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Letter to the Editor

The Diagnosis of Spinocerebellar Ataxia Requires Proof of a Corresponding Genetic Defect

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Letter to the Editor

We read with interest the article by Monteiro *et al*. about a 56-year-old man with spinocerebellar ataxia (SCA) of unknown cause, diagnosed shortly after SARS-CoV-2 infection (SC2I) in June 2020, who underwent physiotherapy treatment using baropodometry and stabilometry as well as a biophysical-social assessment with positive results [1]. The patient showed improvement in vertigo symptoms, no significant changes in balance or mobility, but normalization of bilateral contact area with eyes closed, improvement in the plantar arch index of the left foot with eyes open, and an increase in the pressure center area and mean oscillation velocity under both visual conditions, indicating a decrease in postural stability [1]. The vertigo handicap index improved by almost 50%, particularly in the physical domain [1]. The study is interesting, but some points should be discussed.

The first point is that we disagree with the diagnosis of SCA [1]. SCAs primarily affect the cerebellum and spinal cord, leading to progressive problems with coordination, balance, and movement. SCAs are clinically characterized by progressive ataxia, unsteadiness of gait, slurred speech (dysarthria), difficulty swallowing (dysphagia), and in some cases cognitive impairment or sensory disturbances in the limbs [2]. However, the index patient did not exhibit dysphagia, or cognitive deficits [1]. The patient had no family history of SCA and no genetic testing was performed to confirm the diagnosis. It is also noteworthy that symptoms such as balance disorders, gait disturbances, and falls coincided with a SARS-CoV-2 infection (SC2I) in June 2020, suggesting either an infection of the central nervous system or an immunological reaction (e.g., cerebellitis, brainstem encephalitis, autoimmune encephalitis, transverse myelitis) or a peripheral nervous system disorder (e.g., radiculitis, polyneuropathy) were actually responsible for the clinical presentation. Did the patient undergo a neurological examination, cerebrospinal fluid analysis, and cerebral imaging using contrast-enhanced MRI in June 2022? Were antibodies associated with immune encephalitis detected in June 2022? Infectious and immunological encephalitis and myelitis are common complications of SC2I [3].

The second point is that no cerebral imaging data were reported [1]. In order to assess whether a structural lesion was present and how this manifested clinically, it would have been essential to describe or present cerebral imaging using computed tomography or magnetic resonance imaging. Of particular interest is whether or not there were any residuals of the suspected cerebral involvement in SC2I.

The third point is that the patient apparently had paraparesis and decreased tendon reflexes in the lower extremities, but no adequate explanation was given for this [1]. Was the paraparesis due to SCA, neuropathy, plexopathy, myopathy, or myelopathy? Were nerve conduction studies performed on the patient to determine whether peripheral nerves were affected or not? To distinguish between a central and a peripheral cause of paraparesis, it would be useful to perform an MRI of the spine. Knowledge of the cause of paraparesis and SCA in general is crucial for the design of rehabilitation and the overall course of the disease.

The fourth point is that no explanation was given as to why the patient suffered from rigor [1]. SCA is not usually associated with Parkinson's disease, suggesting that the patient either had an additional condition or that the actual condition was not SCA. Since there was a family history of Parkinson's disease, it is conceivable that the index patient also suffered from Parkinson's disease. It is also conceivable that Parkinson's disease was a complication of SC2I-associated encephalitis.

Overall, the diagnosis of SCA should be based on a positive genetic test and the correlation between genotype and phenotype. Symptoms and signs shortly after SC2I are more indicative of an infectious or immunological cause than SCA.

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