Synapse

Volume 18, Issue 1 Winter 2015

Newsletter of the Spastic Paraplegia Foundation

Letter from the President

Dear Friends,

I am writing this letter with Christmas and 2014 right behind us and the promise of a new year at our doorstep. Before our Foundation can begin our planned goals for 2015, I need and want to say a HUGE THANK YOU to you for your recent donations.

Our Foundation exists because of you. Our cure will be found because of you. You keep our hopes alive. Your total donations were well over our $100,000 year-end fundraising goal and so together with our anonymous matching donor, the total funds raised since mid-November, including the matching gift to date, is $259,948. Your kindness will really do a world of good and is very much appreciated.

Our goals as a Foundation for 2015 are many. Most importantly, of course, these dollars will be invested with or awarded to the very best scientific minds, doing the most promising HSP/PLS research on this planet. Our volunteer SAB Coordinator and attorney, Mark Weber, will continue to make sure that the hundreds of research proposals we receive are ranked by our world renowned Scientific Advisory Board to make sure that your dollars make incredible things happen.

To list just some of our other 2015 goals: SPF is switching to a much improved data base system to better keep records about and communicate with each person in our community in a more personal, individualized way. We plan to fill the missing SPF Ambassador positions in the seven states of: AK, MA, MN, MS, ND, NM and SD. Our ambassador coordinator, Jackie Wellman, will make certain that information and ideas are provided to all of our many state ambassadors.

We are working with many similar non-profit organizations, government agencies and HSP/PLS groups in many countries to create an international, multi-lingual, HSP/PLS Patient Registry that will provide researchers with vast amounts of information about HSP and PLS. Our website will report quarterly in 2015 about what our researchers are working on and what they are discovering. We can't divulge their secrets to competitors, but we can let you know briefly about the GREAT things they are accomplishing.

Social media continues to be a focus. Our Vice-President, Linda Gentner, will continue to welcome and facilitate communication and sharing on our several Facebook pages.

A film will be developed for our website on ways to stretch and exercise. Our medical advisor, Dr. John Fink, is working with SPF and Invitae Corp. to develop a webinar for neurologists about HSP.

Our website will be improved to make sure that data is kept current, up to date and easier to find. Many of the main pages of our website will soon have a button to click to be read in Spanish. We want to encourage more people with HSP or PLS to sign on to our data base so we can keep them informed and draw on their input for our Patient Registry. The editor of our Synapse Newsletter, John Staehle, will continue to improve and organize the stories so that you have the information that you need in an interesting, up to date, informative way.

Cicero once said that "gratitude is not only the greatest of virtues, but the parent of all others." Thank you so much for parenting, nurturing and developing the real hope for a cure for HSP and PLS with your continued donations.

Sincerely,

Frank Davis
President, SPF

Thank you

How to Help

We operate out of the strength of our community, caring friends and sponsors. Your help makes a difference!

Please contact us at volunteer@sp-foundation.org to help in one of the areas below or to suggest another way you can get involved.

Support Research to Speed Our Cures by Volunteering

Below you'll find information on some of the ways you can help SP Foundation in their search for a cure to 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 major fundraising activity consists of a Team*W*alk. Individuals can help organize local fundraisers. People are also needed to secure corporate sponsorships and help with grant applications.

Patient Connection Programs: Organizing a Connections 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 areas with large patient populations, SPF seeks to establish Chapters.

Conference Organizers: The SPF seeks event coordinators in or near metropolitan areas who can work with us to organize Conferences for our 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. 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.

Ambassadors: Ambassadors raise awareness about our disorders as well as enhance community building and industry relationships. You can assist with media relations, share your story, speak at local groups or help with grassroots advocacy.

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 Office applications.

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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).

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

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

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Please direct correspondence to:
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information@sp-foundation.org
www.sp-foundation.org

Connections

Austin Patient Connection

August 30, 2014

This year's Austin Patient Connection was titled "Moving Forward Together" with 18 people attending.

"Moving Forward Together" says that we are NOT ALONE with these two rare disorders Hereditary Spastic Paraplegia (HSP) and Primary Lateral Sclerosis (PLS). We went around the table with each person introducing themselves. In our three hour Connection, people asked questions and made comments. I consider ourselves "the experts" as we are the ones living our lives 24/7 with HSP or PLS or helping a spouse, child or friend with the disorders.

"Moving Forward Together" works well with sharing. Written information was given to attendees for the Spastic Paraplegia Foundation website, www.sp-foundation.org and access for excellent videos of the speakers at the 2014 SPF Annual Conference in Grapevine/Dallas, Texas, http://sp-foundation.org/2014-annual-conference-recap/.

Attendees who wanted to join Facebook for those with HSP or PLS were given the person to contact: lkgentner@aol.com.

Anyone is welcome to hold a Patient Connection. If you are interested, please let me know.

Marlene Doolen
Texas Ambassador
Spastic Paraplegia Foundation

SPF Illinois Connection

Sept 21, 2014

Hosted by Sid Clark (HSP) and Hank Chiuppi (PLS)

We met at the Buffalo Grove Arboretum clubhouse. For lunch we ordered Jimmy Johns' subs and had the driver take the Connection picture. We had 19 in attendance at this our fall meeting.

A lot of material was covered in the meeting. We had a presentation by the Pace paratransit service, an ADA accessible transit operating over the entire six-county Chicago region. We discussed gene testing – Pros / Cons Cost? What is the value to us with MND? What does the future hold? Who provides the test? What is the value in a name and what does it mean? (MND = motor neuron diseases = HSP/PLS +). After lunch we discussed being a mother, wife, mate who has HSP/PLS. How do you do it? How to you cope?

