficult for us to use them, since loading is difficult "up there." Getting the hot cooked food out is even more difficult, since you usually need two hands and the food dish must be carefully held not to spill or burn your hands. We don't have enough hands to hold on while standing up, and also remove the food. It took several years for me to realize that I could buy a small inexpensive counter-top microwave oven that is easy to reach and thus safe to use. There are food and utensil arrangements that also are useful, functional, and safe, too. Put the foods, utensils, pots & pans and special devices you frequently use in the lower kitchen drawers and cabinets. Let your partner get the rare stuff down off the high shelves and cabinets on those infrequent occasions when need them.

Find devices that are easy for you to use and do not require standing or that are difficult or awkward to use if you also need to hold onto something while standing. Electric can openers, popcorn poppers are examples of those that are easy. Some foods can be purchased in "almost ready to use" mode. Older folks remember shaking a pan of popcorn on the stove for 5 or 10 minutes and cooking bacon in a frying pan. I buy almost-cooked bacon that is ready in one minute in the microwave. You can also buy microwave popcorn that takes about 4 minutes to pop in your counter top microwave. Just make sure it has enough power to do popcorn (750 watts and higher). The higher the power, the quicker it will pop - or burn if you overcook it. Although I like to cook, a lot of frozen foods make life easier. Small electric appliances like stirrers for salad dressings and a good electric can opener are easy to operate. Is your kitchen counter too high to use comfortably? Get a small short table, perhaps on wheels, that you can use for food preparation. I had a "cooking shelf" made, at a height just above my knees, which is a lot easier to use than reaching up to the standard counter top.

● ● ● ● ● ● ● ● ● ● ● ● ● ● ●

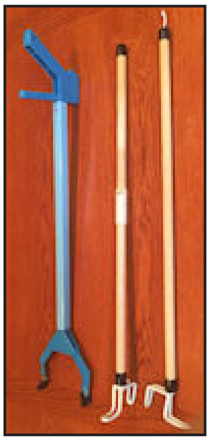
YOUR CAR OR VAN: You may have a handicapped-accessible car or van, or a specific car model that works for you. Examples would be a 4-door car, a large trunk, that would hold a wheelchair or small scooter – perhaps also with a small lift-crane to move it in and out of the trunk – or a handicap van with the "middle seat" removed to allow space for your scooter, with a ramp that suits you and your parking situation.

There is a small metal handle device, the Car Door Grab Bar, sold at a variety of stores and online, that fits into the door latch of most cars. You can use it to assist you when entering or exiting your car on foot and need some extra support while doing so *[Editor's Note: refer to Tina's Tips in the Fall 2016 issue of Synapse for additional information and pictures]*. It is also very helpful to install a few screen-door pulls vertically in the far-back driver's-side corner of the inside of your van, so you can use a Bungee cord between the door pulls to hold vertical things, like an umbrella, grabber, utility stick or guitar from falling onto the floor. I screwed down a small inexpensive utility rug, about 3 x 4 feet, onto the floor of my van, where the scooter sits. This prevents harm to the carpet on your van's floor. The extra rug will accept all the abrasion and damage, rather than the relatively thin and vulnerable car flooring as delivered. You need to screw the rug down. Otherwise the scooter's wheel action will push the rug in the opposite direction every time you drive the scooter across it *(according to Newton)*. If you drive with hand controls, most states require that you have a spinner knob on your steering wheel *(unfortunately, if you don't use hand controls, spinner knobs are illegal in those same states)*. I have two spinner knobs on the steering wheel, at 2:00 and 10:00, very handy for turns in both directions. I also found that spherical spinner knobs are more comfortable than the "flat" palm spinner knobs.

There are a variety of "grabbers," available in handicap/surgical stores as well as from many online stores, with different styles of grab-ends and lengths. I have an assortment of different length grabbers, so I can select a proper length to reach a book or object on a higher shelf.

Continued on next page

A folding grabber is very useful to carry with you, small enough when folded to carry in a wheelchair or scooter pocket. There is a new model that does not break apart when an inside control string breaks. This one, model Able 2-Reacher, with folding clamp, made by Bluestone, is reliable, all-metal, and fits nicely into my scooter pocket. There is also a very useful device consisting of a wood stick a couple of feet long, with a metal hook device on one end for moving things or picking things up, and a small metal hook on the other end for mounting it on a fixed mounting hook. I have a “wood stick” in most rooms, hanging it on a hook on the wall, to be easily available. I’ve placed “room-labels” on many of them, so after use, they get replaced where they came from. I pick up things I’ve dropped, and use them to pick up important things from the floor, like shoes, dirty clothes, water bottles, and anything I can get the metal hook around. I can also move things forward on shelves and elsewhere, where it’s too far away or awkward for me to extend my hand; they are simply a useful extension of my hands. It also grabs the top of my socks to help put them on. One model for travel comes apart in the middle for packing.



