Stiff Man Syndrome: Science and experience

Liz Blows

Article points

1. Stiff Man Syndrome symptoms can include fluctuating stiffness and spasms that occur spontaneously or in response to stimuli and anxiety.
2. Increasing awareness of this rare and severe condition would help increase the speed of diagnosis for individual patients and the number of correct diagnoses in the community.
3. As 30% of SMS patients also have type 1 diabetes, healthcare professionals in the field of diabates are ideally positioned to facilitate diagnosis if they are properly educated about the condition.

Key words

- Stiff man syndrome
- Diagnosis
- Education campaign

Liz Blows is a former nurse. She was diagnosed with Stiff Man Syndrome in 1997.

Stiff Man Syndrome (SMS) may have a comical name, but the condition itself is not to be laughed at. SMS is generally accepted as having an autoimmune origin (McEvoy, 1991; Solimena et al, 1988) with about 30% of people with SMS also diagnosed as having type 1 diabetes (Meinck and Thompson, 2002). This article aims to raise the awareness of SMS and outline a possible role for nurses in the field of diabetes in the early identification of a debilitating condition, currently known by the author to be affecting at least 100 people in the UK.

Stiff Man Syndrome was the name assigned to the condition of 'progressive fluctuating muscular rigidity and spasm' when first identified by Moersch and Woltman (1956). Classical SMS is associated with a number of other autoimmune diseases (Meinck and Thompson, 2002).

The condition is rare: during a 10-year period, 20 individuals were identified from a German population estimated to number between 2 and 3 million (Meinck and Thompson, 2002). The literature states that the average age of onset is in the third to fifth decade of life and in the majority of cases, SMS has an insidious onset with a slow progression over months or even years (Meinck and Thompson, 2002). There is no clear racial or ethnic predisposition (The Offical UK Stiff Man Syndrome Support Group and Charity, 2006), although the syndrome appears to be more common in women than men (Murinson, 2004).

Fluctuating stiffness and paroxysmal spasms of the trunk and legs are the primary features of SMS and its variants. The parts of the body affected include the face, neck, abdomen and/or arms, but more typically the legs or lumbar spine. Some individuals have reported to the author transient stiffness of one or both legs, sudden unexplained falls or failure of gait initiation that tends to manifest during emotional stress. Stiffness can be extremely painful and fluctuates from virtually absent to absolutely incapacitating. Body parts involved become rigidly immobile and often show skeletal abnormalities such as hyperlordosis (extreme lordsosis, where there is an increase in the normal anterior concavity of the lumbar spine) and ankylosis. Occasional fixed deformities of either the hands or feet can be present, as can other symptoms that can – albeit more rarely – affect speech, bladder and bowel function (Meinck and Thompson, 2002).

Spasms can occur spontaneously or be precipitated by a variety of stimuli such as noise or touch and may manifest as an excessive startle reaction (Meinck and Thompson, 2002). Spasms may also be precipitated by simple attempts to move. Emotional upsets, stress and minor motor demands or brisk movements are highly effective at provoking spasms. Excessive startle, space phobia, and spasms – often induced by emotional upset – probably contribute to the initial misdiagnosis of hysteria or depression in many patients (Meinck and Thompson, 2002; Murinson, 2004). After such symptoms are identified by the individual and consultant

physician together, the author believes that specialist nurses can potentially contribute to the speeding up of investigations and diagnosis by being aware of the signs of SMS by helping individuals in describing their symptoms. In the author's experience it is likely the individual will have very little idea about what is happening to their body and may require help in exploring their reaction to the condition.

In the author's opinion, it is vital for specialist nurses to play their part in identifying people who may be suffering from SMS. It is sad but true that despite the severity of the syndrome, because it is rare, there is very little experience and knowledge of SMS in any group of healthcare specialists. This results in the condition not being recognised simply because it may seem like the person has depression or anxiety. Specialist nurses can act as an early warning system and a signpost for consultants. The key is watching for the tell-tale signs described here and making sure any suspicions are followed through with the appropriate tests to determine a diagnosis.

