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CASE REPORT **OPEN ACCESS**

CELL BASED TREATMENT FOR SPINOCEREBELLAR ATAXIA: A CLINICAL CASE REPORT

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ABSTRACT

Background: Spinocerebellar ataxia (SCA) is a dominant and monogenic central nervous system disorder, characterized by progressive motor disorders affecting coordination, balance, speech, and ADL. The prevalence is 2.7 out of 100,000 people. Currently, cell-based therapy is being developed for clinical improvement in SCA.
Case: A 48-year-old woman with weakness in both legs and arms, difficulty walking, dysmetria, and dysarthria since 2015. Examination of SARA scoring is severe ataxia, EMG showed polyradiculopathy, muscle denervation, and suspicion of posterior ramus lesions; complete blood laboratory and tumor markers were routine. MRI brain contrast and whole spine contrast radiology imaging were also performed. Clinical improvement was achieved in 2016 following stem cell injection in Thailand (dose and type of cells unknown). However, clinical worsening occurred from 2020 to 2024. The patient received Umbilical Cord Mesenchymal Stem Cells (MSC) in October 2024, administered intrathecally at a dose of 20 million cells.
Discussion: The first injection showed significant clinical improvement. The second injection showed no clinical improvement, but no worsening of symptoms was found. The difference in results may be due to variations in the route of administration, cell type, cell quality, and the dose administered.
Conclusion: The administration of umbilical cord mesenchymal stem cells (UC-MSCs) in SCA is considered safe, with minimal complications, and can suppress disease progression, although it does not produce clinical improvement.

Keywords: mesenchymal stem cell, neural regeneration, spinocerebellar ataxia, stem cell therapy, umbilical cord

Introduction

Spinocerebellar ataxia (SCA) is characterized by progressive ataxia, speech and swallowing difficulties, loss of coordination, and gait disturbances that significantly impact most aspects of daily living. Spinocerebellar ataxias are monogenic dominantly inherited and progressive disorders of the central nervous system (CNS), mainly affecting the cerebellum, caused by a cytosine-adenine-guanine expansion, which encodes a long glutamine tract (polyglutamine/polyQ) in the respective wild-type protein, causing misfolding and protein aggregation.[1–4] The worldwide prevalence of dominant hereditary ataxias is 2.7 cases per 100000 individuals, varying by ethnicity and geography. The overall European prevalence is 0.9 to 3 per 100,000. Asian countries have limited published data. Among the 47 types of spinocerebellar ataxia identified, types 1, 2, 3, 6, and 7 are the most common worldwide. Type 3 is the most prevalent, and type 2 is the most common in South Korea and India.[1–4]

Over the past decade, efforts have been made to ameliorate disease symptoms in patients, yet no cure is available (no causal or definitive therapy). Recent advances in therapeutic approaches related to stem cell and genetic intervention.[2–4] The exploration of cellular-based therapies for SCA, particularly those caused by polyglutamine (polyQ) expansions, represents a

promising frontier in the treatment of neurodegenerative diseases. While current pharmacological interventions primarily focus on alleviating symptoms rather than addressing the underlying causes, advancements in stem cell technology offer new avenues for potential disease modification. Previous clinical studies have shown that mesenchymal stem cell (MSC) grafts can restore motor impairments in polyQ SCA patients, but with limited sustained effects.[3]

Case Report

A 48-year-old woman with weakness in both legs and arms, difficulty walking, stiffness in both hands, dysarthria, dysmetria, frequent choking when eating and drinking, difficulty writing, buttoning clothes, and holding a spoon since 2015. Family history of ataxia was found in her biological mother and older sister (2 out of 3 siblings suffered from ataxia). In 2016, the patient received intrathecal and intravenous stem cell therapy in Thailand (the dose and type of stem cells used were unknown), resulting in significant clinical improvement, which allowed the patient to walk again. Then, from 2020 to 2024, the patient's complaints began to reappear, starting with slurred speech. Over time, other complaints emerged and worsened, until she could no longer walk. During this time, the patient received no drug therapy or physiotherapy.