One handy idea is to buy small light-weight carts on wheels, available at office supply stores like Staples and Office Depot. Likely there are office, hobby, reading or utility items that you’d like to have available in various rooms. I have a couple of these carts, about a foot wide, that I use to move “my stuff” around, wherever it is needed.

* * *

PHYSICAL THERAPY: This is a reminder of “gadgets and devices,” but not a description of physical therapy. Large devices include walking, rowing, exercise and elliptical machines, as suggested and advised by your physical therapist. Small items include large rubber straps *(e.g., Therabands)*, cold and hot compresses, various hand and foot exercise devices and specialized orthopedic devices. Your physical therapist will also remind you that you weigh a lot less while in water and your physical therapy exercises will be more beneficial and easier if you’re standing in a pool. Better to have someone with you while in the pool, for safety.

There are all kinds of elaborate exercise machines, toilets with water sprays, “safe” bathtubs that easy to get in and out of, fancy and complex automobile devices and other handicap aids not discussed here. Various companies, like Great Stuff, make a wide variety of scooter accessories, such as arm rest bags, covers, ramps, canopies, and cup, cane, and crutch holders.

Regarding convenience in obtaining food, I have found it very useful to have a very small refrigerator in my office or work room, mainly for soft drinks and snacks. Even with a scooter, and especially “on foot” with a walker, going to a different room just to get a can of Coke or a bottle of water is an extra task.

* * *

OTHER HANDICAP AIDS THAT YOU, A CRAFTSMAN OR CONTRACTOR CAN CREATE:

I constructed my own “headboard” bed rail, that goes across the head of the bed. It’s made of PVC pipe, steel pipe, two metal flanges, and two small one-foot square pieces of plywood or pressboard. This was designed to provide a movable rail that I could hold onto with both hands, that goes across the bed to position myself in bed, and to assist in rolling over. One end comes apart, to move out of the way, to get in and out of bed. Commercial cross-bed rails fasten to the headboard or hang down from the ceiling, both of which I did not want nor the expense. The photo shows the installed finished product. The components cost less than $40, and it will take an hour or two to build. Mine is an essential part of my life, enabling me to turn over and move around in bed. If you wish a list of components and instructions for assembly, email me at malind@cox.net.

Another device to assist turning and changing bed position, in particular with adjacent twin beds, can be created by mounting a grab bar in the middle of a

square plywood or pressboard piece. This assembly is then inserted between the two twin beds, the wood base above the box spring and below the mattress. To use, you reach down between the two mattresses, and by holding onto the grab bar, you can easily change your position in bed, in particular to get close to your bedmate. My "turning bar" is placed next to the lower plate of the "socket" end of the overhead bedrail, between the twin beds.

I have a sign next to my doorbell so visitors will not think no one is home when it takes a long time for me to get to the door. You might set your phone to ring a few extra times before rolling to the answering machine.

* * *

Feet and Shoes:

I had built a small wood wedge about a foot wide in size, which sits on the floor. I use this every morning, sitting in my scooter, to put on my shoes, much easier when they are at an upward angle rather than flat on the floor. This is better than the small commercial ones, which are the size of your foot, which move around and are at the wrong angle. Speaking of shoes, I had my mail-order handicap shoes (Propet) rebuilt by my local shoemaker to greatly improve and simplify the task of putting them on. They have no laces or buttons, but simply a large leather flap that goes across the top of my foot and fastens with Velcro. Easy to put on and take off. Also, the "tongue" of the shoe, as manufactured, was too short to allow easy foot access (the acronym "T.G.I.F." also means "Toes Go In First!"). I had a shoemaker cut the open edge of the tongue another two inches forward, so now the tongue is much longer and lifts out of the shoe to give me plenty of room to easily put my toes inside the shoe without the kind of foot contortions I'd done during my pre-disability life. To keep the top strap and the tongue strap out of the way when inserting my foot, I put Velcro strips at the ends of both of these and at corresponding places on the side and front of the shoe to temporarily "park" these straps out of the way while putting my feet into the shoes.