"Care For Candy Fundraiser" for one of our group that needs to purchase a handicap van was highlighted. Handicap vans are very expensive. (Thoughts anyone?) We also referred to the SFP website for "A Patient's Guide Through the Medical Maze". There are also many Resources and Links on the SPF Site.

We never have enough time to cover and share all that we want. For the next meeting we are looking for suggestions on topics that you want covered. By each of us sharing we learn a lot. Our next meeting will be in early spring. And yes we will schedule the pump at the next meeting. All are welcome. For information on future meetings email us at SPFIllinois@gmail.com. Thank you.

Included in the Picture are: Hank & Paulette , Sid & Carol, Steve, Lynn & Ann, Joan, Carolyn & Sarah, Phyllis & Frank, Andy & Candy, Rich, Stacy, Sue, and Tina (SPF board member) & Tim.

Picture by Croghan

* * *

> *"If we all did the things we are capable of, we would astound ourselves."*
>
> *– Thomas Edison*

13th Annual California Team*W*alk for Our Cures and Connection Weekend

September 19-20, 2014

By Linda Gentner

We couldn’t have asked for a more perfect day and, as in prior years, a few new people joined us.

Our Friday night dinner and “Share and Compare” discussion time *(with dessert after dinner)* was particularly inspiring. Kay Brady was so excited to share with the group, and her doctor gave her the okay to join us since her surgery was very recent. What she shared:

“These are the surgeries I had:

1. ***An Achilles tendon advancement. This is als known as a Murphy Procedure.***
2. ***A tibialis anterior tendon transfer.***
3. ***A flexor tenotomy.***

When looking for a surgeon I would recommend finding a surgeon who is board certified in Reconstructive Rear Foot/Ankle Surgery.

I no longer am pigeon toed and my toes lie flat”.

Jean Chambers shared about a special plastic to adhere to shoes to prevent wearing out so fast. She handed out samples to those who needed it. She said it needed to be done by a shoe cobbler. If you want more info, contact Jean directly.

Our yummy Friday night desserts: chocolate cake with our logo, chocolate candy with our logo and chocolate dipped strawberries (who said we liked chocolate – purely medicinal). There were 21 of us for dinner Friday night which included family – 8 with PLS and 3 with HSP. No where other than at our Connection do the PLSers out number the HSPers.

Saturday was another great Team*W*alk – a couple of us represented our friends who were Walkers or Rollers by Proxy. I was proud to wear a name tag for Dave Irvine, our SPF WA State Ambassador. We also had another good year of raffle and silent auction items. Our weekend was a great financial success too for research -- $34,500 and counting. Many thanks to our lead sponsor, Brocchini Farms. Kris had 8 of her sorority sisters in attendance and they buy lots of raffle tickets and bid on silent auction items!! Love those ladies!

Spreading Awareness - One Cup Of Coffee At A Time

By Tina Croghan
Missouri SPF State Ambassador

On a crisp Autumn day, I patiently wait in the drive-thru line of other caffeine-deprived individuals at our local Starbucks Coffee Shop. No, this isn’t a plug for Starbucks, but rather to show you how one small act on your part, may be the key to a cure. You never know!

When I finally drove up to the window and I was allotted my Grande, Non-fat, Pumpkin Spice Latte, I gave the barista an SPF card that I always keep in my car and offered to buy the lady’s coffee behind me. “Just give her this card and tell her to have a good day!”

The cards are of the SPF logo (see below). I don’t put my name on them.

Here are some things I’ve learned:

- Only buy coffee for another (woman/man) [The intention can be misconstrued! You get my meaning?!)
- Look in your rear-view mirror and do it when the person behind you is by herself or loaded down with kids. Not only will the mother really appreciate the coffee, but also the single individual is either going to or coming from work and needs the extra jolt.
- Avoid cars with multiple individuals. They tend to “splurge!” Your bill may be too high.

This can be done anyplace you frequent! Grocery store…fast food…mine just happened to be Starbucks. Just pick a price range you’re comfortable with.


SPF SPASTIC PARAPLEGIA FOUNDATION, INC.
Research / Education / Support
HEREDITARY SPASTIC PARAPLEGIA
PRIMARY LATERAL SCLEROSIS
www.sp-foundation.org

News Flash

Dear SPF Community,

I'm writing to share with you exciting news about a new program offered by Athena Diagnostics to help with the reimbursement process and provide patient assistance to increase access to and affordability of testing for patients who may have Hereditary Spastic Paraplegia (HSP) or Primary Lateral Sclerosis (PLS).

You are undoubtedly aware that patients who show signs of HSP and PLS often face a unique set of problems. Puzzling symptoms, slow referral to specialists, late or no diagnosis, and wrong tests ordered or wrong treatments given can worsen the problem.

For certain disorders, such as HSP and PLS, diagnostic testing is not always covered by the insurer. Many of the newer molecular tests are costly. Patients seeking answers are put in a difficult position of having to use funds that cover life's necessities to pay for the test, or worse, go without the test – including the insights the test result provides. Overwhelmingly, patients cite lack of insurer reimbursement or prohibitive cost as a major barrier to testing – and many continue to suffer from fear, anxiety, lack of control and helplessness as a result.

Athena Diagnostics has recently announced a new program to make diagnostic testing more accessible and more affordable. On September 1, 2014, the company launched the Athena Alliance Program to expand patient access to a variety of diagnostic methodologies and tests, especially those for rare and esoteric disorders.