Reasonably common, co-existing conditions include thyroid disease, vitiligo and pernicious anaemia (Meinck and Thompson, 2002). However, the most common is type 1 diabetes, occurring in approximately 30% of people with SMS (Meinck and Thompson, 2002). Both type 1 diabetes and SMS feature immune responses to a particular enzyme: glutamic acid decarboxylase (GAD; Meinck and Thompson, 2002; Piquer et al, 2005; Raju et al, 2005).

GAD

Everyone has GAD but in the presence of specific antibodies the enzyme can be destroyed. In type 1 diabetes the destruction of GAD affects the transmission of insulin between the pancreas and the liver. In SMS, the destruction of GAD affects the transmission of nerve impulses, causing rigidity and painful spasms.

GAD is an enzyme commonly found in animals and is necessary for the synthesis of the inhibitory neurotransmitter gamma-aminobutyric acid (GABA; Abe et al, 2005). GABA is the most important and abundant inhibitory neurotransmitter in the brain. While the GAD pathway is not the only source of GABA for the central nervous system, it is a significant source and interference of this pathway – such as when GAD is depleted by the presence of antibodies – can lead to rapid GABA depletion (Vianello et al, 2002; Abe et al, 2005). GABA serves as a natural anti-anxiety compound and as such the most potent anti-anxiety medications are based on augmenting the GABA-A receptor (Cheng and Chiou, 2006). A significant percentage of people with SMS have antibodies to GAD (Lohmann et al, 2003; Raju et al, 2005) and as a consequence these people also have elevated anxiety levels, which worsens the symptoms.

Results to date suggest that in both type 1 diabetes and SMS, antibodies to GAD can be detected. In both conditions, B-cells can become activated and lead to the production of antibodies against GAD (Espay and Chen, 2006). The region of the GAD molecule

The author's story

It took the best part of 4 years for medical staff to diagnose me with SMS. With the slow onset and the apparent anxiety, coupled with the rarity of the condition, this is hardly surprising.

But I was lucky. The neurologist I was sent to see in my home county of Yorkshire had heard of SMS and was immediately suspicious when I was eventually sent to him for a consultation. When I was diagnosed in 1997, I was relieved to have a name to attach to the strange – but very real – symptoms I was experiencing. However, my future was unknown and I felt frightened and very alone with a condition that was as difficult to explain medically as it was to understand as a patient. In the 9 years since I was diagnosed, I have gone from walking unaided anywhere, albeit with an awkward gait, to being reduced to only being able to walk unaided (or 'stiff-manning it' as my husband likes to call it) around the familiarity of my own home, knowing there is always something to hold onto if necessary.

In my house, my condition is usually manageable providing I concentrate on where I am going and have taken plenty of medication. Out in the garden anxiety begins to set in. Although I have my three-wheel walker, an unexpected object in the grass is enough to trip me. Beyond the garden gate anything could happen. I may be able to get to the car with just the help of my walker, or I may need assistance. On the rare occasions when I leave the house for any length of time I use a wheelchair. For an SMS sufferer the 'great outdoors' is often anything but 'great' as wind, snow or ice are almost impossible to endure. Walking down steps or slopes poses incredible difficulties. A shiny floor, even if it is not slippery, can make a sufferer feel anxious, which can trigger rigidity. While I try to never go anywhere unaccompanied, perhaps because I am afraid of having an anxiety attack, I am able to drive alone, probably because the car is a confined and secure space, thus making it an extension of my home.

That's my snapshot of life with SMS, but many sufferers are far worse than me and live a life I could not endure.

Support groups

I was inspired to seek out others with the condition, initially finding someone in the UK through a contact in the US. It was wonderful to talk to someone who understood what I was going through. He told me of three other people with the condition with whom he was in touch. These encounters drove me on and in 1998 I set up a UK support group with five members. A landmark came in 2003 when the support group achieved charitable status. There will be many more SMS patients who do not wish to belong to a group; do not know of the group's existence; or are as yet undiagnosed (the vast majority). Even with my limited resources, there are currently 95 members in my UK SMS Support Group.

