NEUROPULSE VOLUME 10

APRIL 4TH, 2025

NEUROLOGY IN FOCUS

STIFF PERSON SYNDROME UNCOVERED

CUTTING EDGE RESEARCH

LIVING WITH SPS: TESTIMONIALS OF AN ARTIST

1 IN A MILLION

THE INVISIBLE ILLNESS YOU NEED TO KNOW ABOUT

CELINE DION

"SINGING WITH STIFF PERSON SYNDROME IS LIKE 'SOMEBODY'S STRANGLING YOU..."

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UNDERSTANDING

STIFF PERSONS SYNDROME

A RARE NEUROLOGICAL MYSTERY

Stiff Person Syndrome (SPS) is a rare neurological disorder, described in the 1950s by Moersch and Woltman.[1] SPS involves increasing muscle tightness and aching spasms that typically start from the torso and then spread toward the arms and legs.[2] For many decades, researchers have widely discovered that SPS is often associated with a specific autoimmune response where the body mistakenly attacks some components of its nervous system, especially enzymes such as glutamic acid decarboxylase (GAD), which are necessary for producing the inhibitory neurotransmitter GABA.[3] This link to autoimmunity improves our comprehension of the way the illness functions and influences how we attempt to handle it by changing the immune system.[2]

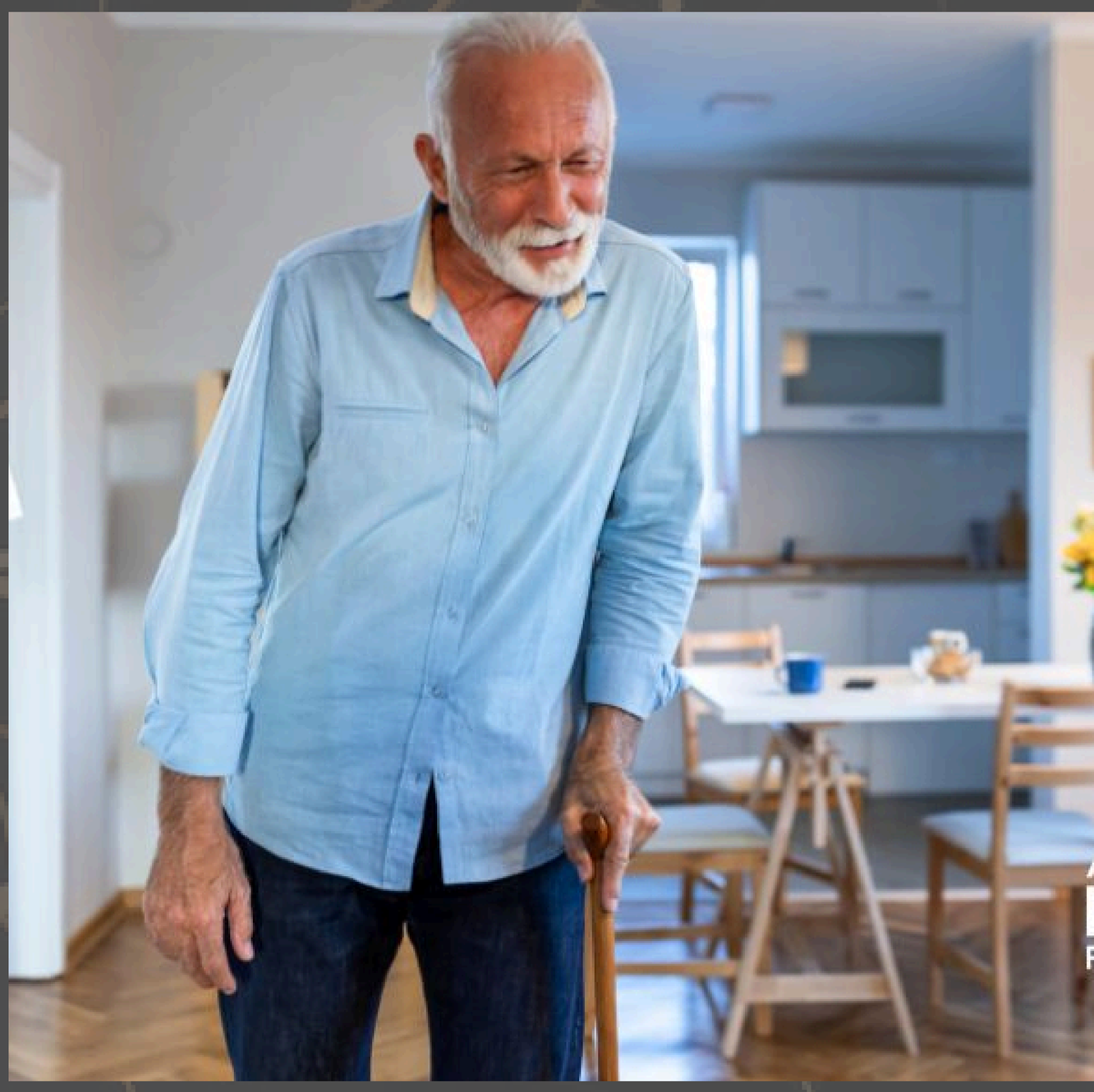
Formerly, SPS was frequently misdiagnosed as either a psychological or functional disorder as the early clinical presentations could resemble anxiety or psychosomatic illness. Today, a more complete comprehension of its immunological basis, with improvements in diagnostic testing, has improved recognition. Even though the condition remains under diagnosed, due to its large rarity. SPS has also been linked to autoimmune conditions such as type 1 diabetes and thyroid problems, which shows the importance of an exhaustive approach when examining people.[3]

1-2

ESTIMATED CASES PER MILLION OF CLASSIC SPS [4]

2x

THE NUMBER OF WOMEN ARE AFFECTED THAN MEN [5]



(AMERICAN BRAIN FOUNDATION, 2025)

SPS DID YOU KNOW?

SPS was originally called “stiff-man syndrome,” and then the name was later updated into “stiff person syndrome” for acknowledgment that the condition affects people of all genders. [6]

(YALE MEDICINE, 2025)

BEYOND THE SURFACE:

THE TELLTALE SIGNS OF SPS

People with SPS typically go through an inconsistent arrangement of muscle stiffness and convulsions. Common signs/symptoms consist of:

1 Persistent Muscle Stiffness

Typically starting in the torso (chest, back, and abdomen), the stiffness can ultimately move toward the limbs closer to the trunk.[2] Quite often, patients report that it feels as if their muscles are "locked in place," making it difficult to bend, turn, and even maintain a normal posture. [3]