Athena's priority is providing patient-centric customer service so that each patient has an individual specialist and a team of dedicated personnel to support them from the time of the order through test results. Athena Diagnostics offers a comprehensive test menu and intellectual property portfolio for neurological, neuromuscular, endocrine, and renal conditions through more than 400 diagnostic tests, including **Hereditary Spastic Paraplegia Evaluation** for the accurate diagnosis of HSP. Athena is offering financial assistance for any insured or uninsured patient who qualifies financially and has a balance that exceeds $250:

- Patients who have incomes of 100% or less of the Federal Poverty Level will not pay more than $250
- Patients who have incomes greater than 100% but less than or equal to 400% of income guidelines will be responsible for 20% of the amount due, not to exceed $600
- Patients who have incomes greater than 400% but less than or equal to 600% of income guidelines will pay no more than $800

Athena handles all billing of insurance and will file appeals, as needed, on behalf of the patient.

While SPF does not make recommendations pertaining to private companies, we do share information about services that offer benefit to our members. Please share information about this program with anyone indicating they are facing financial barriers to diagnostic testing.

If you or your contacts have more questions about the Athena Alliance Program, refer your clinicians to: AthenaDiagnostics.com/alliance

Sincerely,

Frank Davis

President, SPF

Exercise and You

DOCTOR FINK'S RECOMMENDED EXERCISES

(from HSP LISTSERV posting by Dr. John Fink, Mon, 1 Dec 2014)

Hello everyone,

As requested, this is a brief overview of my recommendations for exercise in HSP and PLS. One caveat: my recommendations are not based on scientific research of exercise methods in HSP and PLS. These recommendations are based on talking with many individuals with gait disturbance and finding what seems to be helpful.

The basic concepts are to a) find the problems, b) address the problems specifically both as isolated exercises and importantly, through complex task-based exercises; c) keep score of your progress, d) when tasks become easier, change the routine to make things more challenging; e) expect improvement *(recognizing it will be slow)*; f) core muscle exercise and aerobic conditioning are key.

Here are a few notes:

Identify the factors that make walking difficult. HSP and PLS affect walking differently in each person. For some individuals, spasticity *(affecting hamstrings, quadriceps, adductors, "heel cords" in variable proportion)* is the major problem. In other individuals weakness *(hip flexion, foot dorsiflexion, hamstrings for example)* or endurance is the major problem. Often weakness *(in certain muscles more than others)* and spasticity *(in certain muscles more than others)* occur together *(in variable proportions)* with balance difficulty and slowness in muscle activation.

Consultation with a neurologist, physiatrist, physical therapist, personal trainer are often helpful in identifying which factors are particularly problematic. This is the basis for developing a function-specific exercise program.

Develop an exercise program that a) "starts low and goes slow" *(begin with something you're capable of and increase the frequency and intensity by approximately 10% each week),* b) is graded *(increasing intensity and frequency)*; c) is monitored *(by you, keeping track of performance, and by your therapist or trainer)*; d) addresses the function-specific goals; e) is varied *(monotonous routines are difficult to maintain)*; f) has days off each week where other exercises are performed; g) ideally is done with exercise partners *(activities that are performed completely alone are difficult to maintain)*.

Both complex/contextual exercises *(e.g. climbing gym, water aerobics, kicking a weighted ball)* and isolated exercises *(leg lifts, abdominal exercise "crunch" machine at the gym)* are useful.

In my view, the value of stretching, balance, core exercises, and aerobic conditioning can not be overstated and should have a central place in the exercise routine. In my opinion, "exercise frequency" *(4 to 10 times a week)* is at least as important if not more important than the intensity of a given exercise period.

I hope this is helpful.

Sincerely,

John

John K. Fink, M.D.
Professor, Department of Neurology
University of Michigan

NEW MOBILITY: THE MAGAZINE FOR ACTIVE WHEELCHAIR USERS

www.newmobility.com

"New Mobility" encourages the integration of active-lifestyle wheelchair users into mainstream society, while simultaneously reflecting the vibrant world of disability-related arts, media, advocacy and philosophy. Our stories foster a sense of community and empower readers to:

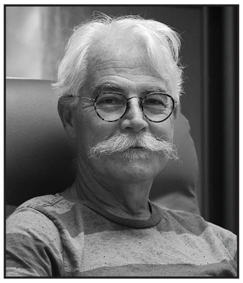
- Participate in all areas of life, including education, work, love, sex, home ownership, parenting, sports, recreation, travel and entertainment;
- Be informed of and take charge of health concerns;
- Obtain appropriate technology; and
- Assert legal rights.

2014 Research Awards

In October, the SPF Board of Directors, on the recommendation of our Scientific Advisory Board, approved the following proposals to receive 2014 research grants, all of which are proposals zeroing in on treatments for HSP or PLS -- a first for us. Science has truly come a long way…although there is still a long road ahead.


John K. Fink, MD


Andrew Grierson, PhD


Alan Mackay-Sim, PhD


Evan A. L. Reid, PhD


Holger Sondermann, PhD

John K. Fink, MD, Professor of Neurology, University of Michigan Medical Center, Ann Arbor, MI, **"New Treatment Strategies for Primary Lateral Sclerosis".**

Andrew Grierson, PhD, Kurt De Vos, PhD, & Mimoun Azzouz, PhD, Sheffield Institute for Translational Neuroscience, University of Sheffield, UK, "**Development of Gene Therapy for treatment of Hereditary Spastic Paraplegia**".

Alan Mackay-Sim, Ph.D, Director, National Centre for Adult Stem Cell Research, Griffith University, Brisbane, Australia, **"Finding Drugs to Treat Hereditary Spastic Paraplegia"**.

Evan A. L. Reid, Ph.D., University Lecturer in Medical Genetics, Department of Medical Genetics, University of Cambridge, UK, **"Defining a Pathway that could be Targeted to Increase Age at Onset of HSP"**.