* * *

Putting socks on: I have adapted some commercial products to make them work better and easier. The photo shows a blue purchased sock gadget to which I've screwed a piece of plywood on the bottom so it won't roll around while being used sitting on the floor. Then I tied a knot in the cord and obtained a "Grip-It" device (made for carrying suitcases and packages) that I slipped in just below the knot to make it much easier to pull the sock-holder with sock over my foot and then pull the sock off the gadget and onto my foot. I use the "stick" described earlier to pull the top of the sock onto my foot with my right hand while I'm pulling the rest of the sock with my left hand holding the handle pictured just under the knot.

* * *

Specific custom devices: I've described many commercial and homemade devices especially helpful to me. Each of you has specific abilities and difficulties, which will stimulate and encourage you to invent and create your specific solutions. Perhaps a plastic seat or chair in the bathtub or shower or a specific transfer board to help you move from one spot to another in your home. I now brush my teeth while in my shower, sitting on a waterproof chair. My toothbrush is kept in a slot in a soap dish on the wall, and the toothpaste tube sits in a plastic cup in the wall-holder. Much easier than trying to stand in front of the sink or even sitting in my scooter at the sink. A handicap-aids company (Stander) has a wide variety of bars, handles, props, guides, bedrails and mobility devices to make life and mobility easier and safer for you. A novel one is the Security Pole and Curve Grab Bar, discussed earlier, which makes standing and sitting easier. It's a floor-to-ceiling bar

Continued on next page

with a clever curved rotating grab-rail in the middle. I have a pair of them next to my bed and my couch, about two feet apart, so I can grab one with each hand when transferring to and from my scooter. The middle grab bar rotates into eight locked positions, so you can custom-configure it for your own specific needs.

How do you easily open or close a door, when your scooter is in the way? See the “Tina’s Tips” article on page 20 of this issue for my solutions to the problem of “Scooter in the Way” when going through a doorway.

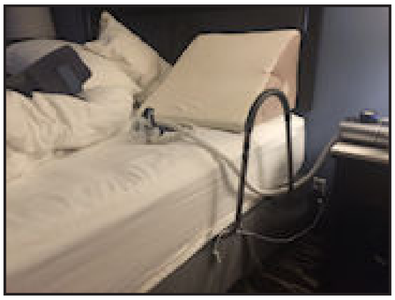
My shower has multiple vertical grab bars fastened into studs behind the tile. It also has folding seats, a wall mount for my toothbrush and toothpaste, clips for brushes, and a teak floor which is, much more durable and safer than a tile floor. Consider putting a small towel on the shower floor, which is much safer than standing on tile. Wringing out a floor towel is very preferable to slipping on the shower floor. You can also put non-slip rubberized pads on the shower floor. I carry several with me when I travel since I can’t know if the hotel bathroom or shower floor is slippery. Getting up from a toilet onto a tile or slippery floor in your bare feet is much safer with that portable non-skid pad to step onto.

Spasticity in our leg muscles produces “scissor walking” where our knees bump together with each step. This can be a major cause of sleeping difficulty; my knees grab each other all night! This problem is greatly alleviated by putting a foam rubber pad between your knees. You can also use a small folded pillow or towel. There are commercial between-the-knee foam rubber pillows (e.g. Hermell products) that can be washed. They also “stay put” and don’t unwrap like folded towels do.

SYNAPSE APPEAL

Synapse costs lots of money to print and mail, and we need your help to keep it going for another year. If you enjoy reading *Synapse* and want to show your appreciation of informative articles like these, please consider using the enclosed response envelope to make a donation.

Every little bit helps!



Finally, I present the ultimate display, which shows five separate handicap/medical aids used at the same time, when I was at a hotel. The message is that you always remain in charge of all your medical and personal needs. Note 1) the white charger cord for my cell phone for medical emergencies – always have a communication method handy, an emergency call device if not a cell phone, 2) the portable bedrail, between the box spring and the mattress, 3) the blue knee pad sitting on the bed, 4) the C-PAPP machine and hose used for sleep apnea, and 5) the foam rubber wedge pillow for head and upper body support, used by folks with GERD/heartburn, to keep the food in the stomach from coming up and causing heartburn and inflammation.

The disabled public is an important group of people whose special and unique needs are being addressed by many manufacturers. There is an enormous variety of wheelchairs and scooters, walkers, mobility aids, bathroom aids, personal devices like transfer boards, pads, safety devices, ramps and a multitude of clever aids for comfort, protection, mobility, bathing, transfers, driving, and safety. Space does not permit discussion of every product here, and I recommend you seek out a large local mobility store/surgical supply house as well as going to an Abilities Expo close to your home. These are held across the country in large cities, once a year, and will show you an amazing and extensive selection of things for people with handicaps. There are devices for quadriplegics to operate computers and scooters, safe bathtubs, service dog agencies, recreational activity demonstrations, health agencies for the handicapped and all manner of unique products you may not have seen or heard about previously.