Examination data obtained in 2024 included the Scale for the Assessment and Rating of Ataxia (SARA)[4,5], which scored 26 (severe ataxia), as shown in Table 1. Electromyography (EMG) examination (NCV and needle EMG) in conclusion obtained results of C5-Th1 and L4-S1 polyradiculopathy and muscle denervation at the left C1-Th3 level, which could also be a posterior ramus lesion as shown in Table 2. Complete blood laboratory results and tumor markers (CEA, alpha fetoprotein, Ca 125, Ca19-9) were within normal limits. Brain contrast Magnetic Resonance Imaging (MRI) obtained a lacunar infarction in the bilateral parietal lobes and left thalamus, and mild focal atrophy of the bilateral frontoparietal lobes, as shown in Figure 1a. A contrast whole-spine MRI revealed mild lumbar scoliosis, as shown in Figure 1b. Due to limited facilities, genetic evaluation and sequencing examinations have not been performed on this patient.

The patient received the second stem cell treatment in Indonesia, an intrathecal injection of Umbilical cord mesenchymal stem cells (UC-MSCs) in October 2024, with a dose of 20 million cells. Clinical evaluation was conducted 4 months after the intrathecal injection of UC-MSCs in patients; the SARA score remained unchanged, indicating no significant clinical improvement. However, there were no signs of worsening clinical symptoms or significant complications and side effects after intrathecal injection of UC-MSCs.

Table 1 Scale for the Assessment and Rating of Ataxia (SARA) in the index patient

Domain	Score
Gait	8
Stance	6
Sitting	1
Speech disturbance	3
Finger chase (R)	1
Finger chase (L)	1
Finger chase (Mean)	1
Nose-finger test (R)	2
Nose-finger test (L)	2
Nose-finger test (Mean)	2
Fast alternating hand movements (R)	1
Fast alternating hand movements (L)	1
Fast alternating hand movements (Mean)	1
Heel-shin slide (R)	4
Heel-shin slide (L)	4
Heel-shin slide (Mean)	4
Total	**26**

R: Right, L: Left, Mean is used for total score summation whenever right and left sides are examined

Table 2: Electromyography (EMG) findings in the patient

Electromyography test	Impressions
SNCS (upper and lower extremities)	Sensory polyneuropathy of bilateral median nerve and bilateral ulnar nerve lesions (axonal lesions); bilateral sural nerve (severe lesions)
MNCS (upper and lower extremities)	Motor polyneuropathy of bilateral axillary nerves and bilateral musculocutaneous nerves (axonal lesions); bilateral radial nerves and bilateral ulnar nerves (demyelinating lesions); right peroneal nerve (axonal lesions)
Proximal peripheral nerves	Polyradiculopathy at C5-C7 bilaterally and C8-Th1 on the left side (severe lesion); C8-Th1 on the right side; L4-L5 bilaterally and L5-S1 on the left side (severe lesion); L5-S1 on the right side
Needle EMG (C1-2, C7, Th3 paraspinal muscle, rhomboid major muscle)	Positive wave (+), fasciculations (+) MUP pattern may support signs of muscle denervation at the C1-Th3 level on the left side, and may also suggest a posterior ramus lesion

SNCS: Sensory Nerve Conduction Study, MNCS: Motor Nerve Conduction Study, EMG: Electromyography, MUP: Motor Unit Potentials, C: Cervical, Th: Thoracal, L: Lumbar, S: Sacral