2 Painful Muscle Spasms

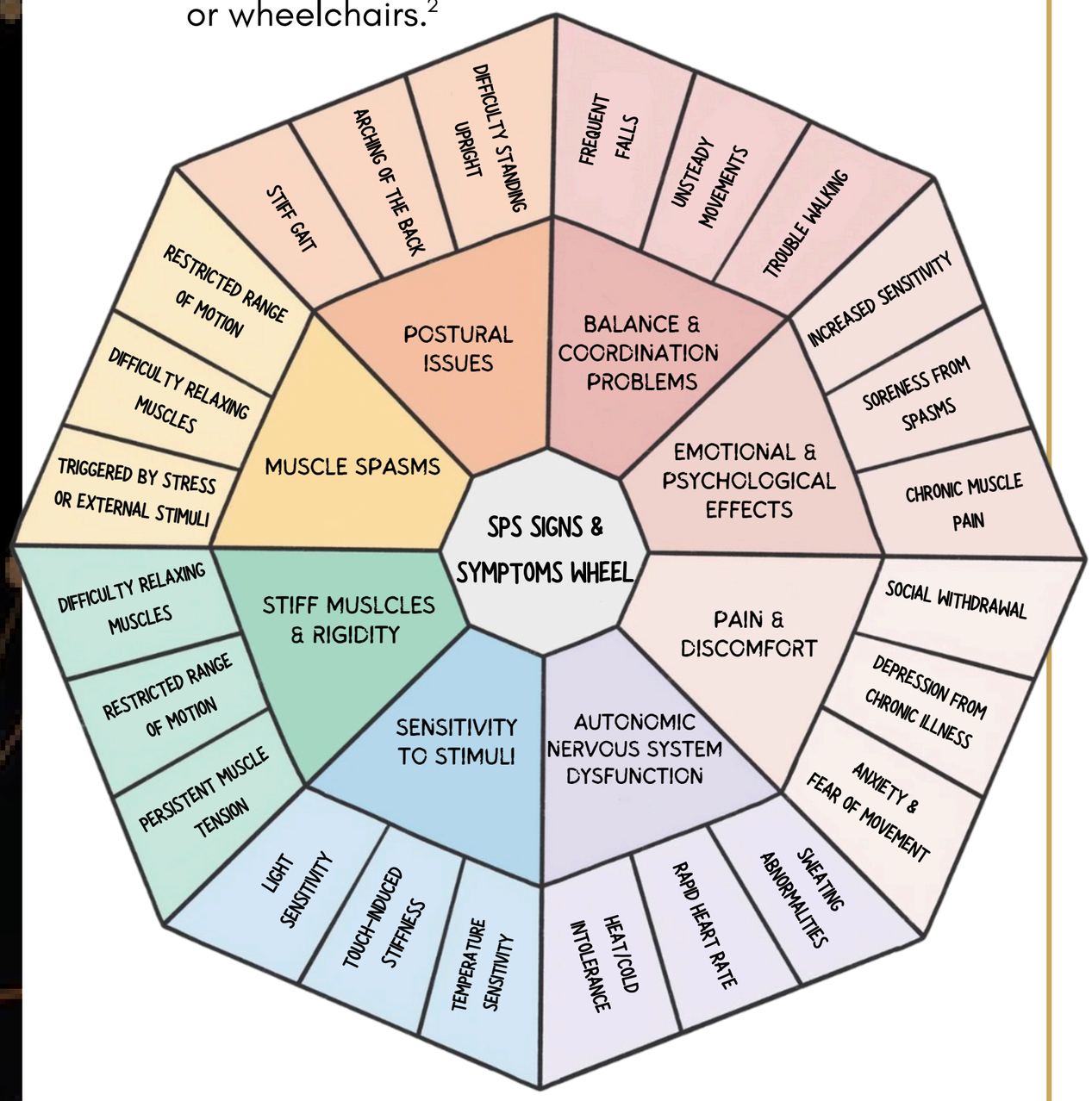
Spasms, unpredictable and sudden, can be triggered by unexpected noises, touch, and strong emotional stress. These contractions are not only uncomfortable, but can be quite intense to the point that they lead to multiple falls or wounds. Spasms may last from a few seconds to several minutes in some cases. Daily activities and general quality of life are greatly affected as a result.[7]

3 Associated Psychological Impact

Due to the unpredictable nature and the debilitating nature among the symptoms, many patients develop anxiety, depression, and even specific phobias.[6] These feelings are thought to be somewhat related to the lower amounts of GABA, which controls muscle movement and also affects how anxiety is managed.[8]

4 Functional Limitations

As SPS keeps advancing, people often have major issues with ambulation and could form very atypical stances (such as hyperlordosis—a considerably exaggerated curve of the lower spine).[9] Over a duration of time, the condition can lead to meaningful physical disability, infrequently necessitating mobility aids such as particular canes or wheelchairs.[2]

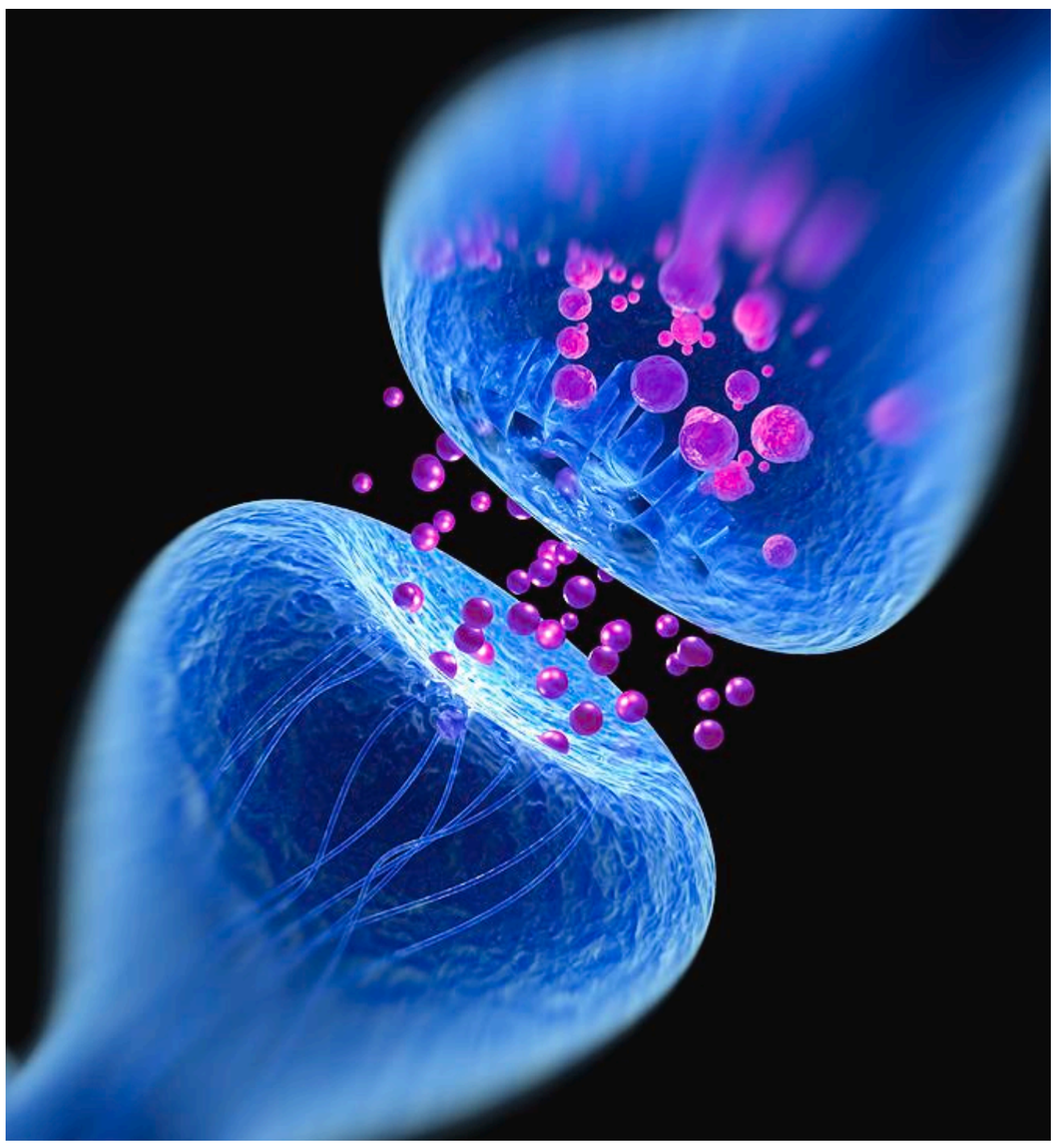


Did you know SPS can go beyond just muscle stiffness. This wheel displays the various primary and secondary effects that a person diagnosed with SPS may expereince throughout their daily life.

UNRAVELING THE MYSTERY:

THE ETIOLOGY OF STIFF PERSON SYNDROME

Stiff Person Syndrome (SPS) is rare and complex disorder, affecting approximately only 1 in 1,000,000 people. While the condition can develop at any age, symptoms typically emerge between the ages of 30 and 60. Notably, SPS disproportionately affects women, with a significantly higher prevalence compared to men.[4] While its clinical manifestations are well-documented, the exact etiology of SPS remains a subject of ongoing research. What scientists do know, however, points to an autoimmune origin, with the immune system mistakenly targeting the body's own nervous system.



At the heart of SPS is the disruption of gamma-aminobutyric acid (GABA) on pre-synaptic or post-synaptic neuronal junctions.[10] GABA is the brain's primary inhibitory neurotransmitter responsible for calming nerve activity. In individuals with SPS, autoantibodies—particularly those targeting glutamic acid decarboxylase (GAD)—are frequently detected. GAD65 is an enzyme crucial for the production of GABA, and its inhibition by these autoantibodies is believed to lead to a hyperexcitable state in the nervous system, resulting in the hallmark muscle stiffness and spasms.[11]

DID YOU KNOW?

35% OF PATIENTS DIAGNOSED WITH SPS HAVE DIABETES MELLITUS TYPE 1 & ~5% HAVE ASSOCIATED AUTOIMMUNE THYROID DISEASE[4]

Interestingly, SPS is often associated with other autoimmune conditions, such as type 1 diabetes, thyroiditis, and vitiligo, suggesting a shared underlying predisposition.[4] Additionally, some cases of SPS have been linked to autoantibodies targeting other proteins, such as amphiphysin and glycine receptors, further complicating the disease's etiology.[12] Beyond its autoimmune origins, paraneoplastic SPS also shows a strong association with cancer. Research indicates that breast cancer is the most prevalent malignancy linked to SPS, followed by lung cancer and lymphoma.[13] In these cases, the immune system mistakenly attacks the nervous system cells, potentially causing symptoms before a tumor is discovered. These findings highlight the heterogeneity of SPS and the likelihood of multiple pathways contributing to its development.