Space here limits extensive discussion of my homemade handicap devices. If you would like more information about a specific device that I’ve mentioned, send me an email at malind@cox.net.

Malin Dollinger, M.D., HSP, SPG4, HUG*

*Handicap-Useful Gadgets

How to Get the Most from Your Doctor

By M. E. Hecht, M.D.

I get a lot of questions from patients surrounding: "How should I treat my doctor appointments?" "What should I ask?" and, "I feel like it's a one-way street." So, I am sharing with you some of the advice I have given to my patients.

For anyone 65 years or more, a visit to their doctor can be a real advantage. It can prevent potential problems and treat known conditions. It is perhaps best scheduled as an event much like the seasons: spring, summer, fall and winter.

I use the word "event" deliberately, rather than a chore to be avoided or dreaded. As an event, there are ways to take advantage of the visit to maximize the benefits. A doctor visit as a planned occurrence can represent a major tool for independence and control, and which of us doesn't relish these life qualities at any age?

For many, it's also an issue of you making the difference and becoming part of the solution. How you prepare, manage and organize the visit is key. With this type of thinking you can make the most of the visit, and believe it or not, make the most of and organize your doctor as well.

If you are prepared to relate precisely what you are feeling and how you are doing, it elicits both attention and interest from your physician.

MANAGE, ORGANIZE AND PREPARE FOR VISITS

You should come to your doctor's appointment ready to describe and quantify the following:

- Note any changes in your condition – when, how and how severe, as well as anything you did for it to modify or ameliorate it
- Note any changes in your response to the medications your doctor has prescribed
- Note any new signs or symptoms
- Note any changes in your activities and the results

REMEMBER TO EXCHANGE INFORMATION

Your appointment must not be a haphazard event. Prepare by writing a list short and to the point for each of these. Leave space beneath each for your doctor's answers and suggestions.

Additionally, be sure to:

1. **Always bear in mind it takes two to tango**, as the saying goes. A white coat doesn't disable the communication or importance of your full presence and understanding. A simple "could you repeat, or explain" if you don't get something, is not an imposition – as a matter of fact, it asks for recurrence or mistreatment, neither of which your doctor wants.
2. **Be sure your doctor answers in "patient language."** If he's using medical-ese, and you don't get it, in effect it hasn't happened.
3. **Deal with each category**, and ask until you fully understand the answer and the solution.
4. **Most importantly, write down the suggestions** the doctor is giving you.
5. **Upon your next visit to his office, refer to this list,** noting whether his solutions have been helpful, so-so or ineffective.

PREVENTIVE CARE TIPS FOR SENIORS

On concluding your visit, be sure you understand what your doctor has recommended. Understand both the effects desired and any major side effects. Remember:

1. **After the call with your doctor, write down the essentials** like change of medication or activities on your visit sheet.
2. **If what he has suggested is not possible or even probable for you to do, let him know** so that he can come up with a modification that makes it achievable.
3. **If it's something that requires a trial and report, be sure you establish when it's best to talk** to him. *(For most doctors if it's an emergency, he will respond even in the middle of seeing patients.)* But, be sure that you are calling about something that needs immediate attention. I always told my patients that at the end of my day, I would be available to answer all questions, even the repetitive or slightly inane.
4. **Keep a notebook of all of your visits and the significance of each.** One sheet *(dated of course),* can serve as your working "visit sheet." The next page is for your reactions to what was proposed.

The bottom line is whenever you visit your doctor, it is important to take responsibility for your health and become part of the solution, and to maximize and organize your visit. With this approach, you will find that your old one-way street has turned into a bright open highway for health – yours!

About the Author: M.E. Hecht, M.D., is a published author, freelance writer and Orthopedic Surgeon. Her published books and articles have been written for *Vogue Magazine*, Sunrise River Press, *The Wall Street Journal*, *American Medical News*, *Medical Tribune*, *Nations Business* and others. She is also author of "A Practical Guide to Hip Surgery" and "The Slip and Fall Prevention Handbook, You Make the Difference" – both books are available online at Amazon.

AUTUMN IN CAROLINA 2017 Is Set!!