It was always going to be tough, but fortunately my husband works in the media and was able to help me raise the profile of the condition. My perseverance is beginning to pay off: awareness of this condition is on the increase. But it is not fast enough. By pushing back frontiers and reaching the key people who can help – especially specialist nurses in an environment where there is such an obvious connection – momentum will gather.

Sadly, because of the rarity of SMS, there are many neurologists who are still not aware of the condition. Rheumatology, immunology, endocrinology and diabetology are all areas that need to be targeted for awareness. Specialist nurses can and must play a major role in this.

Almost 30% of SMS sufferers also have type 1 diabetes, therefore I think that diabetes nurses can be instrumental in identifying the real cause of symptoms indicative of SMS.

This is already happening. Four months after I was given my diagnosis, my diabetes consultant had me videoed with and without diazepam. It was shown at a diabetes conference in London, where a DSN spotted the similarities between myself and a patient of her own. It would have taken much longer for that individual to be diagnosed if it had not have been for the exceptional vigilance of that particular nurse.

targeted by these antibodies seems to differ between people with diabetes and those with SMS (Piquer et al, 2005; Lohmann et al, 2003). The differences between antibodies in both type 1 diabetes and SMS have been considered and research studies have been determining whether those patients with SMS and type 1 diabetes show a different response, human leukocyte antigen profile or clinical features from SMS individuals without type 1 diabetes (Raju et al, 2005). Further investigation into these features could lead to immunomodulatory therapy in both conditions.

Investigation

Because of the rarity and lack of awareness of SMS, reaching a diagnosis can be prolonged and very frustrating for the individual. Thus, it is essential that the following investigations are carried out if SMS is suspected (Meinck and Thompson, 2002).

- Blood tests to look for antibodies to GAD.
- Electromyelogram to measure nerve conduction in affected muscles.
- Magnetic resonance imaging or computed tomography scanning.
- Electroencephalogram to exclude epilepsy.

Central to evaluation for SMS is a detailed history and neurological examination. Hyperreflexia (overactive or over-responsive reflexes) may be the only pathological finding during a neurological examination (Meinck and Thompson, 2002). The cardinal symptoms are essential to the diagnosis of this condition and isolated laboratory results do not stand alone. A related disorder has been found in association with lung or breast cancer and is distinguished by the production of anti-amphiphysin antibodies (Murinson, 2004).

Treatment

The response to medication is important in discriminating other causes of stiffness (Meinck and Thompson, 2002). The first-line medications are diazepam, a benzodiazepine that acts on neurotransmitters and helps in the treatment of anxiety and muscle spasms (Meinck and Thompson, 2002) and baclofen, which is also a muscle relaxant (The Official UK Stiff Man Syndrome Support Group and Charity, 2006). Second-line treatments include tizanidene and gabapentin. The above medications all have a sedating effect. Intravenous immunoglobulin has been shown to decrease the concentration of GAD antibodies and reduce SMS symptoms, with the added benefit of not acting as a sedative (Dalakas, 2005). All of these treatments are, in some respect, 'hit and miss' – it is as much a matter of correct titration of the dosage as changing medications in order to obtain the required effect, all of which takes time.

No treatment yet leads to a cure. However, medications are able to help control symptoms in the majority of cases. This is particularly true in classical SMS, which, if identified and managed appropriately, does not prevent a good outlook for the individual. It tends to be more difficult to control symptoms in the variant stiff limb syndrome due to progressive encephalomyelitis with rigidity.

Conclusion

Raising awareness of SMS is of great importance, as is a knowledge and understanding of SMS by nurses who are, in most instances, best placed to spot the symptoms and report them to consultant physicians to speed up the diagnostic process. ■

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For further information on this condition, please log on to the support group's web site at www.smsgroup.fsnet.co.uk, e-mail the author at liz@blows.fslife.co.uk or telephone the Official UK Stiff Man Syndrome Support Group and Charity: 01482 868881.