Holger Sondermann, Ph.D., Associate Professor, Department of Molecular Medicine, Cornell University, Ithaca, NY, **"Towards a Mechanistic Understanding and Targeted Therapies in HSP type SPG3A"**.

cafemom
the meeting place for moms

CALLING ALL MOMS!!!

Check out a new web site

www.cafemom.com

Do you have a child suffering from any type of spastic paraplegia? HSP? PLS? Then this is the site for you.

www.cafemom.com/group/116957

A place where moms can come together – to share, encourage, and support one another!

Come Check Us Out!

Created by: Jessica Barlow-Anderson

IMPORTANT DATES TO REMEMBER!!

February 28, 2015: The 8th international Rare Disease Day, the main objective of which is to raise awareness among the general public and decision-makers about rare diseases and their impact on patients' lives. This year the theme is:

Living with a Rare Disease
- *Day-by-Day, Hand-in-Hand*.

June 26-28, 2015: The 2015 SPF Annual Conference at the Seattle Airport Marriott. The online registration form is now available at: **sp-foundation.org/2015-annual-conference/.**

MS Drug Aids Walking in Rare Condition

PHILADELPHIA -- A drug that aids walking in people with multiple sclerosis may have a similar benefit in those with a rare form of paraplegia (HSP), a researcher said here, In a small, proof-of-concept study of dalfampridine (Ampyra), 50% of patients with hereditary spastic paraplegia improved on several measures of walking ability, according to Nicolas Collongues, MD, PhD, of the University of Strasbourg Hospital in Strasbourg, France. “The improvements were clinically meaningful as well as statistically significant,” Collongues reported at the annual meeting here of the American Academy of Neurology. “Patients all say they can walk with more facility and can climb stairs more easily,” said Collongues. “This is a real clinical improvement.”

But because of the rarity of the disease -- about three people per 100,000 -- large clinical trials will be difficult to organize, which might rule out formal approval to use the drug in patients, Collongues said. He added that he and colleagues are trying to organize a larger study, using a cross-over design since a standard parallel group design would be impossible.

Source: MedPage Today, May 2, 2014

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Fishing for Causes and Cures of Motor Neuron Disorders.

Patten SA[1], Armstrong GA[1], Lissouba A[1], Kabashi E[2], Parker JA [1], Drapeau P[3].
Author information: [1]Department of Neuroscience, FRQS Groupe de Recherche sur le Système Nerveux Central and CRCHUM, University of Montréal, Montréal, QC H3A 2B4, Canada.
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Abstract

Motor neuron disorders (MNDs) are a clinically heterogeneous group of neurological diseases characterized by progressive degeneration of motor neurons, and share some common pathological pathways. Despite remarkable advances in our understanding of these diseases, no curative treatment for MNDs exists. To better understand the pathogenesis of MNDs and to help develop new treatments, the establishment of animal models that can be studied efficiently and thoroughly is paramount. The zebrafish (Danio rerio) is increasingly becoming a valuable model for studying human diseases and in screening for potential therapeutics. In this Review, we highlight recent progress in using zebrafish to study the pathology of the most common MNDs: spinal muscular atrophy (SMA), amyotrophic lateral sclerosis (ALS) and hereditary spastic paraplegia (HSP). These studies indicate the power of zebrafish as a model to study the consequences of disease-related genes, because zebrafish homologues of human genes have conserved functions with respect to the aetiology of MNDs. Zebrafish also complement other animal models for the study of pathological mechanisms of MNDs and are particularly advantageous for the screening of compounds with therapeutic potential. We present an overview of their potential usefulness in MND drug discovery, which is just beginning and holds much promise for future therapeutic development.

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GENE TESTING FOR HEREDITARY SPASTIC PARAPLEGIA

NOT COMPLICATED OR COMPLICATED

I was diagnosed with Hereditary Spastic Paraplegia (HSP) in 1993 when there was no official test to determine HSP. Testing was done by elimination of other disorders through walking in the doctor’s office, worn toe tips of shoes, an MRI to eliminate other disorders, and my father had been diagnosed with HSP years earlier. I accepted my diagnosis and went on with life.

At the 2014 Spastic Paraplegia Foundation (SPF) National Conference, Dr. John Fink talked about the advances in research and two FDA approved drugs used for other disorders that may help, but not cure, HSP. With this news, I was excited to find out which HSP gene I had. Testing is possible **NOW**. Through advances in research over the past few years, a blood test can determine most known HSP genes that are either HSP Uncomplicated or HSP Complicated.

Linda Gentner, Spastic Paraplegia Foundation Vice-President, www.sp-foundation.org, let people know through SPF FaceBook about a company, Invitae, that does gene testing for HSP. I called Invitae to find out how to get the test done. A Requisition Form was sent to my doctor to complete and return to Invitae. Invitae called me to let me know where to go for the blood draw, and the blood draw materials were mailed to my home to take with me for the blood draw. The blood draw was picked up by Fed Ex at the company of the blood draw and delivered to Invitae. Within a month, the test results were sent to my doctor, who then contacted me and mailed me the genetic testing results.

For HSP gene testing, I highly recommend Invitae, www.invitae.com, 800-436-3037. Their HSP gene testing cost is $1,500 for either HSP Uncomplicated or HSP Complicated. If needed, Invitae can work out a payment plan for you.

Marlene Doolen, Texas Ambassador
Spastic Paraplegia Foundation

* * *

SCRIPPS RESEARCH INSTITUTE SCIENTISTS SHED LIGHT ON CAUSE OF SPASTIC PARAPLEGIA

LA JOLLA, CA—September 29, 2014—Scientists at The Scripps Research Institute (TSRI) have discovered that a gene mutation linked to hereditary spastic paraplegia, a disabling neurological disorder, interferes with the normal breakdown of triglyceride fat molecules in the brain. The TSRI researchers found large droplets of triglycerides within the neurons of mice modeling the disease.

