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CASE REPORT

SUCCESSFUL TREATMENT OF STIFF PERSON SYNDROME WITH INTRATHECAL BACLOFEN

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Intrathecal baclofen therapy is a recognized treatment for severe spasticity. We report here a case of stiff person syndrome in Australia, treated with intrathecal baclofen followed by a rehabilitation programme with substantial clinical and functional improvements. A 59-year-old woman diagnosed with stiff person syndrome had become hoist-dependent and required full care due to severe spasticity over the past 12 years. Treatment with oral benzodiazepines and botulinum toxin injections to the affected muscles had no therapeutic response. After a test dose of 100 µg intrathecal baclofen resulted in a substantial improvement in her physical function, a decision was made to insert an intrathecal baclofen delivery device. This case report supports the use of intrathecal baclofen therapy and a formal inpatient rehabilitation programme for spasticity related to stiff person syndrome.

Key words: stiff-person syndrome; stiff man syndrome; intrathecal baclofen.

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LAY ABSTRACT

Stiff person syndrome, a rare autoimmune and neurological disease that leads to significant disability, can be treated by the use of a device delivering baclofen into the spine combined with physical rehabilitation. A 59-year-old woman was diagnosed with stiff person syndrome and had become dependent, with full-time care, due to severe spasms. Conventional treatment with oral muscle relaxants and botulinum toxin injections to her muscles did not have an adequate effect. A trial dose of baclofen was administered into her spine, followed by implantation of a drug delivery device. This resulted in a substantial improvement in function.

Stiff person syndrome (SPS) is a rare autoimmune progressive neurological disorder characterized by truncal spasticity and painful spasms, with devastating functional sequelae (1, 2). Classically, SPS has been associated with certain autoimmune disorders, such as Grave's disease, pernicious anaemia and diabetes mellitus (3). The mainstay of managing spasticity involves oral anti-spasmodic agents, such as baclofen, which acts on gamma aminobutyric acid (GABA) receptors to decrease hyperexcitability of the central nervous system (4). However, oral baclofen may be poorly tolerated at high doses due to systemic side-effects (5). If the affected muscle groups are localized, botulinum toxin can be trialled followed by a formal rehabilitation programme. International studies reported the use of intrathecal baclofen therapy in occasional cases of SPS (6, 7).

We report here a case of SPS in Australia, which was treated with intrathecal baclofen with substantial clinical and functional improvements.

CASE REPORT

A 59-year-old woman presented to our hospital with progressive stiffness and painful spastic episodes of the trunk, upper and lower limbs for the past 12 years. She had significant functional decline in mobility and required full assistance in personal and instrumental activities of daily living (ADLs).

The patient reported symptoms of abnormal involuntary movements of the arms and legs since childhood. These symptoms appeared sporadically with no specific physical or emotional triggers. Her involuntary movements had worsened progressively over her second to third decades of life. The symptoms peaked in her 50s, when she was

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unable to work due to the prolonged nature of lower leg stiffness and painful spasms. By the time she presented to our hospital she was hoist-dependent and required full-time care. Oral antispasmodic medications were trialled unsuccessfully. She was diagnosed with conversion disorder after seeing numerous neurologists and psychiatrists. Botulinum toxin injections were trialled in an attempt to improve leg function. However, given the systemic nature of her spasticity this was largely ineffective.

Her medical history included hypertension and non-insulin dependent diabetes mellitus. There was no family history of any similar neurodegenerative illnesses or autoimmune conditions.

Initial examination of the patient revealed significant generalized spasticity in the lower limbs with a Modified Ashford Scale (MAS) of 3–4. Gentle tactile and sensory inputs precipitated violent involuntary limb spasms and truncal movements. Her muscle power grade ranged from 0/5 distally to 3–4/5 proximally pre-treatment. There was bilateral non-fatigable ankle clonus. Her bowel and bladder function was intact. Functionally, she required a full hoist transfer and manual wheelchair for mobility. Full assistance was required for self-care, including grooming, dressing and toileting. Her care needs are reflected in her Functional Independence Measure (FIM) (8).

Investigations, including full blood count, urea/electrolyte, liver function test, serum glutamic acid decarboxylase autoantibodies, were unremarkable. Autoimmune screening was negative. Cerebral spinal fluid analysis was unremarkable. A diagnosis of sero-negative SPS was made.

Following a successful test dose of intrathecal baclofen (100 μg), a baclofen pump (Synchromed II, Medtronic) was inserted. The dose of intrathecal baclofen was titrated up gradually from 150 μg/day to 700 μg/day. A comprehensive inpatient interdisciplinary rehabilitation programme was followed, including bed exercise programme, sitting balance, transfer practice, gait retraining and ADL retraining. Inpatient rehabilitation was deemed necessary given the patient's significant distance from services.

After 3 weeks of inpatient rehabilitation, the patient was able to mobilize for short distances without assistance. She was also able to ascend and descend stairs. Her muscle strength, control and co-ordination had improved, with significant reduction in spasticity measured in MAS with scores ranging from 0–1. Subsequent rehabilitation episodes resulted in further functional improvements (**Table I**). These include unsupervised outdoor mobility and driving. The patient remains under outpatient surveillance to this day (3 years).

Table I. Progress of Functional Independence Measure

FIM	On admission	One-month post	Three-months post
Self-care	6/42	39/42	40/42
Sphincter control	14/14	14/14	14/14
Transfers	3/21	18/21	21/21
Locomotion	2/14	12/14	14/14
Total motor	25/91	83/91	89/91
Communication	14/14	14/14	14/14
Social/cognitive	21/21	21/21	21/21
Total cognitive	35/35	35/35	35/35
Total FIM score	60/126	118/126	124/126

FIM: Functional Independence Measure.

DISCUSSION

SPS is a rare autoimmune progressive neurological disorder with a wide clinical spectrum of disease symptomatology, ranging from localized lower limb spasticity (stiff-leg syndrome), facial spasms, status spasticus, cerebellar ataxia, encephalomyelopathy with rigidity and myoclonus and full-body generalized spasticity (7, 9, 10). SPS is a rare condition and the authors advocate tailored rehabilitation management individualized to the patient's functional impairments. We recommend that SPS should be treated in a subspecialty unit with access to intrathecal baclofen as one of the treatment options.

Intrathecal baclofen therapy has potential drawbacks, including surgical risks (11), infection risks (12), pump failure, cost implications, and the requirement for regular quarterly refills. These factors should be considered in combination with the potential benefits of such treatment.

The criteria for a successful trial was a clinically significant reduction in spasticity and involuntary muscle movement. This occurred for a 12-h period following the test dose. Intrathecal baclofen therapy is a well-recognised and applicable treatment for severe spasticity that has failed more conservative and limited treatment. For example, oral baclofen, botulinum toxin, and physical therapy.

Given the life-changing and significant functional improvements that occurred in this case, we conclude that, for best functional outcome, intrathecal baclofen therapy should be considered in similar cases, in combination with a rehabilitation programme. A video of the patient post-treatment is available (13).

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