

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 1,500 individuals). One such rare disease is believed to affect only 1 in 1,000,000 individuals, worldwide.

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

Although actually an Autoimmune Disease, it is most often treated by a Neurologist. It is characterized by rigidity and/or spasticity of the skeletal muscles, tremors, and anxiety. The result of constant spasms is unrelenting pain and commonly, eventual disability. As SPS progresses, virtually no muscle is spared from spasms, and these may be violent enough to result in torn muscle tissue or broken bones. Frequent falls are common among those with SPS, as they lack normal balance. These falls can be serious due to an inability to react, and muscles may become rigid during the crisis.

Unique to SPS is a hyper-excitability of the muscles, known as the **startle response**. Emotional stress, a sudden, unexpected noise, or even a gentle touch, will often trigger a Myoclonic Seizure of prolonged, severe spasms, and extreme rigidity.

Average time to diagnose the disease is 7 years, and misdiagnoses during this period include:

Anxiety (Conversion) Disorder, Phobia, Multiple Sclerosis, Dystonia, PTSD, Fybromialgia, Parkinson's Disease, and Psychosomatic Illness

It is discouraging that a lack of awareness of Stiff Person Syndrome is responsible for this prolonged delay in diagnosis.

The cause of SPS is unknown, and there is no cure. It has been theorized that SPS may be caused by a virus which enters the brain and CSF, and introduces elevated levels of the auto-antibody, glutamic acid decarboxylase (GAD65), preventing adequate production of gamma-Amino butyric acid (GABA) in the brain. This is 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, although ~35% will eventually be diagnosed without any elevation of GAD65.

There are treatments which can relieve the SPS patient of some pain, spasticity, and anxiety. These treatments may slow the progression of the disease but it is rarely halted, and disability can be delayed, although once one with SPS has become disabled, it is doubtful the disability will be reversed.

Common treatments for Stiff Person Syndrome **symptoms** include:

- High doses of Valium (Diazepam) and/or Ativan (Lorazepam) for anxiety and muscle spasms,
- Baclofen, a muscle relaxant,
- Neurontin (Gabapentin), a seizure medicine, and
- Intravenous Immunoglobulin (IVIg), (2G/Kg) helps negate some of the effects of GAD antibodies, and contribute to the production of GABA.

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

Other Autoimmune Diseases, including Diabetes Mellitus, Thyroiditis, Lupus, and Pernicious Anemia, 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.