The event will be held on October 7, just north of Winston-Salem, NC, in Rural Hall. We have used the same location for a number of years. All MND categories are welcome. Normally some folks that travel to attend arrive on Friday and depart on Sunday. We have a block of rooms on reserve at the Double Tree Inn (same motel we have used in the past, just totally remodeled and new name). Those arriving on Friday and anyone living nearby will gather for a Dutch meal at the Mayflower Seafood Restaurant, just a short trip from the motel. Our meeting will be on Saturday will be at Kingswood UMC, again a short distance from the motel, from about 9 am to 3-4 pm. For those staying overnight or locals wishing to join, we will have another Dutch meal at a local BBQ restaurant, again not far from the motel.

I have heard from six folks planning to attend and hope that interest will grow. If you travel and wish to utilize the motel, let me know and it will email information. Sorry Galen, October 7 was the only date that I could block out enough rooms in the motel. Perhaps you will have time to at least stop by.

Don Wilson (don-wilson@earthlink.net)
336-971-1288

● ● ● ● ● ● ● ● ● ● ● ● ● ● ●

SPFIllinois Connection

May 20, 2017

HSP: Sid Clark PLS: Hank Chiuppi

It finally happened. The SPFIllinois connection we held was on a day when the skies opened up with buckets of rain. Try walking with a cane /walker on wet pavement. Nevertheless, we had 17 at the SPFIllinois connection. In spite of some of our regular attendees being in or just released from a hospital, we had new attendees from Wisconsin and Indiana – welcome.

In our round robin, we discussed a number of topics that individuals wanted to bring up. We talked about the care of caregivers (important), home medical equipment, spinal cord stimulators, Medtronic, the Terry Wahls™ MS Diet, painkillers & drugs, Charlotte's Web, home remodeling & bathrooms, edema, Botox, and the upcoming Chicago Abilities Expo and the SPF Atlanta Conference, both on June 24. All of these subjects were discussed in addition to other topics. We cover a lot of ground based on the experience and knowledge of those at our connection. You can learn a lot by listening to what others have tried and what does or does not work for them. In your state gather your fellow HSPers and PLSers and share a get together. You'll make new friends. At our connection, we also talked about signing up for the *Synapse Newsletter* and checking the SP-Foundation website for the latest news and articles. To help SP-Foundation use Amazon Smile to fund SPF research.

Group Photo includes L to R: Back row: Hank Chiuppi, Frank Madrigali, Paulette Chiuppi; Carol Clark, Debbie Sexton, Darrin Duerst, Kyle Swanson, Heidi Swanson, Dawn Hendon, Joan Morris, Debbie Forsythe, and Richard Fairbairn; Center L to R: Phyllis Madrigali, Nancy Stewart, Chris Sexton, and Sid Clark.

● ● ● ● ● ● ● ● ● ● ● ● ● ● ●

Iowa Connection

August 26, 2017

By Jackie Wellman, Iowa State Ambassador, Member of SPF Board of Directors

The Iowa connection was held on August 26th. We munched on tacos, salsa and guacamole for three hours. Lots of gabbing going on, too.

On the right side in the front is David Gibson, Lynda Gibson, Karen Powers, Lois Wagoner, Roger Wagoner, Jackie Wellman, Dina Landphair, Caty Juhlin, Bruce Stolba, Kris Stolba.

Tina's Tips

By Tina Croghan; State Ambassador from Missouri

RAMPS

I have been sent several examples of ramps varying from rising a few inches to several feet. They all have a common thread of when they were installed; a ramp was really not needed. However, they were planning ahead for, "How am I going to be in 5 years?" or in some cases, "…next year?"

John Staehle
North Texas Region State Ambassador

When my HSP progressed to the point I had to use a power chair as my only mode of mobility, there were a number of accessibility modifications we had to make to our home. Being in a chair, I was confined to the main floor of our 2-story home. Since the only bathroom on the main floor was slightly larger than a powder room and had a 24″ wide pocket door, it was obvious we needed to make some significant changes. Fortunately, we had a small bedroom on the main floor to which we had room on the property to add an accessible bathroom. In 2007 we had an addition to that bedroom built with external access. A concrete ramp allows me to drive my chair up to the entrance to my expanded bedroom.

The floor of the garage is about 1.5″ higher than the driveway. I use an EZ Lok device to lock my chair in place when I am in my van. Attached to the bottom of my chair is a large "locking" bolt that reduces the ground clearance of my chair to something less than 1.5″. I asked the contractor that built the addition to make a 36″ wide ramp-like cut in the garage floor to allow access to the garage in my chair.