The findings, reported this week online ahead of print by the journal Proceedings of the National Academy of Sciences, point the way to potential therapies and showcase an investigative strategy that should be useful in determining the biochemical causes of other genetic illnesses. Scientists in recent decades have linked thousands of gene mutations to human diseases, yet many of the genes in question code for proteins of unknown function.

“We often need to understand the protein function that is disrupted by a gene mutation, if we’re going to understand the mechanistic basis for the disease and move towards developing a therapy, and that is what we’ve tried to do here,” said Benjamin F. Cravatt, professor and chair of TSRI’s Department of Chemical Physiology.

There is currently no treatment for hereditary spastic paraplegia (HSP), a set of genetic illnesses whose symptoms include muscle weakness and stiffness, and in some cases cognitive impairments. About 100,000 people worldwide live with HSP.

UNCOVERING CLUES

In the new study, Cravatt and members of his laboratory, including graduate student Jordon Inloes and postdoctoral fellow Ku-Lung Hsu, focused on DDHD2, an enzyme of unclear function whose gene is mutated in a subset of HSP cases. “These cases involving DDHD2 disruption feature cognitive defects as well as spasticity and muscle wasting, so they’re among the more devastating forms of this illness,” said Cravatt.

To start, the researchers created a mouse model of DDHD2-related HSP, in which a targeted deletion from the DDHD2 gene eliminated the expression of the DDHD2 protein. “These mice showed symptoms similar to those of HSP patients, including abnormal gait and lower performance on tests of movement and cognition,” said Inloes.

Prior research had suggested that the DDHD2 enzyme is expressed in the brain and is involved somehow in lipid metabolism. One study reported elevated levels of an unknown fat molecule in the brains of DDHD2-mutant HSP patients. Cravatt’s team compared the tissues of the no-DDHD2 mice to the tissues of mice with normal versions of the gene, and also found that the mutant mice had much higher levels of a type of fat molecule, principally in the brain.

Using a set of sophisticated “lipidomics” tests to analyze the accumulating fat molec-ules, they identified them as triglycerides—a major component of stored fat in the body, and a risk factor for obesity, atherosclerosis and type 2 diabetes. “We were able to show as well, using both light microscopy and electron microscopy, that droplets of triglyceride-rich fat are present in the neurons of DDHD2-knockout mice, in several brain regions, but are not present in normal mice,” said Inloes.

For the next phase of the study, Cravatt’s team developed a complementary tool for studying DDHD2’s function: a specific inhibitor of the DDHD2 enzyme, one of a set of powerful enzyme-blocking compounds they had identified in a study reported last year. "After four days of treatment with this inhibitor, normal mice showed an increase in brain triglycerides," said Inloes. "This suggests that DDHD2 normally breaks down triglycerides, and its inactivity allows triglycerides to build up."

Finally the team confirmed DDHD2's role in triglyceride metabolism by showing that triglycerides are rapidly broken down into smaller fatty acids in its presence. "These findings give us some insight, at least, into the biochemical basis of the HSP syndrome," said Cravatt.

LOOKING AHEAD

Future projects in this line of inquiry, he adds, include a study of how triglyceride droplets in neurons lead to impairments of movement and cognition, and research on potential therapies to counter these effects, including the possible use of diacylglycerol transferase (DGAT) inhibitors, which reduce the natural production of triglycerides.

Cravatt also notes that the same approach used in this study can be applied to other enzymes in DDHD2's class (serine hydrolases), whose dysfunctions cause human neurological disorders.

Other contributors to the study, "The hereditary spastic paraplegia-related enzyme DDHD2 is a principal brain triglyceride lipase," were Melissa M. Dix, Andreu Viader, Kim Masuda, Thais Takei and Malcolm R. Wood, all of TSRI. Ku-Lung Hsu is now an assis-tant professor of chemistry at the University of Virginia.

Support for the study came from the National Institutes of Health (DA033760, DK099810, DA035864 and GM109315).

ABOUT THE SCRIPPS RESEARCH INSTITUTE

The Scripps Research Institute (TSRI) is one of the world's largest independent, not-for-profit organizations focusing on research in the biomedical sciences. TSRI is internationally recognized for its contributions to science and health, including its role in laying the foundation for new treatments for cancer, rheumatoid arthritis, hemophilia, and other diseases. An institution that evolved from the Scripps Metabolic Clinic founded by philanthropist Ellen Browning Scripps in 1924, the institute now employs about 3,000 people on its campuses in La Jolla, CA, and Jupiter, FL, where its renowned scientists—including two Nobel laureates—work toward their next discoveries. The institute's graduate program, which awards PhD degrees in biology and chemistry, ranks among the top ten of its kind in the nation.

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Be Informed…Be Prepared

Tina Croghan

Now that our preoccupation with getting the house ready for "The Holidays" is over, we must not relax and ignore preparing ourselves for emergencies. Emergencies happen when you least expect them. Emergencies aren't just weather related either.

Some things you might consider are highlighted in FEMA's mailer.

Be Informed!

It is important to know what types of emergencies are likely to affect your region. For more information about specific types of emergencies, visit www.ready.gov/be-informed.

Have a Plan!

Being a high school teacher in theatre for many years, I know rehearsal is key! Make sure you have physically rehearsed your plan. It's not enough to KNOW your lines. Just the same, it's not enough to just KNOW what you would do if the unthinkable happened.

If you have a **fire** in the middle of the night, how would you get out?

Plan two ways out of every room in case of fire. Check for items such as bookcases, hanging pictures, or overhead lights that could fall and block your escape path.

