

## Research

Genetic testing may confirm a suspected diagnosis or rule out disorders with similar symptoms. A genetic diagnosis may also help predict disease progression or inform family planning. Seek board-certified genetic counselors to discuss test options and results.

Genetic research takes many years of gathering information and blood samples from patients, their family members and research volunteers to form the medical data or observational studies. Scientists use the data to perform complicated genetic analyses. Observational studies aim to better understand the relationship between the clinical characteristics (phenotype) and genes (genotype), and to develop biomarkers that might be useful in aiding therapy development for this group of disorders.

The state of HSP and PLS research is “exploding,” according to Professor John Fink, M.D. Since SPF’s inception in 2002, its all volunteer-driven efforts and fundraising activities are helping leading scientists unravel many of the questions regarding the complicated biochemistry of these diseases. Medical breakthroughs look to be very close and only seem limited by the amount of research that the Spastic Paraplegia Foundation can afford to sponsor. SPF has funded over \$7.5 million in medical research to date, but each year several research proposals are left unfunded due to a lack of financial resources.

SPF research grants offer primarily “seed monies” to assist investigators with new ideas, those in the early or pilot phase of their studies, or as additional support for ongoing investigations with a demonstration of need. In addition to furthering HSP/PLS research, scientists receiving SPF grants can leverage these funds to attract new and greater grants from other sources, like the National Institutes of Health. Researchers submit highly technical proposals. Our Scientific Advisory Board recommends the best for funding.

The Spastic Paraplegia Foundation is dedicated to advancing research and finding cures for HSP and PLS. Dramatic breakthroughs have been made in the past decade that gives those that suffer every reason to hope for treatments and therapies in the coming years. Moreover, uncovering more of these riddles may also lead to important findings for related conditions such as Parkinson’s disease, MS, ALS, spinal cord injury and Alzheimer’s disease.



**Spastic Paraplegia Foundation**  
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## **Like, Follow, Subscribe, Share SPF**



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Raise Funds for  
Medical Research  
& Awareness for  
#HSPandPLS

# #HSPandPLS

# SPASTIC Paraplegia Foundation

SPF operates with the strength of our friends, & sponsors, & community.

Donate, Fundraise, Participate.

Help Us Find a Cure.

### DONATE

Your support is needed now more than ever! Donate today – together we can make a tremendous difference.



### FUNDRAISE

Raise funds to help find the cure! Learn about how to organize a local fundraiser and help a great cause.



### PARTICIPATE

Support research that's working towards a cure. Find out more about how you can help the SPF.



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**What are HSP and PLS?**  
 Hereditary Spastic Paraplegia (HSP) and Primary Lateral Sclerosis (PLS) are chronic, degenerative neuromuscular diseases, like Lou Gehrig's (ALS). These diseases greatly diminish the quality of life of those affected. Many patients with HSP and PLS are initially

misdiagnosed with ALS, Multiple Sclerosis (MS), or Cerebral Palsy (CP), causing confusion and resulting in improper treatment. Currently, there is not an accurate count of people affected by these diseases, so we are trying to find outreach measures. The Spastic Paraplegia Foundation, Inc., is working to find ways to locate every person affected with HSP or PLS in order to gain the information for scientists to work toward finding treatments and cures. Get your name on our list!

**What is the Spastic Paraplegia Foundation (SPF)?**

The SPF is a volunteer driven, non-profit organization dedicated to finding cures for Hereditary Spastic Paraplegia (HSP) and Primary Lateral Sclerosis (PLS), closely related groups of rare, neurological disorders. The Spastic Paraplegia Foundation, a 501c3 organization, is the only organization in the USA dedicated to HSP and PLS, helping leading scientists unravel many of the questions regarding the complicated biochemistry of these diseases. The SPF is committed to funding research to find treatment or cures, educating people affected by these conditions, and raising awareness of HSP and PLS

The SPF Board of Directors is comprised of individuals, affected by HSP or PLS or who have close ties to either disorder. Board members do not receive compensation for their involvement. The SPF Medical Advisor is John Fink, MD. He is the Director of the Neurogenetic Disorders Clinic and Professor at the University of Michigan and a leading investigator of HSP and PLS.

**What is #HSPandPLS?**

The hashtag (#) is used within a message to identify a keyword or topic of interest and facilitate a search for it. Whenever a user adds a hashtag to his/her post, it is able to be indexed by the social network and becomes searchable or discoverable by other users. Use, Share, and Search for #HSPandPLS to broaden awareness of Hereditary Spastic Paraplegia (HSP) and Primary Lateral Sclerosis (PLS).



**Symptoms**

One of the most noticeable symptoms is impaired walking, which may result in the use of a cane or walker. Ultimately, it may be easier to physically use a wheelchair, power chair or a scooter. In PLS, the disease also affects muscles in the upper body and may cause complications with the arms, voice, and swallowing. Adults and children with HSP or PLS can also suffer from great pain, balance disturbance, bladder and/or bowel issues. Some forms of more complicated spastic paraplegia can cause dementia, epilepsy, vision problems, skin problems and/or other neurological problems.

According to the National Institute of Neurological Disorders and Stroke, HSP (Hereditary also known as Familial Spastic Paraparesis or Paraplegia) is "a group of inherited disorders that are characterized by progressive weakness and stiffness of the legs". PLS, Primary Lateral Sclerosis is "a neuromuscular, motor neuron disease characterized by progressive muscle weakness in the voluntary muscles."



Both HSP and PLS are motor neuron diseases sharing a common symptom; weakness and spasticity (sudden, involuntary muscle spasms, or also known as clonus). HSP is a genetic disease that many times affects several members of the same family in which symptoms can occur at any age. Although hereditary, anyone may be the first to experience the genetic mutation in a family.

PLS symptoms usually begin in the legs, but may also start with the tongue or hands. Scientists believe PLS is not hereditary, but progresses gradually over a number of years or even decades. There is no evidence of the degeneration of spinal motor neurons or muscle wasting (amyotrophy) in PLS that occurs in amyotrophic lateral sclerosis/ALS (Lou Gehrig's disease). Diagnosis of PLS is often delayed because it is mistaken for ALS.

There are currently no cures available to prevent, stop, or reverse HSP or PLS. Current treatments are focused on symptom relief, such as medication to reduce spasticity; physical therapy and exercise to help maintain flexibility, strength, and range of motion; assistive devices and communication aids; and supportive therapy and other modalities.

**Education**

SPF needs your help as we continue to reach out to an increasing number of people struggling with HSP or PLS. We need to use a variety of tools and initiatives, including TeamWalks, Golfing For Rare Disease, events, support groups, conferences, and publications to bring awareness to HSP and PLS. SPF must continue to raise funds for the important medical scientific research and grow awareness in the community.

**What are SPF Ambassadors?**

SPF Ambassadors (SAs) are key volunteers that demonstrate a commitment to helping people in their area. Their involvement can be in multiple capacities with an interest in advocacy, education, awareness, support and fundraising for the SPF. More SAs are needed to help connect with local people, resources, and events. SAs welcome new members to better inform them and their communities about #HSPandPLS. If interested in representing an area, please reach out to one of the SPF Ambassador Co- Coordinators to help with outreach efforts.



**Is your Physician in our Directory?**

Finding a neurologist that understands #HSPandPLS may seem challenging. Help us spread the word about SPF and update our Physician Directory. Is your current doctor or neurologist listed in our Physician Directory? If not, let us know so we can add them to our database.

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