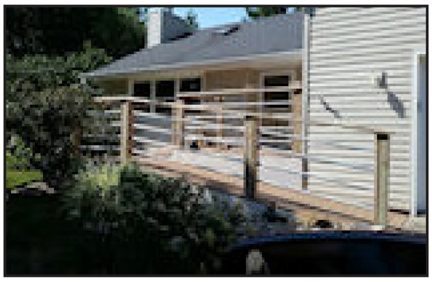
Several other ramps were added to allow alternate paths for me to enter the house, which I gratefully use when it is raining. Inside the garage I had a combination ramp and platform built to compensate for the 3″ difference between the garage floor and the floor level of the house. This ramp/platform, which leads to a covered patio, is constructed from plywood and 2x4s since it is totally protected from the weather. Tiny glass beads were added to the paint to improve traction especially on the incline.

Ramps from the garage to the patio and into the house were constructed from steel diamond plate and were primed with a rust-inhibiting paint to protect them from the weather.

Anne Moxley
HSP from CA

I keep my Scooter in the house at night, and I have a ramp on my patio door, which is wonderful. It is only 3″, but makes it possible. I love it.

Jackie Wellman
State Ambassador from Iowa



Our porch rotted off. I do not need a ramp yet, but we had one put on for the future. It was cheaper than rebuilding another porch. It is pretty awesome looking.

Continued on next page

Martha Weavers
From Ontario
I have a portable ramp in my garage so I can get my chair into the house. When we need to take the chair with us, the ramp can be moved so we can load the chair into the van. We have also taken the 'guts' out of the doorknob *(garage door into the house)* so all I need to do is run into the door for it to open.

Elizabeth M. Marin
I have an automatic ramp closer on my minivan.

Lewis "Sid" Clark
Co-State Ambassador for Illinois

Since I can no longer walk, we put a permanent ramp in our house which we could not do without. It is important to stay active and involved, so I recommend it. I did get a portable ramp for our son's house for family events. However, portability is in the eye of the beholder. We need his help putting it up, but if it was any lighter, it would not hold up. I have heard of some who got a cheaper, lighter ramp only to have it bend. Being on a ramp and having it fail underneath you is not something I would want to happen or experience. If you need a ramp to stay active, get one and stay active.

DOOR CLOSING

John Staehle
North Texas Region State Ambassador

Exiting the house in my chair is not without difficulty closing the doors behind me. I exit going forward and then have to turn around and drive back far enough to reach the door handle. Then I have to move the chair back a little, pull the door until it hits the chair, back up a little more, pull the door again and repeat this process until the door is totally closed. To solve this problem, I attached some nylon twine between 2 hook eyes that I screwed into the door *(or in one case, between the door knob and a hook eye)* that I can grab from outside the door threshold and pull the door closed. I start to close the door by grabbing the twine closer to the hinge side of the door and move along the twine toward the doorknob as the door begins to swing close. Nylon twine is tough enough to withstand pulling on it and it will last for several years.

There is a 36″ wide solid wood door between the dressing/vanity area of my accessible bathroom and the room with the toilet and the shower. I replaced the standard middle hinge on the door with a spring-loaded hinge. A door stop keeps the door open until I need to enter the toilet and shower room. I release the stop and as I roll in, I let the door close behind me. When I exit, I flip the door stop down and push the door open. The stop prevents the door from closing on me while exiting the room.

Ann Marker

I am a full-time wheelchair user, when someone cannot shut the door for me, I have a 30″ shoehorn I use to pull the door closed.

Don Wilson
State Ambassador from North Carolina

The best thing that I have found to assist in opening and closing doors while using a wheelchair is rubber hose (surgical hose). One 3-foot piece is tied to the inside door knob and a knot is tied in the loose end. A like piece is attached in the same manner to the outside knob. Using the wheelchair, unlatch the door and holding the knot, back up and the door opens. Drop that piece, go through the door and grab the loose end knot tied on the outside knob. As one goes though the door holding the hose, it will close behind you.

Mike Church
HSP from Missouri

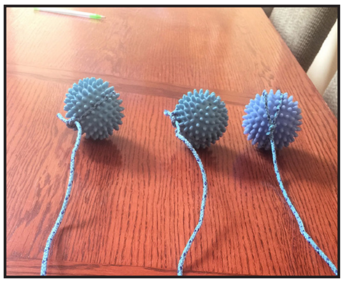
Parts needed, rope, 3 screw eye screws, 1 tennis ball or figure out another pull down idea. I roll through the door and close the door behind me just by pulling the tennis ball down.