While the autoimmune component is well-established, the triggers for this immune dysregulation remain elusive. Genetic predisposition, environmental factors, and viral infections have all been proposed as potential contributors. For instance, genetic predisposition has been traced to specific major histocompatibility complex (MHC) class II alleles, particularly DQB1 and DRB1, increasing the risk of both idiopathic and paraneoplastic variants of SPS.[4] Additionally, certain human leukocyte antigen (HLA) genotypes have been associated with an increased risk of developing SPS, pointing to a genetic vulnerability. Meanwhile, viral infections, such as those caused by the Epstein-Barr virus, have been hypothesized to initiate or exacerbate the autoimmune response in susceptible individuals.[12]

Despite these advances, many questions remain unanswered. As researchers continue to unravel the intricate mechanisms behind SPS, the hope is that a deeper understanding of its etiology will pave the way for more targeted and effective treatments, offering relief to those living with this debilitating condition.

Pathophysiology

The development of SPS is driven by B-cell-mediated autoimmune inflammation, which disrupts inhibitory GABAergic neurons and their synapses.[14] Autoantibodies targeting GABA-related antigens, specifically GAD, interfere with inhibitory signaling, leading to motor cortex hyperexcitability and the inability to relax truncal and axial muscles. Anti-GAD65 antibodies are a key marker of classic SPS. They appear in 70-80% of cases, and are also associated with other autoimmune neurological disorders, known in the medical community as GAD antibody-spectrum disorders (GAD-SD). Titers refer to the concentration of specific antibodies (anti-GAD) present in a person's blood and are measured through laboratory tests to assess immune response or diagnose autoimmune and infectious diseases. While low anti-GAD titers are seen in type 1 diabetes, high titers are specifically linked to GAD-SD. Other antigens involved in SPS include GABA(A) receptor-associated protein (GABARAP), dipeptidyl-peptidase-like protein-6 (DPPX), and glycine receptor (GlyR). A paraneoplastic variant of SPS is associated with antibodies against amphiphysin or gephyrin, which disrupt GABA signaling by depleting presynaptic vesicle pools and reducing GABA receptor expression.[4]

RESILENCE

Emily Carter's Story of Strength and Survival

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In this exclusive interview, we sit down with Emily Carter, a 38-year-old artist, and author of "Written in the Wind" from Portland, Oregon, who has been living with Stiff Person Syndrome for nearly a decade. She shares her journey of resilience, the challenges she faces daily, and her hopes for the future.

Q: Emily, can you tell us a bit about when and how you were first diagnosed with SPS?

Emily Carter: Absolutely. It started in my late twenties with occasional muscle stiffness, mostly in my lower back. At first, I thought it was just stress or poor posture from long hours painting and writing. But over time, the stiffness became more intense and unpredictable. Eventually, I began experiencing painful muscle spasms that left me temporarily immobile. I saw multiple doctors who initially misdiagnosed me with anxiety, fibromyalgia, and even multiple sclerosis. It wasn't until a neurologist ran a series of tests, including a GAD antibody test, that I finally got the correct diagnosis—Stiff Person Syndrome. That was in 2015.

Q: What was your reaction to the diagnosis?

Emily Carter: Honestly, it was a mix of relief and fear. Relief, because I finally had an explanation for my symptoms and knew I wasn't imagining things. Fear, because I had never heard of SPS before, and when I started researching, I realized how rare and complex it is. I had to come to terms with the fact that this was a chronic, progressive condition with no known cure.

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Q: What kind of treatments or therapies have helped you manage your symptoms?

Emily Carter: My treatment plan includes a combination of medication, physical therapy, and lifestyle adjustments. I take muscle relaxants and GABA-enhancing medications to help control the spasms. IVIG therapy has also been beneficial in reducing the severity of my symptoms, although access to it can be challenging due to cost and availability.

I also do gentle stretching and yoga when I can, but I have to be careful not to overdo it. Mindfulness and meditation have been incredibly helpful in managing stress, which is a huge trigger for me. And, of course, I've had to learn to listen to my body and rest when I need to.

Q: What has been the most difficult part of living with SPS?

Emily Carter: The unpredictability of it. I think that's what makes chronic illness so mentally and emotionally exhausting. One day I might feel strong enough to go for a short walk, and the next, I might be unable to leave my bed. It's also tough dealing with the lack of awareness about SPS, even within the medical community. I've had emergency room visits where doctors didn't know what SPS was, which is terrifying when you're in crisis and need immediate help.

Q: How does SPS affect your daily life?

Emily Carter: Every day is different. Some mornings, I wake up feeling almost normal, while other days, I struggle just to move. The muscle stiffness can be so severe that simple tasks—getting dressed, cooking, even holding a pen—become challenging. The spasms are unpredictable and can be triggered by stress, cold temperatures, or even loud noises. It's frustrating because I never know when my body will betray me.

I've also had to adjust my social life. I used to be an extrovert, always attending gallery openings and poetry readings, but now I have to plan everything around my physical limits. I use mobility aids when needed, and I rely on a strong support system of family and friends who understand that I might have to cancel plans at the last minute.

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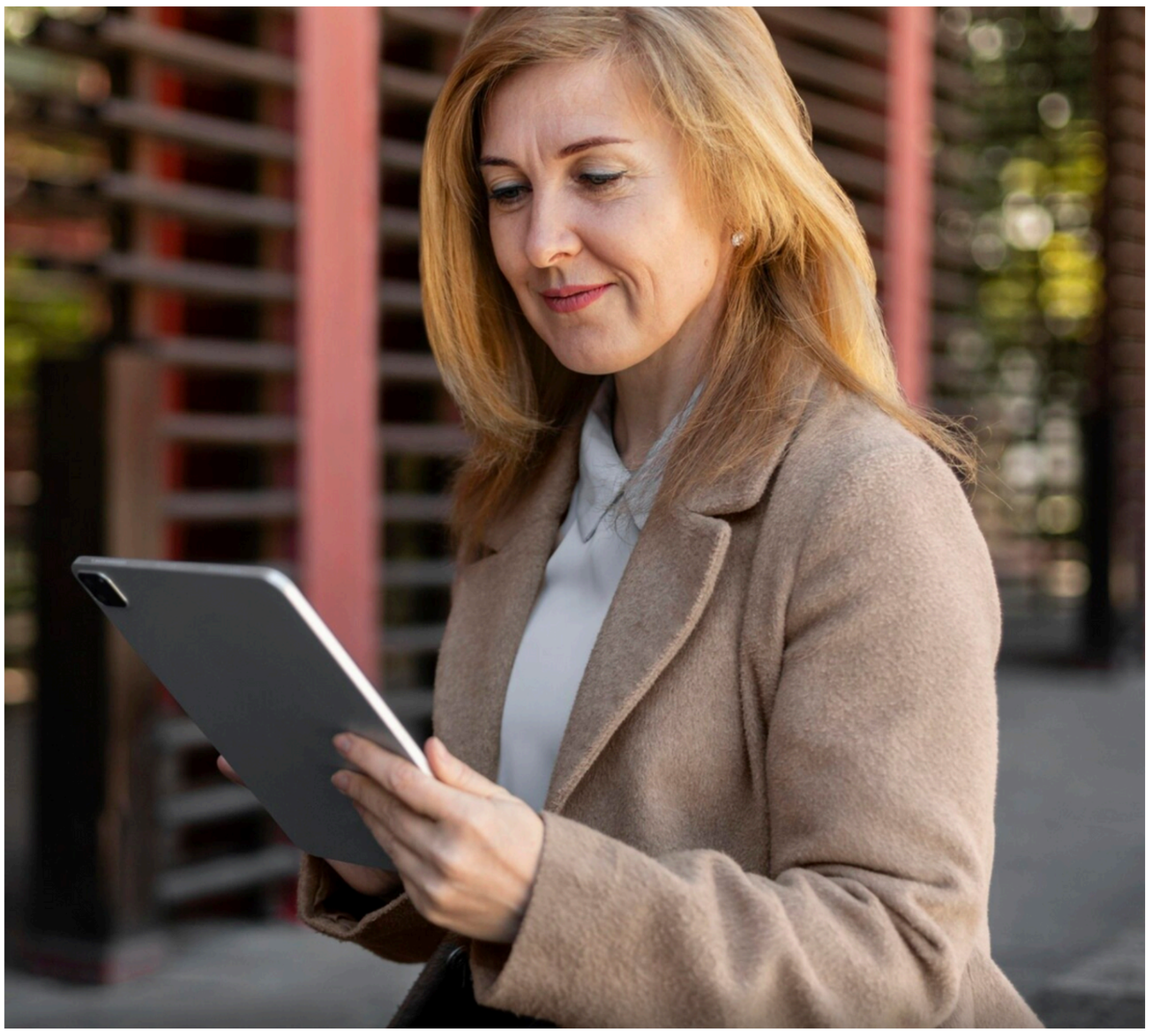
Q: How did your experience living with SPS influence your bestselling novel "Written in the Wind" ?