For more fire safety tips, go to http://www.usfa.fema.gov/citizens/disability.

Be Informed…Be Prepared
(Continues next page)

Remember, you're going to be stiff! With the added anxiety, you will be even stiffer and may have clonus, too! Don't try to stand up. That's where the smoke is anyway. Grab your pills, cell phone and *CRAWL* to safety! Once you get out of the burning house, don't go back in for ANYTHING! Chances are your loved ones or your pets are already out—remember they have animal instincts. Have a meeting place--the neighbor's mailbox across the street (remember the fire trucks are going to be blocking yours!) Your family might not be together when disaster strikes. Have a plan how you will contact each other.

Those of you on the West Coast have experience in dealing with an **earthquake.** Once again, drop down on your hands-and-knees and crawl to a doorway or under something like a table or desk. Clasp your hands over your head to protect it.

What if the emergency is a **tornado?** Fortunately we have the capability now of being pre-warned! Heed those warnings! Do you have a basement? I keep a backpack loaded with things I think I might need**.**

I have a flashlight, a bottle of water, a radio and extra batteries. I will grab my pill case and my cell phone before I head down to the basement. Before I got my chair lift, I would scoot down on my bottom to reach the basement.

The first decision you are going to have to make is whether to stay or evacuate. Use common sense, however, if you go to a public shelter, it is important to remember that the law provides that only service animals will be admitted. You should know in advance where you would take your other pets. Visit http://www.ready.gov/animals.

Create a Personal Support Network

Everyone should make a list of family, friends and others who will be part of your plan. Make sure that someone in your personal support network has an extra key to your home and knows where you keep your emergency supplies.

If you use a wheelchair, oxygen or other medical equipment, show friends how to use these devices so they can move you or help you evacuate. Practice your plan with your personal support network.

If you undergo routine treatments at a clinic or hospital, or if you receive regular services at home such as home health care, meals, oxygen, or door-to-door transportation, talk to your service provider about their emergency plans.

If you use medical equipment in your home that requires electricity to operate, talk to your health care provider about a back-up plan for its use during a power outage." -FEMA

Another good idea from FEMA is to --

Contact Your Local Emergency Information Management Office. Some local emergency management offices maintain registries for people with disabilities. Some registries are only used to collect planning information; others may be used to offer assistance in emergencies. If you add your name and information to a registry, be sure you understand in advance to meet access and functional needs what you can expect. Be aware that a registry is NEVER a substitute for personal preparedness. Even if the registry may be linked to first responders, assistance may not be available for hours or days after a disaster. Contact your local emergency management agency to see if these services exist where you live, or visit http://www.ready.gov/ to find links to government offices in your area."

Keep 2 Kits!

One supply kit for sheltering-in-place. Figure on being cut off for 2-3 days. In which case, you're going to need food and water. I keep a case of bottled water in the basement along with a box of granola bars. I will grab my pill case and cell phone and all of the things in my smaller kit for evacuating. For a suggested list of supplies, visit http://www.ready.gov/

***"Include Important Documents in Your Kit -** Include copies of important documents… such as family records, medical records, wills, deeds, social security number, charge and bank account information, and tax records. Also be sure you have cash or travelers checks in your kits in case you need to purchase supplies. It is best to keep these documents in a waterproof container," says FEMA.*

Be Informed…Be Prepared
(Continues next page)

Be Informed...Be Prepared (Continued)

I put all of my RX list on a card in my wallet. I use this same one when I go to the doctor and they want all of my medical history. This saves me LOTS of time! You can do the same thing for your emergency kit. This is the larger one that STAYS. FEMA suggests you put important information on a "flash" or "thumb" drive. (If you don't know what this is, ask your kids to do this for you!) Put this in your kit along with some cash and access to your finances to protect your family in case an emergency happens.

Get Involved!

People with disabilities often have experience in adapting and problem solving that can be very useful skills in emergencies. To find out more about potential volunteering and emergency response training opportunities, go to http://www.ready.gov/volunteer.

[Editor's Note: For additional information, see publication FEMA 476, "Preparing for Disaster for People with Disabilities and Other Special Needs," also available from the American Red Cross as publication A4497. This booklet is online at www.fema.gov and www.redcross.org. It is also available by calling FEMA at 1-800-480-2520, or writing: FEMA, P.O. Box 2012, Jessup, MD 20794-2012. JDS, Ed.]

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Useful Tips

from Ronnie Grove

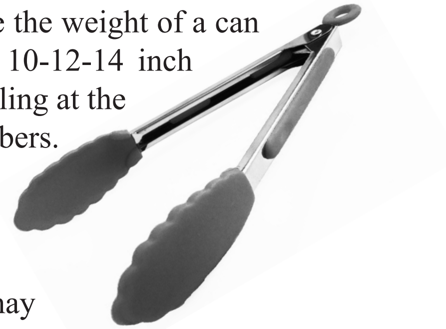
Great Guns…I thought my son had taken mine (BBQ tongs) home, but found it hid behind my cookbooks. It is wonderful for picking up things. One of the best for me is ice cubes. Lol! My fridge doesn't have ice in the door so I have to stand next to it and hold the cabinet top with one hand and reach over and get ice out of the bin in the freezer. I usually drop one or two. If you have never tried to pick up an ice cube, you should try it. Ha! I used to have to get it wedged against the wheel of my chair and finally be able to hold it after several attempts. I keep the gadget in my trash can in the kitchen and am forever using it.

Thanks…Arlene in Texas

Dear Arlene in Texas,

I know that ice cube routine by heart. Paper towels are good for getting a grip on those run away cubes if the tongs are misplaced.