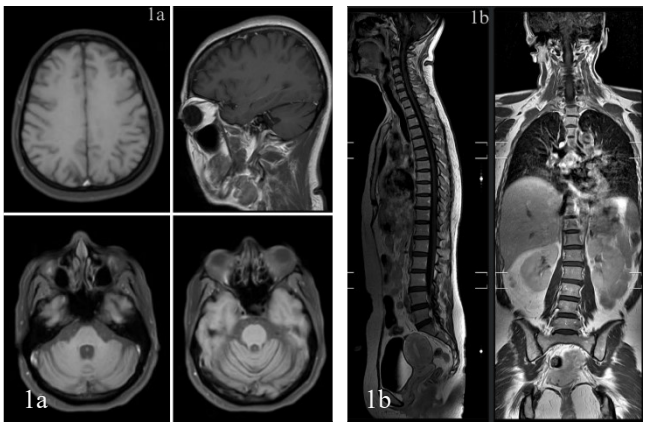

Figure 1a. Brain contrast MRI findings: bilateral parietal lobes subacute lacunar infarction, bilateral frontoparietal lobes focal mild atrophy, not obtained brain stems and cerebellum atrophy, 1b. Whole spine contrast MRI findings: mild narrowing of the disc space C4-5 and C5-6, mild scoliosis of the lumbar, no visible pathological signal intensity in the spinal cord and bone marrow

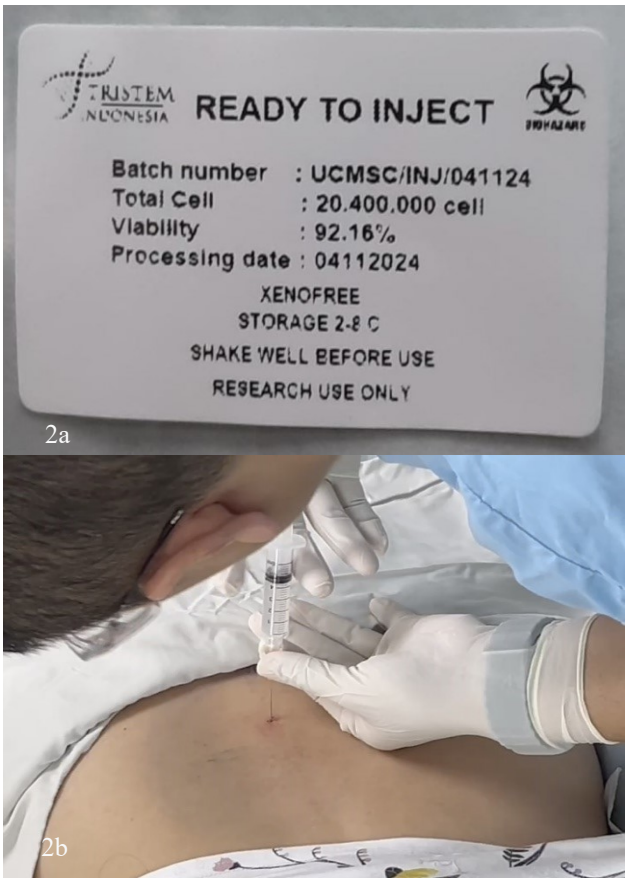

Figure 2a. Patients received a dose of 20 million umbilical cord mesenchymal stem cells (UC-MSC), 2b. Intrathecal administration of UC-MSC (start with lumbar puncture of 3cc CSF, then insert UC-MSC in 4cc of NaCl)

Discussion

Spinocerebellar ataxia (SCA) is primarily characterized by progressive ataxia, speech and swallowing difficulties, loss of coordination, and gait disturbances that significantly impact most aspects of daily living. Spinocerebellar ataxias are monogenic dominantly inherited and progressive disorders of the central nervous system (CNS), mainly affecting the cerebellum, caused by a cytosine-adenine-guanine expansion, which encodes a long glutamine tract (polyglutamine/polyQ) in the respective wild-type protein, causing misfolding and protein aggregation.[1–4] polyQ repeats play a crucial role in patient clinical manifestations. The progressivity and classic clinical findings of SCA, related to its anatomical involvement, include an unsteady walk as the initial sign, followed by slurred and scanning speech, dizziness, cranial nerve palsy, ophthalmoplegia, visual problems, dysarthria, dysphagia, cognitive problems, and motor-sensory problems. Mortality in the later stages due to progression, which leads to immobility and an inability to breathe. Some experts have classified SCA into four subtypes: type 1 as an early-onset disease with extrapyramidal signs and spasticity, but minimal ataxia; type 2 as midlife progressive ataxia; type 3 as later-onset ataxia accompanied by neuropathy, amyotrophy, and loss of reflexes; and type 4 as Parkinsonism with and without ataxia.[2,4]