Randy and Carol Barta

We have a 2-story house with an elevator. I can not close the door due to my left leg stretched straight out - my arms are not long enough - LOL - to grab the door handle and pull it closed. The door has to be closed as well as the gate inside the elevator before you can use it. SO - my husband and I went to Hobby Lobby's drapery section and purchased 3 ½ yards of the decorative cording and I cut it in half - one half for each door - the one downstairs and the one upstairs *(after wrapping clear tape around the place I was going to cut so the cording didn't unravel)* and knotted a "handle" to slip over the door knob and then a smaller one at the other end to grab and pull the door closed as I backed into the elevator *(very carefully)*. I also worked out a way to get the gate closed but that's not the "door" issue so I won't go there. But here's a picture of one of the doors with the rope handle - works great and doesn't get in the way of the elevator. We purchased a gold color to semi-match the paint on the door. The cording comes in many colors but we wanted it to match as closely as we could to the door color.

Malin Dollinger, M.D.

HSP from California



How do you easily open or close a door, when your scooter is in the way? Here is my solution to the problem of "Scooter in the way" when going through a doorway. When going out through an outside doorway, where the door opens outward, of course I need to close it again after I go through. But if I'm close to the door, my scooter is in the way. So I created a device to pull the door closed from a few feet away. Obtain a few rubber "laundry balls," made to toss in the wash. Find some strong thin cord and cut a piece about 2 ½ feet long. Wrap one end around the laundry ball (this ball has projections, which easily hold the cord), and the other around the stem (round part) behind the doorknob. After you exit, you run your hand down the cord and grab the ball. Then back up in your scooter, holding on, and you can pull the door closed without you being in the way. I use this on inside doors as well, whenever a door opens toward me in my scooter, and with the ball I can back up a couple of feet, holding onto the ball. In the photo of the ball/cord hanging in a doorway, there is also a red pocket reminder bag, to put your keys, cell phone, money, credit cards, and anything you will need and don't want to forget, such as a shopping or project list.

A safety/ ease of opening variation for the <u>bathroom</u> door is to place several layers of masking tape over the door latch opening in the door jamb. That prevents the latch from engaging the strike plate. In my home we rarely lock the door of the bathroom I use, just in case there's a problem/accident, and quick access is critical. With a ball door opener on that same doorknob, I ignore the doorknob and simply grab the top of the cord, slide my hand down to the ball, pull the door open, and let go of the ball as I'm going through. That door has a spring that closes it by itself, but because of the tape, it does not "latch."

Another help is to install one-piece door handles, like the kind you see on screen doors which are available in hardware stores, about 1/3 of the way between the hinges and the doorknob. When the door is wide open, it's hard for us to reach through the doorway and grab the doorknob. But that handle is much closer and easy to reach while sitting in your scooter and with it you can easily close the door.

Remember, if you have a tip or suggestion, send it to <u>tinacroghan@yahoo.com</u>.

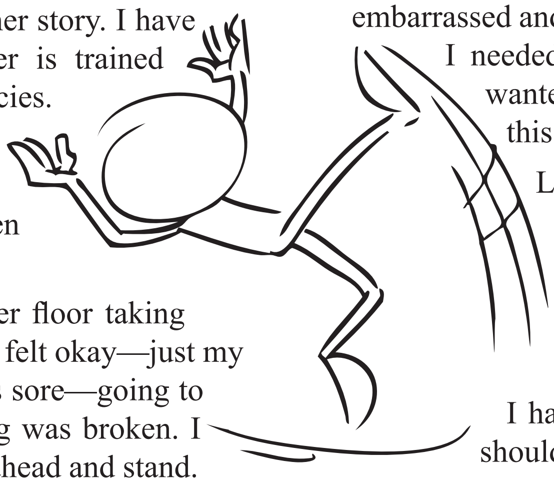
"I'VE FALLEN AND I CAN'T GET UP!"

By Tina Croghan, Missouri State Ambassador

I thought I knew how to fall. I never dreamed it would happen to me—although I should have known better.

Just after returning home from the SPF Annual Conference in Atlanta, I was taking my shower after exercising and stretching. My service dog "manned" his position ready to spring into action—sleeping under my vanity table.

After drying off, I went to step out of the shower and fell right on my bum! I continued to hold onto the grab bar with my right hand. I knew that when I fall to "just go with it" and not try to brace myself—I learned that the hard way! But that's another story. I have a service dog. After all, Thunder is trained specifically to help me in emergencies.



I remember seeing my arm in slow motion, thinking that I had never seen it turn that way. I then heard a loud "POP!"

