Article • Spinocerebellar Ataxia: The Role of Optometry and Binocular Vision Therapy in Rehabilitation and Improving Quality of Life

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ABSTRACT

Background: Spinocerebellar ataxia (SCA) is a group of progressive, inherited neurodegenerative disorders that impact coordination, balance, and vision. SCA subtypes, especially SCA7, present unique ocular manifestations, such as reduced visual acuity, color vision deficits, retinal degeneration, and oculomotor dysfunction. This multidisciplinary care approach enables patients to live more confidently and independently despite the visual and neurological challenges posed by SCA.

Case Report: A 65-year-old male with spinocerebellar ataxia type 3 (SCA3) presented with diplopia, primarily at distance, and limitations in extraocular movements. Clinical evaluation revealed esotropia at distance and intermittent exotropia at near, managed effectively with 3$^{\Delta}$ base-out correction and binocular vision therapy including vergence exercises. Despite underlying neurodegenerative pathology and coexisting hyperthyroidism, the patient achieved significant symptomatic relief and visual comfort over two years.

Conclusion: This case highlights the role of non-surgical, tailored optometric intervention in managing complex diplopia cases associated with neurological conditions.

Keywords: binocular vision therapy, double vision, orthoptics, spinocerebellar ataxia

Introduction

Spinocerebellar ataxia (SCA) is an inherited progressive neurodegenerative disorder characterized by late-onset cerebellar and brain dysfunction. It is an autosomal dominant condition with mono-allelic pathogenic expansion in the ATXN7 gene.[1-3] Patients have neurological deficits, including ataxia (impaired balance or coordination, can be damage to brain, nerve or muscles) and dysarthria (weakness in the muscles used for speech, which often causes slowed or slurred speech).[1-5] Visual symptoms are the first presenting signs in patients with SCA,[7] including severely affected visual acuity and color vision, ocular motility impairment, and retinal macular degeneration.[6-7] Also, there is a case report of the existence of keratoconus in patient with SCA.[7] The age of onset of SCA7 is usually 30-50 years and is inversely correlated with the length of cytosine-adenine-guanine (CAG) repeat expansion.[1-6] The first neurological features of the disease are truncal or gait ataxia and dysarthria.[8-10]

SCA7 is distinguished from other SCAs by the presence of visual dysfunction in affected individuals. In this case report, we provide a detailed overview of SCA, focusing on its ocular and systemic manifestations, diagnostic criteria, and management strategies. We also present a case report of a 65-year-old male with SCA type 3 (SCA3), highlighting the challenges in managing the disease.

SCA7 is unique among the SCAs due to its early visual dysfunction, which can precede other neurological symptoms.

Ocular Manifestations in SCA7

1. Visual Acuity: Reduced visual acuity is one of the most prominent features in SCA7, often progressing to blindness.
2. Color Vision: Deficits in color vision frequently appear early in the disease.
3. Ocular Motility: Impaired ocular motility can lead to binocular diplopia and significant functional impairment.
4. Saccades: Patients may experience slowing of saccades, affecting the quick, simultaneous movement of the eyes.
5. Ophthalmoplegia: This condition involves paralysis of the eye muscles, further classified into external and internal types.
6. Gaze-Evoked Nystagmus: Nystagmus that occurs when the eyes are held in an eccentric position but not in the primary position.
7. Retinal and Macular Degeneration: A hallmark of SCA7, detectable via electroretinogram ERG or optical coherence tomography prior to visible fundus abnormalities.

Systemic Features of SCA7

- Dysphagia: Difficulty swallowing is common in SCA7 patients.
- Memory Loss: Cognitive decline, including memory loss, can occur.
- Muscle Stiffness and Cramps: These symptoms contribute to impaired mobility.
- Peripheral Neuropathy: Loss of sensation in the extremities is frequently observed.
- Incontinence: Bladder control issues are reported in SCA7.