The standard protocols include clinical findings of ataxia and related signs and symptoms, exclusion of a direct or secondary ataxia, a positive family history, and specific gene testing. The family history indicates that the inheritance pattern for the condition is autosomal dominant, with no skipped generations and both males and females affected. While most SCAs are autosomal dominant, a small but increasing number are recessive.[1,2,4,6] Clinical scales are considered essential prerequisites for observational studies; the currently most widely used ataxia scale is the Scale for Assessment and Rating of Ataxia (SARA). The SARA sum score ranges from 0 to 40, with zero indicating the absence of ataxia and 40 being the most severe degree of ataxia.[5,7–9]

Neuroimaging plays a crucial role in assessing and understanding SCA, revealing distinct patterns of brain atrophy and other structural changes (cerebellar atrophy, ventricular enlargement) associated with different subtypes of the disease, which may contribute to the clinical symptoms experienced by patients. Some specific focal or regional atrophies appreciated in certain SCAs are gross cerebellar atrophy (SCA2), pontocerebellar atrophy with enlargement of the fourth ventricle (SCA3), atrophy of vermis sparing brainstem (SCA5, SCA8, SCA10), isolated cerebellar atrophy (SCA6, SCA12), etc. Other experts categorized MRI imaging into four groups: pure cerebellar atrophy, cerebellar atrophy with additional degeneration of neuroanatomical structures, selective cerebellar atrophy, or no cerebellar atrophy.[6] Advanced imaging techniques can detect metabolic changes in the brain, such as Positron Emission Tomography (PET) and

Magnetic Resonance Spectroscopy (MRS), which are not routinely performed due to their cost and availability.[2,4,6,10–12]

Electrophysiological testing provides additional information on neuronal function but lacks specificity for subtype differentiation. Reduced sensory nerve action potential (SNAP) and nerve conduction velocity (NCV) are the primary abnormalities in nerve conduction studies (NCS). Motor and sensory nerves are affected in SCA1, SCA2, and SCA3, with sensory nerve abnormalities more pronounced than those in motor nerves. Pure sensory nerve studies are abnormal in SCA4, SCA18, and SCA25. EMG abnormalities are of three types for SCA1, SCA2, and SCA3, and contain fasciculation potentials, giant motor unit action potentials (MUAPs), and reduced recruitment.[2,6,13]

Advances in molecular genetic analysis and testing facilitate early classification and diagnosis of SCA. Identifying a specific mutated gene allows for testing in other family members, making genetic testing the definitive method for determining SCA subtypes, especially in cases with a positive family history. Polymerase chain reaction (PCR) is used to analyze nucleotide repeats at various SCA gene loci, thereby helping to identify the specific gene and its associated repeats.[1–3,11–15]

Spinocerebellar ataxia has a heavy overlap with other neurodegenerative disorders, making it difficult to diagnose. Genotype has become the gold standard for diagnosis, but clinical manifestation and characterization are imperative before genetic analysis.[2,4] In this case, the results of several examinations, such as clinical manifestation, SARA scoring, and EMG impressions, point to SCA 2. However, the neuroimaging results did not find specific findings for SCA. A diagnosis of probable SCA can still be established; however, it would be beneficial to perform genetic examination, given the positive family history.