Emily Carter: Writing "Written in the Wind" was deeply personal for me. Living with SPS gave me a unique perspective on vulnerability, control, and resilience—all themes that are woven into the story. The protagonist's struggles with her own limitations and the unpredictability of life mirrored what I was facing physically and emotionally. In many ways, writing the novel became a form of therapy, allowing me to process my journey and turn something challenging into something creative and meaningful. It also allowed me to subtly raise awareness about invisible illnesses without making it the focal point, which I think resonated with readers.



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Q: What keeps you motivated and hopeful?

Emily Carter: My art, my writing, and my loved ones. I've learned to adapt my creative process—on bad days, I might sketch in bed or dictate poetry instead of typing. Expressing myself through art gives me a sense of purpose and control in a body that often feels uncooperative.

I also find hope in advocacy and connecting with others in the SPS community. Sharing my experiences, raising awareness, and educating people about this condition makes me feel like I'm making a difference. I want others with SPS to know they're not alone.

Q: What do you wish more people knew about Stiff Person Syndrome?

Emily Carter: That it's real, it's debilitating, and it's not just 'a little muscle stiffness.' It's a neurological disorder that affects every aspect of my life. I wish there was more research, more awareness, and more compassion for those of us living with rare diseases. Just because something is invisible doesn't mean it isn't life-altering.

Q: Finally, what message would you like to share with others who are newly diagnosed with SPS?

Emily Carter: Be patient with yourself. This is a marathon, not a sprint. Educate yourself, advocate for your needs, and surround yourself with people who support you. Some days will be incredibly hard, but there will also be moments of joy and triumph. You are more than your diagnosis. Keep fighting, keep hoping, and never be afraid to ask for help.

AN SPS PATIENT JOURNEY TOWARDS A DIAGNOSIS

Stiff Person Syndrome is a rare neurological disorder that often gets misdiagnosed due to symptom overlap with other conditions. Diagnosis is a multi-step process through clinical evaluation, lab tests and specialist consultations. Due to it's rarity, patients often endure protracted delays in diagnosis, seeing multiple specialists before reaching the correct one.

START OF SYMPTOMS

Patients seek answers for unexplained rigidity or "anxiety-like" symptoms

ONLINE RESEARCH & INITIAL CONCERNS

FIRST DOCTOR VISIT

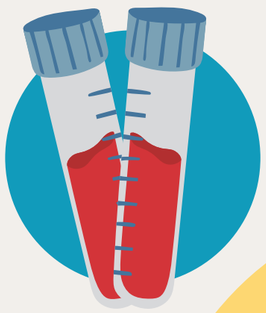
Primary Care or Neurologist: Initial assessment, basic reflex tests, blood work (e.g., CBC, metabolic panel)

BASIC TESTING & INITIAL WORKUP

Blood Work: Thyroid function, vitamin deficiencies.
Reflex Tests: Hyperactive reflexes noted.

SPECIALIST REFERRAL

Neurologist/Immunologist:
Suspected autoimmune or movement disorder.



KEY DIAGNOSTIC TESTS

ANTI-GAD ANTIBODY TEST
Elevated levels (>90% specificity for SPS)

ELECTROMYOGRAPHY (EMG)
Measures abnormal muscle activity and hyperexcitability.

LUMBAR PUNCTURE (SPINAL TAP)
Checks cerebrospinal fluid for immune markers

MRI/CT SCAN
Rules out other neurological conditions

CORRECT DIAGNOSIS

FINAL DIAGNOSIS & TREATMENT PLAN

Subtype Confirmation, Treatment, Interdisciplinary Team

RETESTING

NO DIAGNOSIS

MIS DIAGNOSIS

TEST RESULTS

PATIENT SUPPORT & MANAGMENT

SPS Foundation, Global Autoimmune Institute & Physical therapy for mobility, counselling for anxiety/depression

A Deeper look into the
DIAGNOSIS JOURNEY

Diagnosing Stiff-Person Syndrome (SPS) is a path fraught with uncertainty, mistakes and persistence. For patients struggling with inexplicable stiffness, spasms and fear of the unknown, the road to answers often rests on a few critical medical tests. These tests which include everything from blood work to advanced imaging are not just diagnostic tools; they are lifelines that separate SPS from mimics like multiple sclerosis or anxiety disorders.

We turn to the science behind the key investigations in this exploration:

ANTIBODY BLOOD TESTS:

Approximately 70-80% of patients with SPS possess particular antibodies (such as anti-GAD65) at levels greatly exceeding those discovered in other conditions, like type 1 diabetes. One may also detect additional antibodies, such as those directed toward amphiphysin or glycine receptors, especially throughout paraneoplastic variations or instances exhibiting progressive encephalomyelitis alongside rigidity and myoclonus (PERM).[4]

ELECTROMYOGRAPHY (EMG):

EMG is an important diagnostic tool for SPS. It is tested through electromyography, which assesses the electrical activity of muscles at rest and during voluntary contractions. In SPS, EMG usually demonstrates continuous motor unit activity, even during attempted muscle relaxation, which is a characteristic of this syndrome. There is also frequently co-contraction of agonist and antagonist muscles which cements the diagnosis.[4]

Tip: Consider the EMG findings as almost an overactive "alarm system" in the muscles so that even in a state of attempted relaxation, the muscles are constantly "on."

CEREBROSPINAL FLUID (CSF) ANALYSIS:

Even though several CSF results throughout typical SPS are frequently normal, the existence of many anti-GAD antibodies within the CSF may support a diagnosis, especially if serum quantities are weak or uncertain.[15][15]

(UT Southwestern Medical Center, n.d.).

MRI AND OTHER IMAGING MODALITIES:

Although the formal exclusion of other causes for stiffness (e.g. multiple sclerosis, spinal cord compression, neoplastic processes) involves magnetic resonance imaging (MRI) of the brain and spinal cord, it is not diagnostic for SPS. On rare occasion particularly in patients with the PERM variant MRI may show subtle changes in the brainstem itself or spinal cord.

Note: Although imaging is primarily a "rule-out" tool rather than a diagnostic test for SPS.[15]

DIAGNOSTIC CRITERIA

1. "Stiffness in the limb and axial muscles, prominent in the abdomen and thoracolumbar region" [2]
2. "Painful spasms precipitated by unexpected tactile and auditory stimuli" [2]
3. "Evidence of the continuous motor unit activity in agonist and antagonist muscles demonstrated by EMG" [2]
4. "Absence of other neurological impairments that could support an alternative diagnosis" [2]
5. "Positive serology for anti-GAD65 or anti-amphiphysin autoantibodies" [2]
6. "Clinical response to therapy with benzodiazepines" [2]

Clinical Classifications of Stiff Person Syndrome

Understanding the Variants

Stiff Person Syndrome (SPS) is not a one-size-fits-all diagnosis. SPS is a rare neurological disorder with several distinct forms with different challenges and symptoms. Athough the hallmark of SPS is muscle rigidity and spasms, the way it affects the body can differ.