At first I sat calmly on the shower floor taking stock of my injuries. I surprisingly felt okay—just my right arm was numb. My bum was sore—going to have a nasty bruise there! Nothing was broken. I didn't see any blood. I tried to go ahead and stand. Then the pain started slowly seeping in. I couldn't get my feet under me. My right arm was useless. My left was wet from the shower floor and couldn't support my weight.

All I could think of at this moment was that silly commercial—"I've fallen and I can't get up!" I only had the towel to kind of drape over me in case 911 was called. This was going to take my husband's help!

I called out for Thunder. Normally, he would come to my rescue with healing kisses and a wagging tail. This time he just stood there safely out of reach in case there was some catch to my pleas for help.

I told him, "Go get Timmy!" He knows that command. His tail started wagging. "This must be some new game!" He bounded out of the bathroom. Unknowingly, I had the bedroom door closed. Now Thunder has a special rope on the door handle and knows how to open the bedroom door. However, Thunder thinking, "We have never played this game before. I know I'm supposed to do something here. I'll just go into my kennel and hide!"

After 2 or 3 minutes of cries and shouts for help, Tim came to check up on me—thankfully!

As he was helping me off the floor of the shower, he continued a barrage of, "What was I doing?! I should have known better! Didn't I know that if I broke something, it would take me forever to heal?! Why didn't I send Thunder?!"

Okay—caregivers out there—I already felt stupid, embarrassed and now really hurting. The last thing I needed right now was a lecture. I just wanted a little compassion and to get off this wet floor!

Long story—I went to the doctor the next day and found that nothing was broken but I had dislocated my shoulder. That was the loud "POP" I heard. It fortunately, went back in on its own right away, but I had pulled all of the muscles in the shoulder area!

After a few days of ice packs, massages, chiropractics and slow stretching, I was able to type without waves of pain.

What I learned from this: My husband and I made a pact for me to never take a shower without him being within earshot and he has installed these "skid strips" (pictured).

Also, Tim vowed he wouldn't lecture me when I fell. *(He later apologized! He was just scared and reacted that way.)*

Now, I leave the bedroom door wide open, not just the bathroom door, for Thunder to, "Go get Timmy."

We have a new game now—"Go get Timmy!"

2017 Annual Conference

Thanks for the Memories

We'll See You Next Year!

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

1605 Goularte Place
Fremont, CA 94539-7241

NON-PROFIT ORG.
U.S. POSTAGE
PAID
Permit No. 1001
San Antonio, TX

How to Help

We operate out of the strength of our community, caring friends and sponsors. All of the hard workers in our Foundation, including the Board of Directors, are volunteers. Your help can make a difference!

Please contact us at volunteer@sp-foundation.org to help in one of the areas below or to suggest other ways you would like to get involved.

Support Research to Speed Our Cures by Volunteering

Below you'll find information about some of the ways you can help the SP Foundation in their search for cures for PLS and HSP.

Raise Funds: The primary focus of SPF is to raise funds to support research to find the causes, treatments and cures for Hereditary Spastic Paraplegia and Primary Lateral Sclerosis. Our fundraising activities include: SPF TeamWalks (actual and "virtual"); Golf Tournaments; national and international fundraising events like Rare Disease Day (the last day of February each year) and Giving Tuesday; and local special fundraisers. Individuals like you can help organize local fundraisers. Volunteers are also needed to secure corporate sponsorships and help with grant applications.

Patient Connections: Organizing a Connection, a social gathering for people to meet, share stories and help one another, is a great service. Events can be as simple as meeting for coffee! In many states, the SPF State Ambassador organizes and hosts their respective Connections (see below).

Conference Organizers: The SPF seeks event coordinators in or near metropolitan areas who can work with us to organize Conferences for our SPF community. These events feature speakers and programs on special topics of interest to our community as well as provide the opportunities for individuals to meet others with similar conditions. Conferences can be half-day or full-day events..

Communications: Individuals with writing, research, website or graphic design skills are needed to assist with various communication initiatives, including SPF's closed Facebook group and this newsletter, *Synapse*.

Ambassadors: Ambassadors welcome new members of the SPF community who reside in their respective states or regions and invite them to participate in local events. Visit the SPF website, www.sp-foundation.org, click on Get Involved, then Join the State Ambassadors to see a complete description of the State Ambassador job.

Business and Administrative Support: Volunteers with business and administrative skills can play a valuable role in administering the work of the SPF. Most of the help is coordinated through email correspondence and uses popular MS Office applications.