What in the World is Stiff Person Syndrome?

In the United States, a rare (or orphan) disease is defined by the National Organization of Rare Diseases (NORD) as a disorder affecting less than 200,000 Americans (approximately 1 in 1500 individuals).

According to the National Institutes of Health (NIH), there are nearly 6,800 rare diseases affecting more than 30 million people of all ages in the country. Nearly 1 in 10 Americans has one of these rare diseases.

One such rare disease is believed to affect only 1 in 1 million individuals, worldwide.

This orphan is known as Stiff Person Syndrome (SPS).

Although considered a Neurological disease it is often classified as an Autoimmune Disease, characterized by alternating rigidity and spasticity of the muscles, tremors, anxiety, and a hyper-excitability of muscles (known as the startle reflex). Emotional stress or even a gentle touch are known to cause prolonged, often severe, spasms.

The result of constant spasms is unrelenting pain and commonly, eventual disability. In middle to late stages of SPS, virtually no muscle is spared from spasm, and spasms can be violent enough to result in torn muscle tissue, and broken bones. Frequent falls are common among those with Stiff Person Syndrome, as they lack normal balance. These falls can be serious due to an inability to react, and muscles may become rigid during the crisis.

Average time to diagnose the disease is *7 years*, and misdiagnoses during this period include Anxiety or Adjustment Disorder, Phobia, Multiple Sclerosis, Dystonia, Fybromialgia, Parkinson's Disease, and Psychosomatic Illness.

It is discouraging that a lack of awareness of the disease is responsible for this prolonged delay in diagnosis.

The cause of SPS is unknown, and there is no cure on the immediate horizon.

It has been theorized that SPS may be caused by a virus in the brain and CSF which for an unknown reason went awry and introduced elevated levels of the auto-antibody, *glutamic acid decarboxylase* (GAD65), which prevents adequate production of *gamma-Aminobutyric acid* (GABA) in the brain, a crucial neurotransmitter and one of the body's natural anti-anxiety chemicals.
Diagnosis is most often made from a blood test which reveals even the slightest elevated level of GAD65.

Treatment, which first requires a proper diagnosis, can relieve the patient of some pain, spasticity, and anxiety. Progression of the disease can be slowed although rarely halted. Disability can be delayed, although once a patient has become wheelchair-bound it is doubtful the disability will be reversed.

Common treatments for Stiff Person Syndrome *symptoms* include:
- High doses of Valium (Diazepam) for anxiety and muscle spasms
- Baclofen, a muscle relaxant (often dispensed from an implanted pump, called Intrathecal Baclofen Pump)
- Neurontin (Gabapentin), a seizure medicine, and
- Intravenous Immunoglobulin (IVIg), made from thousands of healthy blood donors to help negate some of the effects of GAD antibodies, and contribute to the production of GABA.

These treatments will be required for the life of the patient, in varying doses and frequency.

Other Autoimmune Diseases, including Diabetes Mellitus and Thyroiditis, are commonly found in those with Stiff Person Syndrome. Depression is common and difficult to treat due to Neurological side effects from most Antidepressants, exacerbating SPS symptoms.

There is support on the Internet for those dealing with Stiff Person Syndrome. Among the sites are several facebook "communities", and forums dedicated to the disease.
SPS - Australia (Facebook Page)
SPS - Moersch & Woltman – Stiffman (Facebook Group)
Stiff Person Syndrome - The Official UK and Ireland Support (Facebook) Group
NEW! Official *Stiff Person Syndrome Association* Website

This page was created by a person with SPS, and should not be considered medical advice. "I am not a doctor. Nor do I play one on TV!"
It is simply, *and only*, a means of helping to raise awareness of Stiff Person Syndrome. Carl David Ericson – Las Vegas, NV, USA