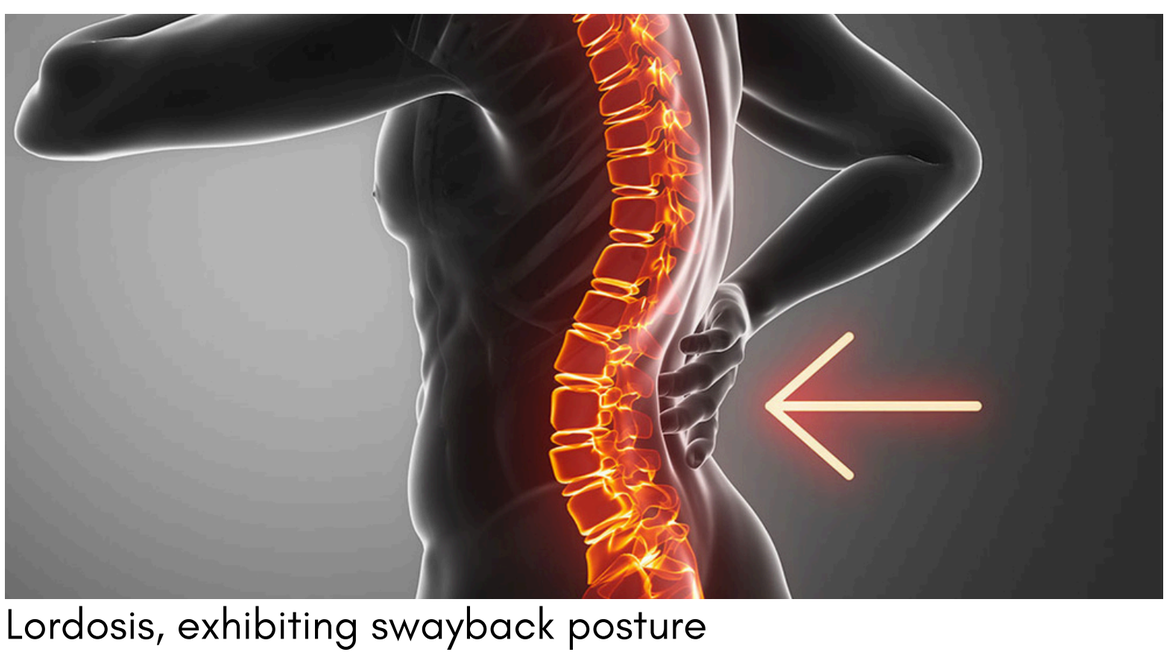
Let's explore the **three main clinical classifications** of SPS:

1. CLASSIC SPS - THE MOST COMMON FORM

This classification of SPS is what most people associate with the disorder. It is a progressive neurological condition where patients often have severe muscle spasms[17], which can be painful and debilitating. These spasms often start in the trunk and then spread to the[17] limbs, causing daily movement and balance difficulty.

In the early stages of classic SPS, patients may have periodic muscle rigidity, which gradually worsens over time. As this condition develops, the muscles become more stiff, and a condition known as *lordosis* may develop, where the patient develops an excessive swayback posture.[17] This condition can also progress into permanent physical deformities like joint deformities, leading to a "statue-like" appearance.[4]

Unfortunately, if no interventions are being made, this condition can become life-threatening, as it can impair breathing or lead to fractures and dislocations, which is why early diagnosis and intervention are essential for symptom management in classical SPS.[17]



Lordosis, exhibiting swayback posture

2. PARTIAL SPS

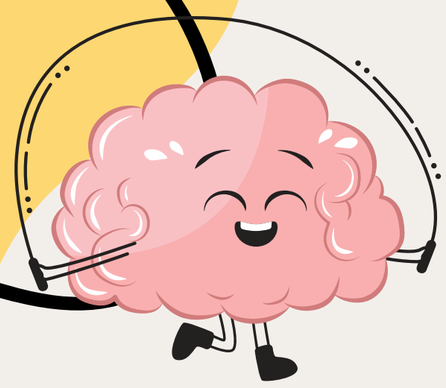
Not all cases of SPS affect the entire body. *Partial SPS* targets only specific regions of the body rather than spreading across the whole body. This type of SPS often affects one limb or just the trunk muscles.

There are several types within partial SPS that often relate to the affected region, such as *stiff limb syndrome*, where spasms occur only in the affected limb, with very minimal spasms in the trunk muscles.[4]

3. PROGRESSIVE ENCEPHALOMYELITIS WITH RIGIDITY AND MYOCLONUS (PERM)

This is a more severe form of SPS, which includes more widespread symptoms. This could entail classic rigidity with severe myoclonus (involuntary muscle jerks) and autonomic instability,[18] making it more dangerous and severe than any other forms of SPS.

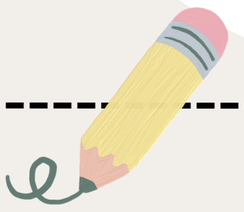
PERM can also affect the central nervous system (CNS) in addition to the muscles and is often seen in patients with anti-GAD antibodies. This variant may involve altered consciousness, muscle dysfunction in the brainstem,[4] and even issues with eye movements and autonomic functions like blood pressure regulation.[17]



CHALLENGE CORNER

TEST YOUR KNOWLEDGE!

MATCHING



A Midpoint Check in!

COLUMN A: DESCRIPTIONS

1. A blood marker found in ~80% of SPS cases, indicating an autoimmune attack on the nervous system.

2. A neurological test that detects continuous muscle activity, a key feature of SPS.

3. The most common form of SPS, affecting the trunk and limbs, causing severe stiffness and painful spasms.

4. A rare, aggressive variant of SPS associated with brain inflammation, leading to rigidity, seizures, and cognitive issues.

5. A localized form of SPS, affecting only one part of the body, such as the legs or torso.

6. A cancer-associated form of SPS, often linked to tumors (e.g., small-cell lung cancer, breast cancer).

7. An exaggerated muscle spasm response to sudden noise, touch, or stress, common in SPS patients.

8. A pronounced inward curve of the lower back, often seen in SPS due to chronic muscle contractions.

9. An increased sensitivity to external stimuli, such as loud noises, bright lights, or physical touch, which can trigger spasms.

10. A condition where the immune system mistakenly attacks healthy cells, leading to diseases like SPS.

COLUMN B: TERMS

A.Hyperlordosis

B.Electromyography (EMG)

C.Startle Reflex

D.Autoimmune Disorder

E.Focal Stiff Person Syndrome

F.Progressive Encephalomyelitis with Rigidity and Myoclonus (PERM)

G.Sensory Sensitivity

H.GAD65 Antibodies

I.Paraneoplastic Stiff Person Syndrome

J.Classic Stiff Person Syndrome (SPS)

HOW DID YOU DO?

9-10 CORRECT: SPS EXPERT!

6-8 CORRECT: GREAT EFFORT! KEEP LEARNING

0-5 CORRECT: TIME TO REVIEW!

ANSWERS

MATCHING:
1 → H, 2 → B, 3 → J, 4 → F, 5 → E, 6 → I, 7 → C, 8 → A, 9 → G, 10 → D

Stiff Person Syndrome

TREATMENTS

CURRENTLY, THERE IS NO CURE FOR SPS. MUCH OF THE CARE AND MEDICATIONS PRECRIBED TO PATIENTS AIM TO RELIEVE THE SYMPTOMS AND MAINTAIN MOBILITY.

MUSCLE RELAXERS

Drugs that act to relieve muscle spasms or tension by directly targeting the CNS and skeletal muscle.

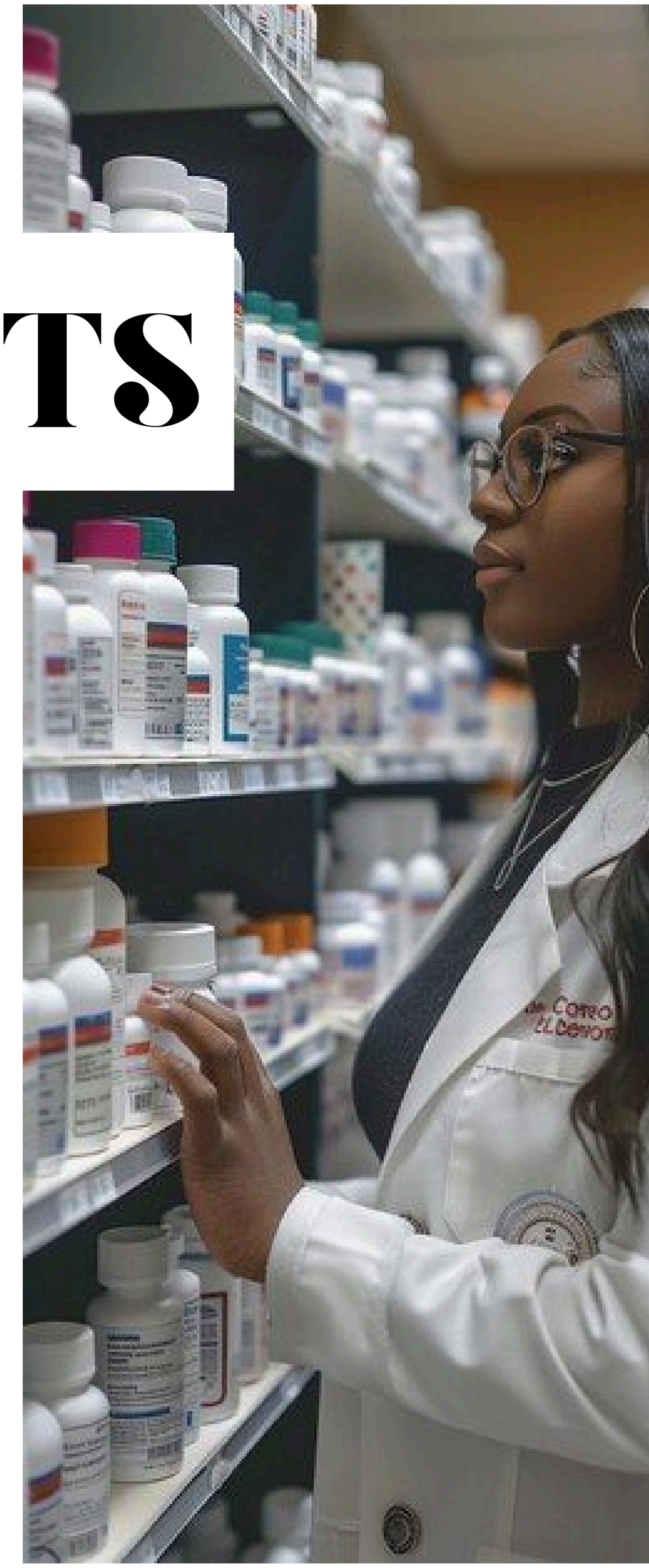
Diazepam & Clonazepam: Drugs in the benzodiazepine class that work by enhancing GABA receptor affinity. Typically used as an initial treatment for SPS. (19)
Baclofen: A GABA receptor agonist, which helps inhibit excitatory signals in the spinal cord, and reducing muscle stiffness. It can be taken orally or administered intrathecally. Used if benzodiazepines fails to control the symptoms. (19)
Tizanidine - An α2-adrenergic agonist, which reduces muscle spasms by impairing release of excitatory neurotransmitter at the spinal cord level. (19)