Case Report

In this case report, we discuss a patient diagnosed with SCA3, also known as Machado-Joseph Disease (MJD), an autosomal dominant genetic disorder first described by Nakano et al. in 1972 among Portuguese immigrants in the United States. SCA3 is characterized by progressive motor dysfunction, with early manifestations including coordination and balance issues.[1-8] Other clinical features include dysarthria, dystonia, spasticity, rigidity, tremors, bulging eyes, and diplopia.[15-16]

In addition to these motor symptoms, patients with SCA3 often present with sleep disturbances, such as restless leg syndrome and REM sleep behavior disorder. Restless leg syndrome is typified by sensations of tingling or numbness in the legs, prompting an urge to move them, while REM sleep behavior disorder involves abnormal muscle activity during the REM phase of sleep, causing patients to physically act out their dreams.[11-16] Although SCA3 shares many clinical similarities with other subtypes, particularly SCA7, it is distinguished by a wide array of systemic and ocular manifestations.

A 65-year-old male presented to our neuro-ophthalmology clinic with a primary complaint of diplopia, most noticeable at distance. His best-corrected visual acuity was 6/6 in both eyes, with a prescription of plano-2.50x015 in the right eye and +0.75-1.75x175 in the left eye. The patient had been using prism glasses with a 3^{Δ} base-out correction in both eyes, though the lenses were significantly scratched. A cover test revealed 5^{Δ} esotropia for distance and 9^{Δ} intermittent exotropia for near. Examination of extraocular movements showed a -2 limitation in all directions of gaze except down gaze. Sensory evaluation using the Worth four-dot test demonstrated normal retinal correspondence for near and diplopia for distance, with stereoacuity measured at 400 seconds of arc using the Titmus fly test.

The patient's medical history was notable for hyperthyroidism, for which he was not taking any medication, in addition to a diagnosis of SCA3. The limitations in extraocular movements were hypothesized to be related to SCA3. The patient was referred to the binocular vision therapy clinic for further management and prism correction. During his review at the binocular vision therapy clinic, he was advised to start vergence exercises using the Brock string at home. Progressive addition lenses with 3^{Δ} base-out prisms in both eyes were prescribed to address his visual needs. The 3^{Δ} base-out was prescribed after clinical assessment revealed that it was the minimum effective prism needed to achieve comfortable fusion and reduce diplopia symptoms. Color vision and contrast sensitivity were normal in both eyes, as assessed using the Ishihara chart and the Bailey-Lovie chart, respectively. The fundus examination showed right eye within normal limits and left eye central serous chorioretinopathy but maintaining aided 6/6 vision in both eyes.

At the initial evaluation, nystagmoid movements were observed during fixation. These movements were absent at a later visit, where the patient reported diminished distance vision in the left eye, though end-gaze nystagmus remained present. After two years of his follow-up, the patient continued using

the same prism correction in his glasses and reported comfort with no diplopia for either distance or near vision. A cover test at this time showed orthophoria for distance and mild exophoria for near. At the final follow-up, a prism adaptation test confirmed the same prism correction, with no diplopia noted for either near or distance vision. The patient was able to appreciate all patterns on the Brock string, although maintaining them for extended periods remained challenging. He was advised to maintain greater compliance with fusional vergence exercises. The patient expressed satisfaction with the treatment plan, noticing significant relief from diplopia through the prescribed exercises and prism glasses.

This case emphasizes the value of individualized binocular vision therapy and precise prism correction in addressing diplopia related to SCA3. While evaluating the patient, several differential diagnoses were considered, including SCA7 (notable for early onset of visual problems), Friedreich's ataxia, multiple system atrophy, ataxia with oculomotor apraxia, episodic ataxia, progressive supranuclear palsy, and toxic or metabolic cerebellar disorders. Other conditions such as myasthenia gravis, internuclear ophthalmoplegia, and paraneoplastic cerebellar degeneration were also considered due to similar ocular signs. Detailed clinical assessment, genetic confirmation, and exclusion of other neurological conditions helped establish the diagnosis of SCA3.

The case highlights that a timely and holistic treatment approach can lead to noticeable improvements in patient comfort and functionality. Since SCA3 presents with both visual and neurological challenges, collaboration among optometrists, neurologists, physiotherapists, and occupational therapists is crucial. Addressing only the visual symptoms may not lead to the best outcomes. Therefore, a team-based strategy that includes motor coordination, balance training, and speech support can greatly enhance the patient's overall well-being. Through such multidisciplinary care, eye care professionals play a significant role in restoring independence and improving life quality for individuals living with SCA3.