You know, sometimes we know one thing works better than something else but don't stop to try to figure out why. The tong thing came to me a couple weeks ago and I don't even remember what I was doing. I have the grippers, like everyone else. And I use them for lots of things from time to time, but the tongs are still my favorite. It is because they are shorter. I guess it is physics—is that the word I want? It is much easier to handle the weight of a can of veggies at the end of 10-12-14 inch tongs than when it is dangling at the end of 24-30 inch grabbers. I can't live without my tongs. I keep a pair on the walker all the time.



Another thing I may or may not have mentioned that works wonders for me. I bought a carpenter's apron and tied it around the back side of my scooter basket (facing me). I laced the tie strings in and out of the front of the basket. Now I have pockets I can reach for my van keys and remote, pens and paper, a couple tools---a nice long screwdriver that I sometimes find helpful for knocking stuff off the shelves in grocery stores, my tongs, my reading glasses and my camera. Scripts from the doctor go in there where I can easily find them when I get to the pharmacy. When I clean it out. once a year or so, it's like Christmas. Never know what might be down in those pockets: change, maybe a bill or two, someone's email address on a scrap of paper, piece of wrapped candy, hand sanitizer, note pads, road maps…surprise after surprise. Couldn't do without it.

Happy "tong-ing." We could have an ice cube fund raiser. Have people pay so much a cube for all we can pick up in a given amount of time. You and I, of course, would have the advantage. We've had lots of practice.

Up Close and Personal

HSP and Happiness

By Mary B. Schultz

You are NOT your HSP! You are a happy person with a condition called HSP. HSP does not define you as a person.

The scientific study of happiness is relatively new. Traditionally, psychologists studied depression, anxiety, addictions, and UN-happiness. *(Good news: If you suffer from depression, anxiety, or addiction, there is a lot of psychological and psychiatric help for you.)* Recently, there has been more attention on the UP side of life, happiness as opposed to UN-happiness. Scientific studies consistently substantiate a very surprising fact: Life's circumstances, for example HSP, are relatively insignificant (about 10%) in contributing to your overall happiness. Generally, there are three categories of factors that determine your happiness: (1) circumstances (10%); (2) genetics (50%); and (3) factors YOU control (40%). So, HSP decreases your overall happiness, at most, 10%. It is not very significant.

Life's circumstances not only are relatively insignificant in contributing to your happiness, any effect of a change in life's circumstances is short lived. In other words, changing life's circumstances, like wiping out HSP entirely (we wish) would at most improve your happiness a small amount, and such a change would not have a lasting effect on your overall happiness.

Personally, I have a form of HSP that is known as "adult onset." Although I recall problems with my balance, sudden weakness in my joints and legs, and muscle spasms, even when I was a little girl, I did not manifest significant symptoms till I was in my 40s, and was not diagnosed with HSP till I was in my 50s. Yet, I am not UN-happier now that before I was diagnosed with HSP! *(I am increasingly angry with HSP, and frustrated that I can no longer "do" what I formerly did routinely and without thinking about my accomplishment, but not any more unhappy….)*

There is a total of 90% of factors that contribute to our happiness, other than HSP. There is a body of "happiness psychology" that suggests we already have within us the ability to be happy. There is a significant concept in modern psychology that we can change our genetically determined "set point" of happiness, the 50%. Overcoming genetics to promote our own happiness is a worthwhile endeavor, but beyond the scope of this short article. While there are many psychologists that are working to overcome genetics, you should work on factors under your control that contribute to your happiness, the **40%, RIGHT NOW**.

My own one-word imperative after having read some of the "happiness literature" is "**NOW!**" Strive to live and be present in the NOW – the present, not the past, not the future. Avoid thinking "If only…"

And among the most important factors that we control, and that contribute to our happiness is to "connect." Do not battle HSP on your own. Although HSP is rare, there are many others who also have HSP. The Spastic Paraplegia Foundation offers "connection" through mutual support and sharing with others who have HSP.

This article is intended to provide general information only. It does not constitute, nor should be relied upon, as providing professional advice or opinion relating to specific facts or circumstances.

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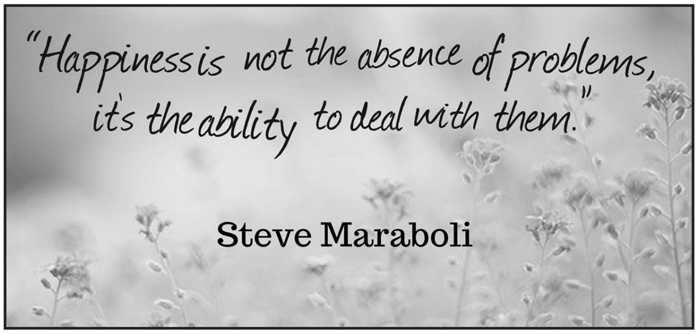

SYNAPSE APPEAL

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

My Airport Experience

by Tracy Scott, HSP

August fifteenth, I decided to take a trip to Nashville, Tennessee with my bird, Smoke. I searched hotels that allowed animals, made reservations, and even booked my flight on expedia.com. I emailed site-seeing places, to see if Smoke could come with me. I checked to see if Nashville had a disabled ride, like here in Fairfax County Virginia, it is called Metro Access. I found one called Access Ride. I printed the application and mailed it in.

Since this was my first trip after a car accident I was involved in December 2008 that left me with a Traumatic Brain Injury with short term memory problems. I also use a walker because my balance is so awkward. I use a walker for short distances and a wheelchair for longer. I even made an itinerary for my Mother, what I would be doing each day with addresses and telephone numbers, so she would not be worried.