NON-MUSCLE RELAXERS

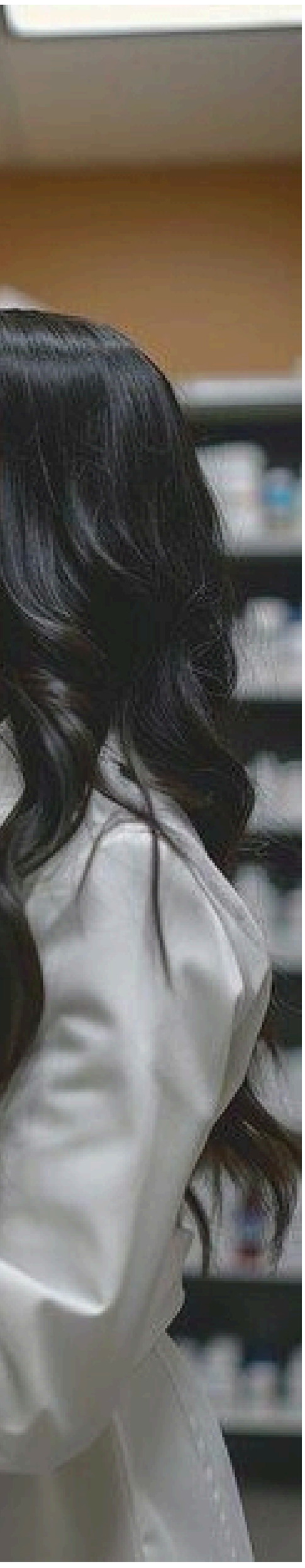
Drugs that are targeted to relieve inflammation and pain but not directly targeting the CNS.

Gabapentin & Pregabalin: These drugs are calcium channel modulators that reduce abnormal neuronal excitability and help manage neuropathic pain. An anticonvulsant (20).
Tiagabine: A GABA re-uptake inhibitor, inhibiting a GABA transporter (GT-1) and which increasing concentration in the nervous system, and prolonging synaptic inhibition (21).

Botulinum toxin: Botox can be injected into specific muscles that are particularly stiff or spastic. It blocks the release of acetylcholine at the neuromuscular junction, preventing excessive muscle contractions (22).



"AI IMAGE GENERATED FROM FREEPIK “PORTRAIT OF FEMALE PHARMACIST WORKING IN THE DRUGSTORE” .



“THERE IS NO CURE.”

IVIG consists of pooled antibodies from healthy donors, administered intravenously to modulate the immune system. It works by neutralizing autoantibodies against GAD, reducing inflammation and immune overactivity inhibiting immune cell-mediated damage (23).

IMMUNOTHERAPY

Intravenous Immunosuppressant Therapy

Rituximab (anti-CD20 monoclonal antibody): Targets and depletes B-cells, which are responsible for producing autoantibodies, including anti-GAD antibodies in SPS (24).

Oral Immunosuppressant Therapy:

Mycophenolate mofetil: Inhibits purine synthesis, suppressing T and B lymphocyte proliferation, reducing autoantibody production (24).

Azathioprine: A purine analog that leads to a blockage in purine synthesis. Interferes with DNA synthesis in rapidly dividing immune cells, reducing immune-mediated damage (2).

Plasmapheresis:

Therapeutic plasma exchange involves removing a patient’s plasma using membrane filtration (containing autoantibodies against GAD and other immune components) and replacing it with donor plasma or albumin. This helps reduce the autoimmune attack on the nervous system.(Rossi et al., 2010)

SUPPORTIVE THERAPY

Physical Therapy:

Helps maintain mobility and prevent muscle contractures. Stretching and relaxation techniques can help patients manage discomfort, muscle spasms and pain. Aqua therapy, occuring the pool, is also beneficial as it relieves stress on joints and bones (15).

Cognitive Behavioural Therapy (CBT)

Psychological therapy that can help patients address depression and emotional distress that may come with having SPS. Working on relaxation techniques can help patients manage stress-induced muscle spasms (15).

A prescription pill bottle spilling out an assortment of pills by Burlingham

Céline Dion's Battle with Stiff Person Syndrome

Canadian Global Icon Raising Awareness

CELINE DION'S BRAVE JOURNEY

Celine Dion, an iconic Canadian singer known for her powerful voice and international hits, made headlines in 2022 when she shared to the world about her battle with Stiff Person Syndrome (SPS). In a heartfelt video message to her fans, she revealed her diagnosis of SPS, a rare neurological condition that significantly impacted her daily life.[25]

For Dion, the battle has been challenging, as the disorder affects not only her daily activities but also her ability to perform and sing, which is the very thing that brought her worldwide fame and success.

In her emotional announcement, Dion expressed the physical toll on her body she has experienced with SPS.[25]

"My voice was struggling, I was starting to push a little bit."

She spoke of how the condition caused her muscle spasms, making it difficult for her to walk and use her vocal cords.[25] Dion, who has always been known for her work ethic and passion for performing, initially refused to stop her ongoing tour despite her symptoms.[25]

"I needed to find a way to be on stage,"

However, after her official diagnosis of SPS, she had to make the heartbreaking decision of pausing her tour to focus on her health.[25]

After Dion's announcement, she was met with an outpouring of support from her fans and the public. After sharing her diagnosis, Dion has become one of the most prominent figures raising awareness for SPS, a condition that has never heard of before by many.[25] Her courage in opening up about her struggles not only highlights the challenges of living with SPS but also brings light to a disorder that often remains in the dark due to how rare it is.

Since her diagnosis, Dion has focused on treatments and therapies to help manage her symptoms of SPS. She has also been vocal about the importance of early diagnosis and continued research into SPS through her documentary with Amazon Prime. Through her transparency about her experience with SPS, Celine Dion has helped break down the stigma around rare conditions such as SPS and shown that even global superstars are not immune to health struggles. With her advocacy, Dion is not just a global singer, but also a global voice for those who often go unheard.