Discussion

SCA3 is a progressive neurodegenerative disorder for which, despite extensive research, no curative or disease-modifying treatments have been developed. Like other inherited ataxias, SCA3 is characterized by the degeneration of the cerebellum and other parts of the central nervous system, leading to symptoms such as gait disturbances, coordination issues, and a range of neurological deficits. These include speech difficulties, muscle rigidity, dystonia, and visual disturbances. The genetic basis of the disease, particularly the CAG trinucleotide repeat expansion, complicates efforts to develop effective therapies, resulting in a focus on symptomatic management to improve patients' quality of life.[17,18]

Given the complexity of SCA, researchers have investigated drugs that show efficacy in treating other neurodegenerative disorders. For instance, Grobe-Einsler et al.[19] explored the use of rivastigmine, a cholinesterase inhibitor effective in treating cognitive dysfunction and motor symptoms in Parkinson's disease. As Parkinson's shares certain symptoms with SCA, such as gait disturbances and motor control issues, the trial sought to determine whether rivastigmine could provide similar benefits to SCA patients. Although modest improvements in gait and motor control were observed, the overall impact on disease progression remains limited, highlighting the need for more targeted treatments.[20]

Parkinsonian symptoms in SCA patients, such as rigidity and bradykinesia, have also been managed with dopaminergic agents like levodopa, which can offer relief for motor symptoms but do not halt neurodegeneration.[20] Additionally, common non-motor symptoms in SCA, such as restless leg syndrome, respond to similar treatments, providing symptom relief without altering the disease course. Sleep disturbances, another significant issue in SCA, can be managed with sympathomimetic agents like modafinil and amphetamines to reduce daytime sleepiness, though the long-term benefits and risks of these interventions remain under investigation.[3]

Visual disturbances, including diplopia and nystagmus, are prominent symptoms in SCA, significantly impairing patients' ability to perform daily tasks and contributing to social isolation.[21] Optometrists play a crucial role in managing these symptoms through interventions like prism glasses, which help realign the visual fields and reduce double vision. However, prism correction alone is often insufficient, and additional therapies are needed to improve long-term visual outcomes.

Binocular vision therapy is one such approach, aiming to improve oculomotor coordination and maintain binocular single vision. Exercises such as Brock string training can strengthen vergence movements and help patients achieve better visual

stability.[22,23] Although binocular vision therapy cannot reverse the progression of SCA, early intervention and consistent practice can delay severe oculomotor dysfunction and enhance functional independence.

Several additional professionals played a significant role in the care of this patient. The neurologist played a key role in confirming the diagnosis of SCA3 through clinical examination and genetic testing and provided ongoing neurological evaluation to monitor disease progression and manage systemic symptoms such as ataxia, dysarthria, and muscle stiffness. The occupational therapist focused on improving the patient's ability to perform daily tasks independently by addressing fine motor coordination, hand function, and adaptive strategies for home and work environments. The physical therapist worked on enhancing balance, strength, and gait through customized exercises aimed at reducing fall risk and maintaining mobility. Together, this multidisciplinary team supported the patient in achieving better functional independence and overall quality of life.

Given the complexity of SCA, a multidisciplinary approach is essential for effective management. Optometrists, neurologists, physical therapists, and occupational therapists must work together to address the wide range of symptoms, including balance, coordination, and speech deficits. Comprehensive care that integrates visual and systemic management can greatly improve patients' quality of life.

Conclusion

In conclusion, while there is no cure for SCA, symptom management through pharmacological treatments like rivastigmine and dopaminergic agents, as well as non-pharmacological approaches like binocular vision therapy, can provide significant relief. Addressing visual symptoms, in particular, can help reduce social isolation and improve daily functioning. By offering a coordinated and holistic approach to care, clinicians can empower SCA patients to maintain a better quality of life despite the progressive nature of the disease.