Spinocerebellar ataxia is a genetic disease with no definitive cure. Treatment mainly focuses on symptomatic relief, such as seizures, tremors, depression, ataxia, and eye symptoms. Antiepileptic drugs, botulinum toxin injections, beta-blockers, primidone, antidepressants, and levodopa are used for symptomatic treatment. Other treatments include N-acetyl cysteine, citalopram, dantrolene, chlorzoxazone, zolpidem, and varenicline. However, any therapeutic procedure that clears accumulated misfolded mutant proteins can be a potential treatment option. Chemical chaperones, proteasome activity, and mTOR inhibitors can accelerate the degradation of mutant proteins. Antisense oligonucleotides can decrease cerebellar ataxin expression, delay the onset of SCA, increase the firing frequency of Purkinje cells, and improve motor function. Neurorehabilitation and physical therapy are essential for enhancing motor functions and maintaining independence. Occupational therapies, such as wheelchair support and adaptive devices, can also be augmented with communication devices and behavioural intervention.[2] All these treatments have been made to ameliorate disease symptoms in patients, yet no cure is available. Previous studies have proposed using stem cells as promising tools for regenerating central nervous system tissue.[3,14–22]

Regenerative medicine offers promising tools through cell-based therapies, focusing on restoring cellular circuitries. Four regenerative treatments are available: stem cell therapies, platelet-rich plasma-based therapies, lipogems, and prolotherapy. These treatments promote ligament and tendon healing, as well as neurogenic inflammatory pain. Recent studies suggest that transplanted cells can provide environmental enrichment or promote the development of neural networks. Pluripotent stem cells can be obtained from the inner cell mass (ICM) of the blastocyst as embryonic stem cells (ESCs) or from somatic cells through reprogramming approaches that originate induced pluripotent stem cells (iPSCs). Differentiating the three germ layers (ectoderm, mesoderm, endoderm) is possible by inhibiting OCT4, SOX2, and NANOG factors in pluripotent cells. The neural progenitor cells (NPC) are derived from the ectoderm, and the mesenchymal stem cells (MSC) are derived from the mesoderm. Multipotent cells such as NPC can give rise to neural and glial cells, and MSC can be differentiated at least into adipocytes, chondrocytes, or osteoblasts.[3,14,16–18,23]

Stem cell therapy for polyQ disorders is primarily in the pre-clinical research stage, with only a few clinical studies examining its safety and efficacy. Human MSCs from the umbilical cord or adipose tissues are preferred for SCA patients due to their safety and neuronal feeding factors. A Phase II trial is currently underway to assess efficacy. MSC therapy can be administered to reach the subarachnoid space through various routes, including intracranial, intra-arterial, intravenous, intralesional, and intrathecal. Intra-arterial or intravenous routes have been observed to exhibit suboptimal cell delivery into lesion areas, owing to the possible inability of cells to cross the blood–brain barrier (BBB) or potential retention in other organs. Several recent studies have shown significant results in intravenous UC-MSC administration, such as improved motor deficits and alleviated cerebellar atrophy, promoted Purkinje Cell (PC) survival, increased expression of IGF-1, NGF, VEGF in cerebellum, decreased apoptosis in the cerebellum and pons, improved neuronal-muscular electrophysiology, and also MSCs remained viable and secreted cytokines until 20 weeks post-transplant.[14–20,24]

The intravenous route remains the most commonly utilised, followed by intrathecal injection. This preference may be attributed to ethical considerations arising from the potential risks associated with lumbar puncture and MSC administration. Some studies have found that administering MSCs via intrathecal delivery results in a minor increase in adverse effects (AEs) associated with musculoskeletal and connective tissue disorders, but no severe AEs have been reported.[3,15,20] In this clinical case report, the first injection showed significant clinical improvement. The second injection showed no clinical improvement, but no worsening of symptoms was found. The difference in results may be due to variations in the route of administration, cell type, cell quality, and the dose administered.

Conclusion

The administration of intrathecal umbilical cord mesenchymal stem cells (UC-MSCs) in SCA is considered safe, with minimal complications. It can suppress disease progression, although it does not produce clinical improvement. Further evaluation is needed regarding dosage, route of cell administration, choice and quality of stem cells, various background conditions of SCA survivors, inhibiting factors, and therapy side effects to obtain more significant results.

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