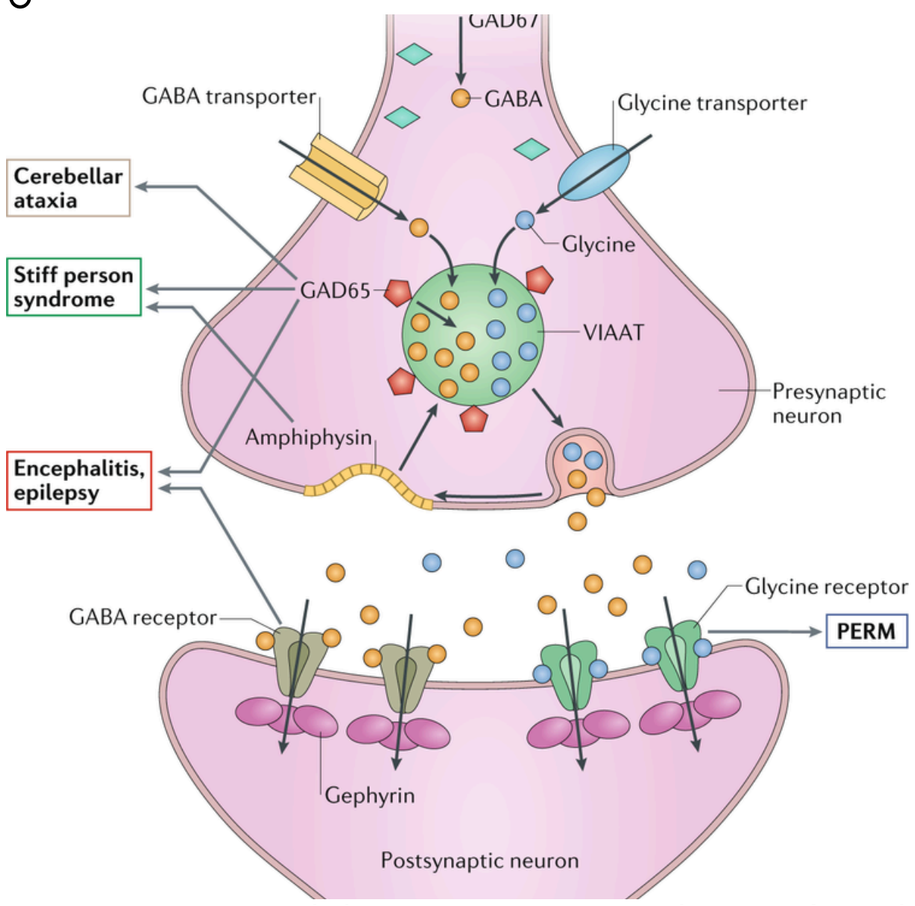
The Path Forward for Stiff Person Syndrome

New Research and Future Findings

While Stiff Person Syndrome (SPS) is a rare condition that is difficult to understand, recent research is shedding light on new treatment options and the disease's underlying mechanisms. Scientists are making efforts to understand how SPS develops and how to manage it more effectively, offering hope for both patients and researchers.

UNRAVELING THE MYSTERY OF ANTI-GAD ANTIBODIES

One of the most debated topics in SPS research is the role of anti-glutamic acid decarboxylase (anti-GAD) antibodies. While these antibodies are commonly found in patients with SPS, we are still unsure whether there is a direct causal relationship between the disorder and anti-GAD antibodies or if it is just a sign of a broader immune dysregulation.[26] Some recent findings suggest that these antibodies might not be as pathogenic as we thought, suggesting that the immune system's response could be more complicated than we thought.[26]

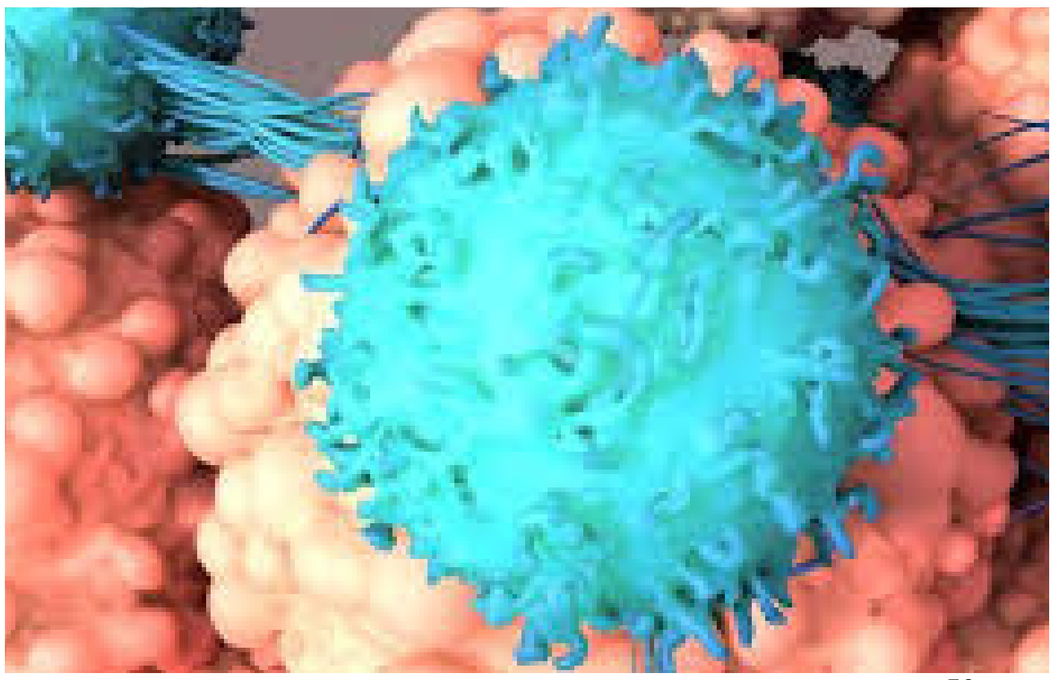


Autoimmune targets in inhibitory synapse with correlated neurological disorders including SPS[27]

NOVEL THERAPIES: THE FUTURE IS BRIGHT

There have recently been new therapies on the horizon. *Efgartrigimod*, a new drug targeting a receptor important for the homeostasis of IgG (FcRn), has shown some promising results in reducing the levels of harmful antibodies in the body.[28] Additionally, therapies like *anti-CD20/anti-CD19 therapies* are being explored for their potential to help patients with severe, treatment-resistant SPS.[29]

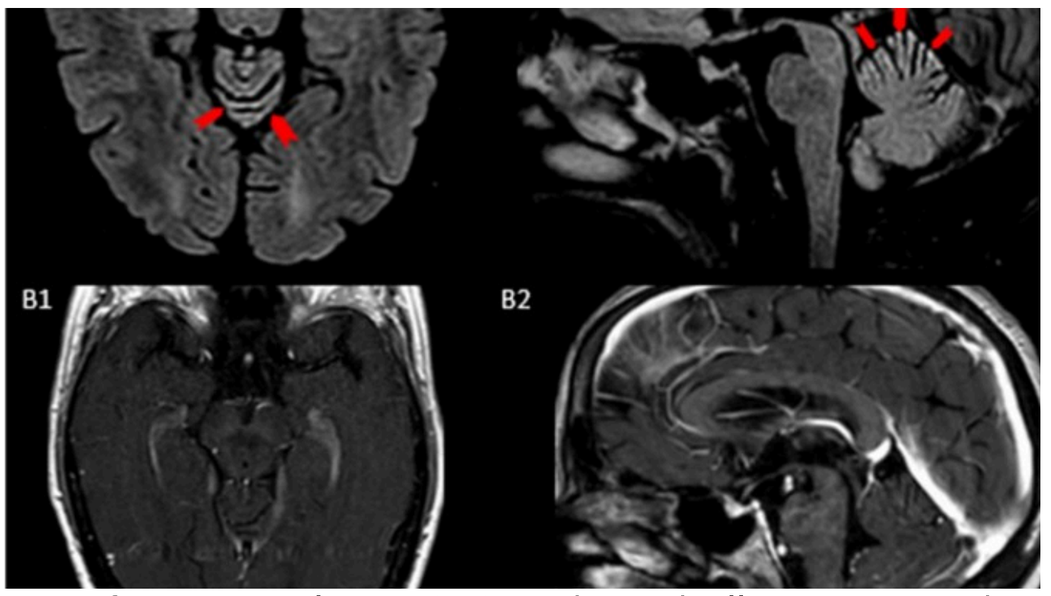
The most exciting news for SPS therapy is the rise of CD19-CAR T cell therapy, which uses the body's immune cells to target and eliminate disease-causing antibodies.[26] Although this therapy is still in its early experimental stage, it has shown some promising results for patients whose SPS does not respond to the usual treatments we have so far,[26] offering hope for those who need better options.



