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Spinocerebellar ataxia type 8 presenting as parkinsonism syndrome: a case report

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Introduction

Spinocerebellar Ataxia Type 8 (SCA8) is a genetic disorder caused by a trinucleotide repeat expansion in the ATXN8OS/ATXN8 gene, primarily presenting with ataxia and gait instability. While Parkinsonism symptoms such as bradykinesia, rigidity, and gait disturbances can also be observed in SCA8, the disease is often misdiagnosed as Parkinson's disease (PD). The overlap of symptoms between these conditions can complicate the diagnosis, especially in the absence of characteristic signs like rest tremor and a poor response to dopaminergic therapy. In these cases, genetic testing becomes essential for accurate diagnosis and to differentiate SCA8 from other movement disorders including PD.

Case

A 59-year-old woman with a history of diffuse large B-cell lymphoma (DLBCL) and right breast cancer presented with progressive gait disturbance, balance loss, and left lower limb pain. Symptoms began in September 2023 and worsened after chemotherapy and autologous stem cell transplant for DLBCL in June 2024, eventually leading to wheelchair dependence. PD was initially suspected due to her gait slowness, rigidity, and functional decline. However, normal FP-CIT PET findings, showing preserved striatal uptake, and absence of basal ganglia pathology on brain MRI helped rule out PD as the cause. In September 2025, a spine MRI and electromyography confirmed lumbar pathology, including bilateral L5 radiculopathy, which was linked to her lower limb pain. Lumbar nerve root injections were administered to address the pain, but her gait instability persisted. Upon admission, manual muscle test revealed good grade in both upper and lower limbs, but the berg balance scale score was 5/56 points, showing a significant reduction in balance relative to her muscle strength, which indicated a central nervous system issue affecting her coordination and gait. Although no significant abnormalities were observed on the brain MRI (Figure 1) and the previous brain PET scan was also negative, further investigation was conducted given the patient’s medical history. Paraneoplastic antibody testing was performed to exclude issues related to her previous cancers, and the results were negative. Therefore, the possibility of a genetic disorder was investigated. Subsequently, genetic testing was done, which confirmed SCA8, explaining her gait instability and ataxia. Importantly, there was no family history of similar conditions. However, considering the possibility of incomplete penetrance, it is possible that the patient ' s family carries the gene repeat but does not show symptoms, which could explain the sporadic presentation in this patient. The final diagnosis was SCA8 with bilateral L5 radiculopathy, which contributed to both her motor symptoms and lower limb pain. Treatment was adjusted accordingly, and the diagnosis of SCA8 helped guide future management, focusing on gait stability and symptom control.

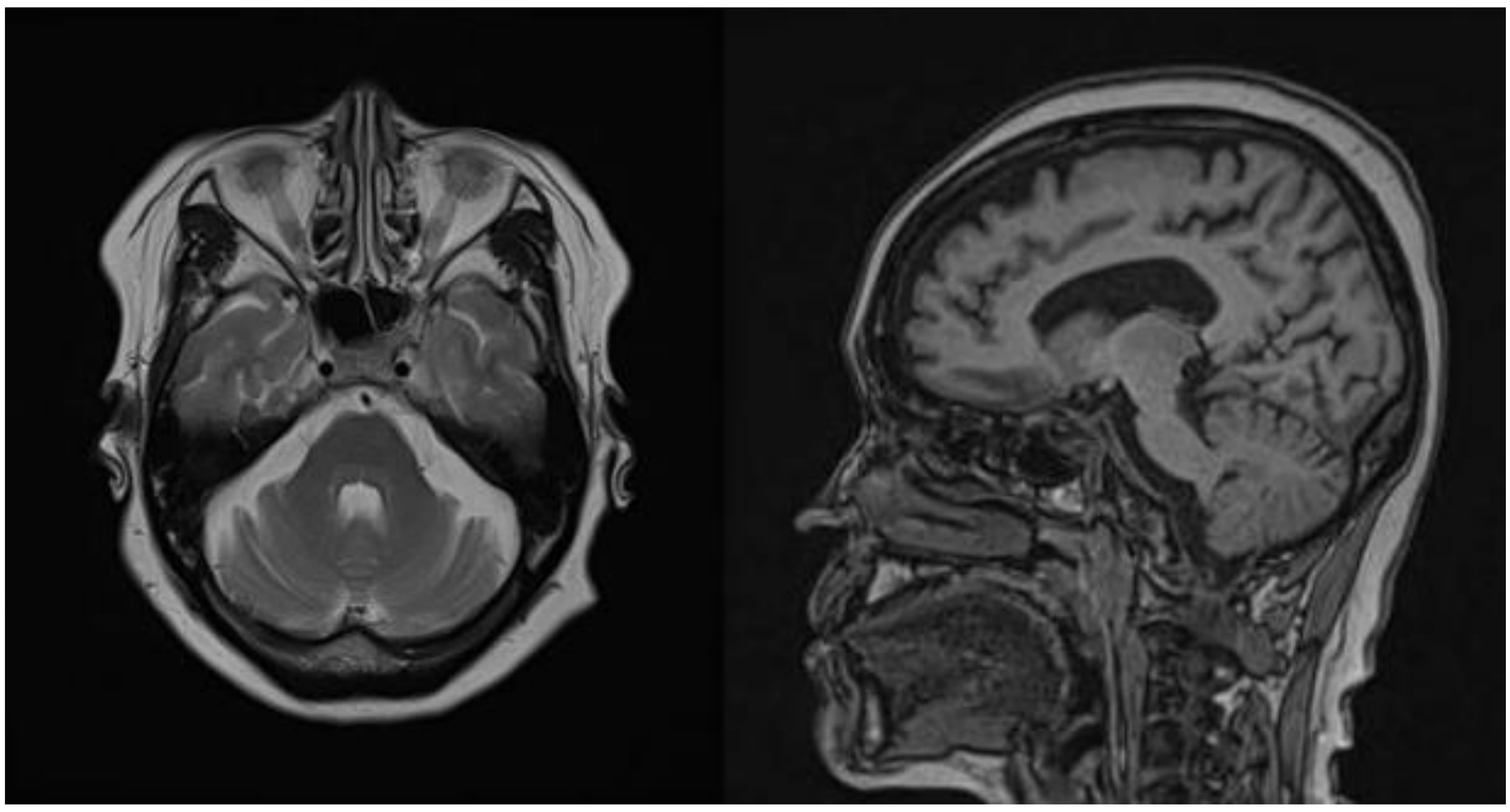

Figure 1.
MRI of the brain revealed no cerebellar atrophy.

Conclusion

This case highlights the critical importance of a thorough diagnostic approach, combining clinical evaluation, imaging, and genetic testing. The presence of parkinsonism-like symptoms in SCA8, such as gait instability and ataxia, can easily lead to diagnostic confusion, making early genetic testing essential for an accurate diagnosis. Identifying SCA8 through genetic analysis not only aids in appropriate management but also facilitates genetic counseling, especially in cases with no family history. This case underscores the need for careful consideration of rare neurodegenerative diseases when common diagnoses do not fully explain the patient’s symptoms, ensuring better treatment outcomes and patient care.