I scheduled Metro Access to take me to Reagan National Airport and scheduled all my Access Rides in Nashville. The day came and Smoke and I were ready. I put Smoke in his carrier and off we went in my wheelchair. The Metro Access driver was very nice. I do not think she had dropped people off at the airport before. After taking all the straps and seat belts off me and my wheelchair, we got out and a gentleman came up to her and said, "This is not where you drop off people. Go over to section B." Back into the van we went and got all hooked up again. I told her, "No, worries, I am four hours early."

I wanted to be early because I did not know how long it would take to get my & Smoke's ticket. I thought going through security will take longer and I wanted to have plenty of time. The automobile accident I was in, left me with a metal plate where my humerus bone was and my pelvis, where the screw was, is removed, but the nut is still in there.

When I get in the door, I asked a man, "Where is American Airlines." He pointed to his right. I was on my way. People would cut me off, walk in front of me and come to a complete stop and start talking to the person next to them. I passed many employees that just looked at me. No one offered to help me. I finally made it over to American Airlines and when I showed them the Expedia American Airlines confirmation code, he said I needed to go to U.S. Airways. I thought to myself, this is not good!

Then he told me that this other man will help me get to U.S. Airways. That man was pushing an older lady who was in a wheelchair. Well, I waited about six minutes and just wheeled myself to U.S. Airways. When I finally got there, a young man looked at my papers and asked me if I'd like to leave earlier and I said, "Sure."

Then another man walked up to me and said, "What is in that thing" and pointed to Smoke in the carrier. I said, "A bird." Then he said, "No birds allowed." I said, "The American Airlines website lets pets." He went to get his supervisor. An older man with white hair came over and said, "No birds!" I said, "Well then I'd like to cancel and get a refund." He walked away and came back to me and said, "Here is your refund." I did not even look at it. I just put it in the folder I had full of my papers for my trip.

I wheeled myself right out the door because I knew I needed to get a taxi to go home. Many workers passed me, while I was crying. They just looked at me and walked right by. I saw a taxi and caught his attention. He tried to get my wheelchair in his car, but it would not fit. He told me to go downstairs where the taxi's can pick up people. I went in the airport again, people running right into me. I shook my head and started crying again.

I found the elevator and went down stairs and left this awful place. This is no place for someone with a disability. This has been my worst experience ever!

When I got home, I looked at the paper that the supervisor gave me and it was a refund for someone else. I called American Airlines to see if I could get a refund of $472.70 and they said I have a credit. I never want to fly U.S. Airways or American Airlines!

Rebecca Hart and Schroeters Romani Start Season with High Score at White Fences

Wellington, FL - December 17, 2014 -

Rebecca Hart and Schroeters Romani shined Saturday afternoon during the Polar Express show held at the White Fences Equestrian Center in Loxahatchee, Florida. The talented duo rode to the score of 78.382 percent for the FEI Para Team Grade II test during the national show, kick starting their season as they prepare for the Global Dressage Festival CPEDI3* in January 2015.

Rebecca Hart and Schroeters Romani prepare for their FEI Para Team Grade II test.

Hart traveled to Florida in early December to work with Wellington-based international dressage trainer Todd Flettrich out of Cherry Knoll Farm, which is also a partial owner of Romani. Hart and Flettrich have worked consistently on warm-up and preparation with Romani after taking a break from showing when the mare returned from the 2014 Alltech FEI World Equestrian Games (WEG) in Normandy, France. White Fences offered the ideal venue for Hart to prep for the upcoming CPEDI3*.

“She felt really good,” Hart smiled. “We wanted to come out and use this show as a schooling session to get her ready for the CPEDI3* that we have in about three weeks. I just wanted to get her out and about because she had a little bit of a break after WEG. This was her first time out at a recognized show since then.”

Hart continued, “I am really happy with the direction we are going. She was looking at the atmosphere and looking at everything, but she was able to keep her focus, which was really nice. We are definitely growing as a pair. I am more confident in my ability and my cues with her. It’s nice to be able to give her that confidence.”

The pair rode a fluid test with great relaxation and expression, and they earned the highest score of the weekend at White Fences. Hart, a two-time Paralympic dressage rider, has her eyes set on the 2016 Paralympic Games in Rio de Janeiro, Brazil. There are still many more strides to make before their journey to the podium, but Hart feels confident in their growing partnership.

“Leading up to the next event, I have been working on getting the rhythm, the cadence and the relaxation in order to sustain it through the test. I am working on really getting her to focus on me during the ride. I want to get us into our own little bubble out there so that we can block out everything. We are really going in the right direction, and I want to keep building on what we did here. I think this is a good baseline, but we can be even better,” Hart concluded.

Phelps Media Group, Inc. International

Rebecca Hart and Schroeters Romani rode to the high score of 78.382% during the Polar Express Show at White Fences.

Fantastic
Fabulous
Exceptional CONGRATULATIONS
CONGRATULATIONS
Fantastic Fabulous
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Fabulous OUTSTANDING Exceptional
Terrific

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2015 Annual Conference

We have another wonderful conference to look forward to this year. Seattle is a beautiful city and our speakers are exceptional, with something to offer everyone attending. Dr. Fink will once again be speaking and facilitating a Breakout Session. His session last year was one of the most well-attended we have ever had! Rebecca Hart, who was unable to attend last year due to weather, will be speaking this year. We all look forward to hearing from her. New to the conference this year are Dr. Christina Fournier from Emory University and Dr. Catherine Lomen-Heorth from UCSF. I know we will all benefit from what each speaker will contribute this year.

The online registration form is now available. Please pay special attention to all conference deadlines and email me at claudiadavis6@yahoo.com if you should have any questions. Remember, Seattle is a beautiful city and would make a great place to extend your conference stay for a family vacation.

We look forward to seeing each of you there!

https://sp-foundation.org/events/2015-annual-conference/