KYV-101, an anti-CD19-CAR T-cell therapy[30]

FUTURE FOCUSES

As research advances, researching genetic variations and their role in the immune response has become more important than ever, especially in cases like paraneoplastic SPS, where cancer triggers autoimmune reactions of SPS.[31] Researchers are also exploring more broader implications of nerve-muscle communication disorders, which could provide more insight into better treatments for SPS.[31]

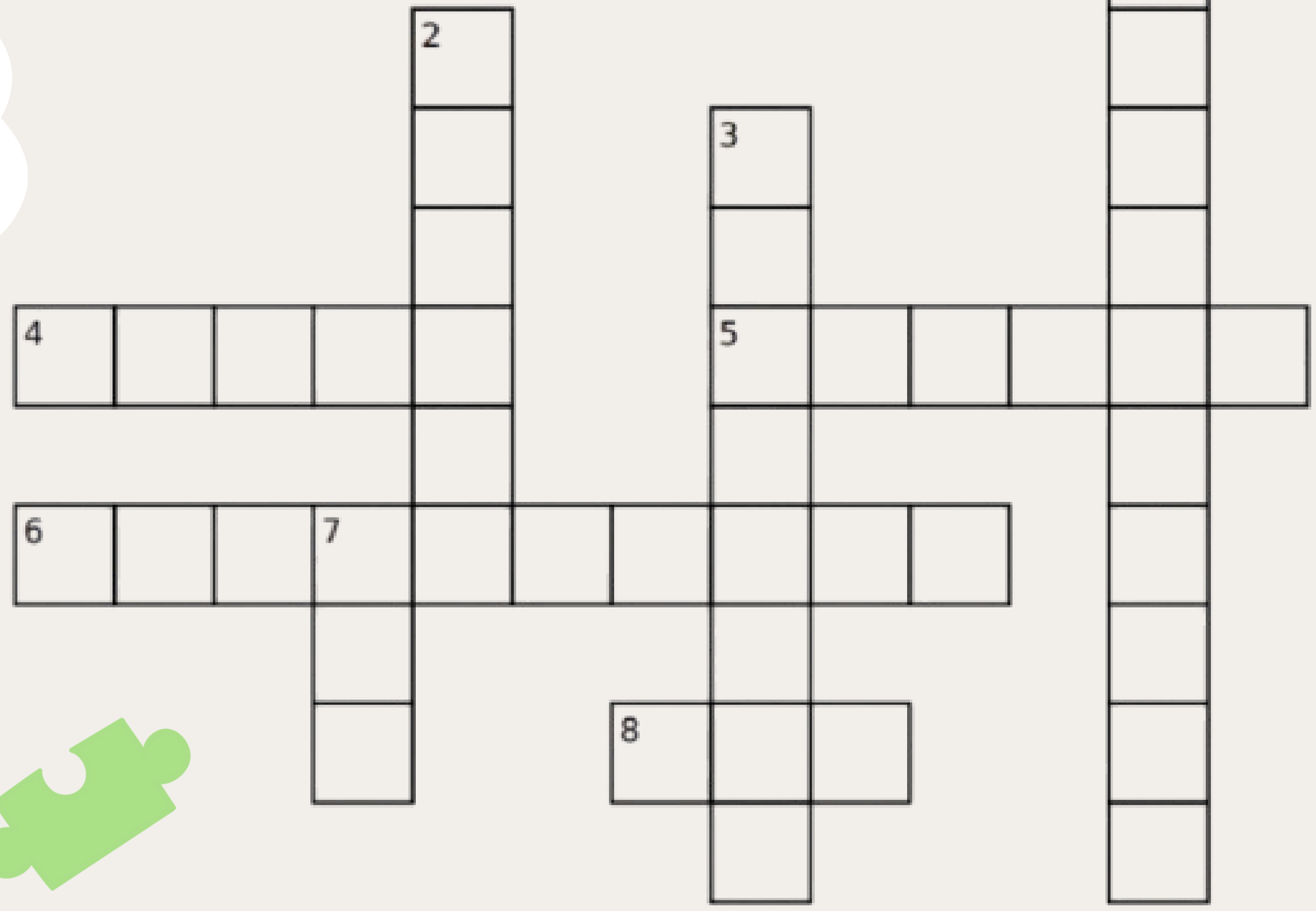


MRI of paraneoplastic SPS and cerebellar ataxia with anti-GAD65 abs and thymoma[32]

PUZZLER

DO YOU KNOW SPS VOCABULARY

DON'T FOGERT TO CHECKE YOUR ANSWERS BELOW TO SEE HOW WELL YOU DID !



ACROSS

1. Hallmark symptom of SPS, causing stiffness and spasms.
2. Chronic discomfort often experienced by SPS patients.
3. Hyperactive _____es may be seen in neurological exams for SPS.
7. Imaging scan used to rule out other causes of stiffness.

DOWN

4. Medical specialty that diagnoses and treats conditions like SPS.
5. Involuntary muscle contractions that may co-occur with SPS.
6. Autoantibody target in SPS.
8. Common immunotherapy for SPS, given intravenously.

ANSWERS:

Down:
1. Neurology
2. Dystonia
3. GAD
7. IVIG

Across:
4. Rigidity
5. Pain
6. Reflex
8. MRI

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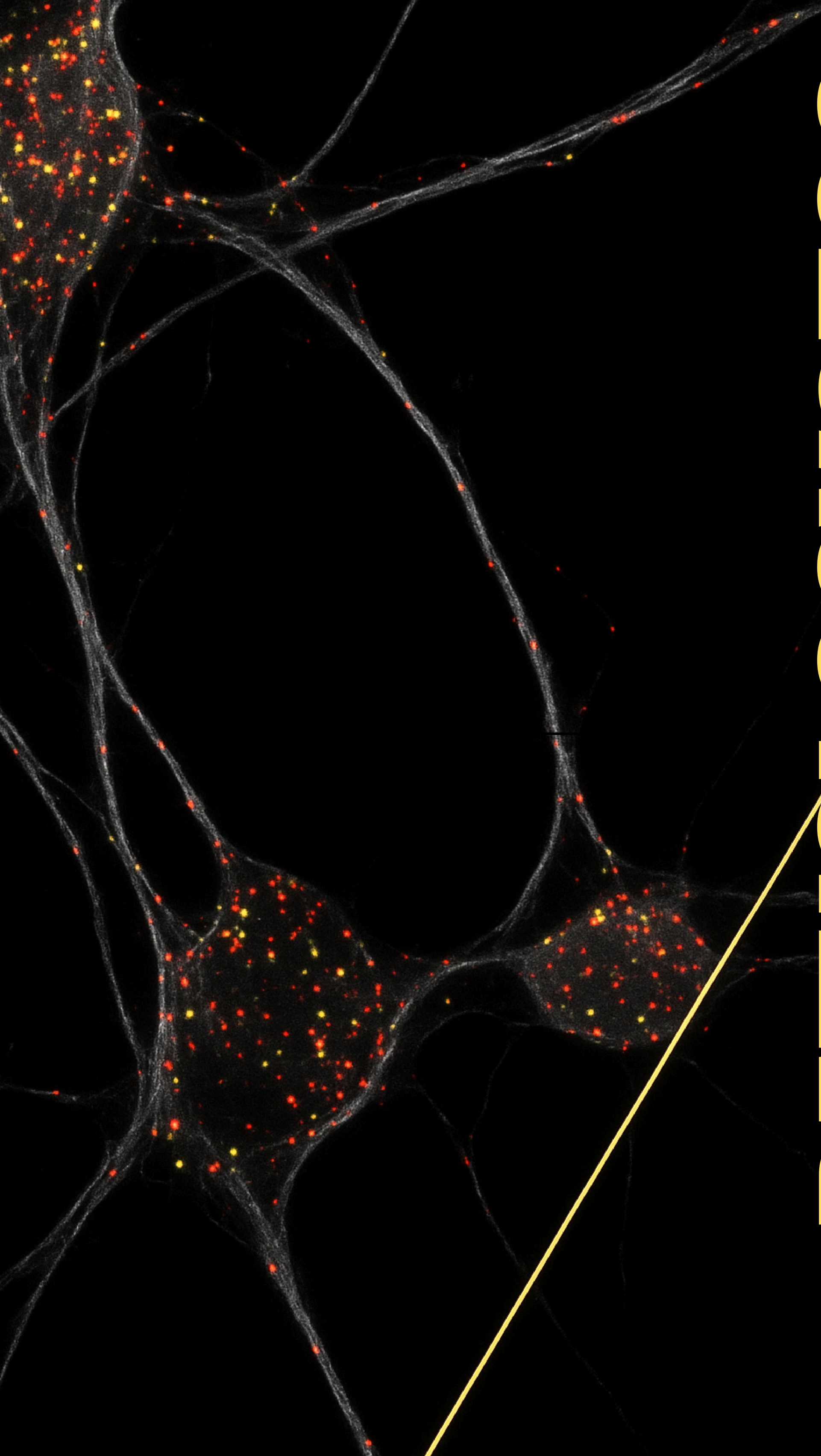

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