References

1. Campos-Romo A, Graue-Hernandez EO, Pedro-Aguilar L, et al. Ophthalmic features of spinocerebellar ataxia type 7. Eye 2018;32(1):120-7.
2. Park JY, Joo K, Woo SJ. Ophthalmic manifestations and genetics of the polyglutamine autosomal dominant spinocerebellar ataxias: A review. Front Neurosci 2020 Aug 21:14:892. doi: 10.3389/fnins.2020.00892.
3. Lebre AS, Brice A. Spinocerebellar ataxia 7 (SCA7). Cytogenet Genome Res 2003;100(1-4):154-63.
4. Fagerström R. Fear of a cataract operation in aged persons. Psychol Rep 1993 Jun;72(3 Pt 2):1339-46.
5. Bhandari J, Thada PK, Samanta D. Spinocerebellar Ataxia. 2023 Sep 15. In: StatPearls. Treasure Island (FL): StatPearls Publishing; 2024 Jan.
6. Soong BW, Morrison PJ. Spinocerebellar ataxias. Handb Clin Neurol 2018;155:143-74.
7. Jacobi H, du Montcel ST, Bauer P, Giunti P, et al. Long-term evolution of patient-reported outcome measures in spinocerebellar ataxias. J Neurol 2018;265(9):2040-51.
8. Ashizawa T, Xia G. Ataxia. Continuum 2016;22(4 Movement Disorders):1208-26.
9. Németh AH, Kwasniewska AC, Lise S, et al. Next generation sequencing for molecular diagnosis of neurological disorders using ataxias as a model. Brain 2013;136(pt 10):3106-18.
10. Zeigelboim BS, Teive HA, Santos RS, et al. Audiological evaluation in spinocerebellar ataxia. Codas 2013;25(4):351-7.
11. Abe T, Abe K, Aoki M, Itoyama Y, Tamai M. Ocular changes in patients with spinocerebellar degeneration and repeated trinucleotide expansion of spinocerebellar ataxia type 1 gene. Arch Ophthalmol 1997;115:231-6.
12. Ahn JK, Seo JM, Chung H, Yu HG. Anatomical and functional characteristics in atrophic maculopathy associated with spinocerebellar ataxia type 7. Am J Ophthalmol 2005;139(5):923-5.
13. Bettencourt C, Lima M. Machado-Joseph disease: From first descriptions to new perspectives. Orphanet J Rare Dis 2011 Jun 2:6:35. doi: 10.1186/1750-1172-6-35.
14. Klockgether T, Mariotti C, Paulson HL. Spinocerebellar ataxia. Nat Rev Dis Primers 2019;5(1):24. doi: 10.1038/s41572-019-0074-3.
15. Camey S, Jardim LB, Kieling C, Saute JA, Vigo A. A prospective study of SCA3 gait ataxia described through a Markovian method. Neuroepidemiol 2010;34(3):163-70.
16. D'Abreu A, Franca MC Jr, Paulson HL, Lopes-Cendes I. Caring for Machado-Joseph disease: Current understanding and how to help patients. Parkinsonism Relat Disord 2010 Jan;16(1):2-7. doi: 10.1016/j.parkreldis.2009.08.012.
17. Protasova MS, Grigorenko AP, Tyazhelova TV, et al. Whole-genome sequencing identifies a novel ABCB7 gene mutation for X-linked congenital cerebellar ataxia in a large family of Mongolian ancestry. Eur J Hum Genet 2016;24(4):550-5.
18. Pierson TM, Adams D, Bonn F, et al. Whole-exome sequencing identifies homozygous AFG3L2 mutations in a spastic ataxia-neuropathy syndrome linked to mitochondrial m-AAA proteases. PLoS Genet 2011;7(10):e1002325 doi:10.1371/journal.pgen.1002325.
19. Grobe-Einsler M, Vogt IR, Schaprian T, Hurlemann R, et al. Effects of rivastigmine on patients with spinocerebellar ataxia Type 3: A case series of five patients. Neurodegener Dis 2020;20(2-3):104-9.
20. Henderson EJ, Lord SR, Close JC, Lawrence AD, et al. The ReSPonD trial--rivastigmine to stabilise gait in Parkinson's disease a phase II, randomised, double blind, placebo controlled trial to evaluate the effect of rivastigmine on gait in patients with Parkinson's disease who have fallen. BMC Neurol 2013 Dec 3;13:188. doi: 10.1186/1471-2377-13-188.
21. Moscovich M, Okun MS, Favilla C, Figueroa KP, et al. Clinical evaluation of eye movements in spinocerebellar ataxias: A prospective multicenter study. J Neuroophthalmol 2015;35(1):16-21.

22. Coetzee D, Pienaar AE. The effect of visual therapy on the ocular motor control of seven- to eight-year-old children with developmental coordination disorder (DCD). Res Dev Disabil 2013;34(11):4073-84.
23. Shandiz JH, Riazi A, Khorasani AA, Yazdani N, et al. Impact of vision therapy on eye-hand coordination skills in students with visual impairment. J Ophthalmic Vis Res 2018 ;13(3):301-6.

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