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(CASE REPORT)

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Resolution of acute cerebellar ataxia with vitamin B12 supplementation in a 19-year-old female with normal vitamin B12 levels

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Abstract

Acute cerebellar ataxia, characterized by incoordination of movement and gait instability, presents a complex diagnostic challenge for clinicians. Vitamin B12 deficiency is a well-established cause of neurological dysfunction, including ataxia. However, diagnosis can be complicated by the presence of normal serum vitamin B12 levels. This case report presents an unusual presentation of acute cerebellar ataxia in a young woman with normal vitamin B12 levels. The case highlights the importance of considering a broader diagnostic approach and the potential therapeutic benefit of vitamin B12 supplementation even in the absence of overt vitamin B12 deficiency.

Keywords: Acute cerebellar ataxia; Vitamin B12; Vitamin B12 deficiency; Vitamin B12 supplementation; Neurological manifestations; Neurological disorder

1. Introduction

Vitamin B12 (also known as cobalamin) is an essential water-soluble nutrient vital for hematopoiesis, DNA synthesis, energy metabolism, homocysteine regulation, and myelination, underscoring its paramount significance in maintaining overall health [1]. Severe deficiency primarily stems from inadequate consumption of animal-derived foods and the development of pernicious anemia, resulting from autoimmune atrophic gastritis leading to intrinsic factor loss, affecting both children and adults globally [2].

Current research highlights the health implications of vitamin B12 deficiency, especially among vegetarian populations, during pregnancy, in the elderly, and in developing nations. This deficiency is a well-established cause of hematological and neuropsychiatric disorders, including peripheral neuropathy, subacute combined degeneration of the spinal cord, dementia, cerebellar ataxia, and optic atrophy [3]. Notably, neurologic symptoms can manifest even without anemia or macrocytosis, and the absence of these hematologic changes does not rule out vitamin B12 deficiency, as illustrated by this case of acute cerebellar ataxia with normal vitamin B12 levels, which demonstrated rapid improvement with vitamin B12 supplementation.

2. Case description

A 19-year-old African American female with no past medical history presented with complaints of new-onset progressive tremors, difficulty walking and slurred speech for the past 3 weeks. She denied any trauma and reported going on a cruise to Honduras for 5 days around 4 weeks prior to her symptom onset. The patient denied any sick contact or any symptoms while she was on the cruise. She reported taking oral contraceptives and no other medications. The rest of her history including surgical, family, and social was unremarkable. The physical examination was suggestive of cerebellar ataxia, central tremor and dysarthria.

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Neurology and Infectious Disease were consulted upon admission. The initial laboratory investigation only showed microcytic anemia. Non-contrast computed tomography (CT) scan of the head, computed tomography angiography (CTA) scan of the brain, magnetic resonance imaging (MRI) with/without contrast of the brain, and magnetic resonance imaging (MRI) of the spinal cord normal anatomy and no evidence of acute pathology as shown in figures 1, 2, 3, and 4. Extensive work-up including cerebrospinal fluid (CSF) analysis, heavy metals screen, alcohol & drug screen, paraneoplastic panel, autoimmune panel, and CT chest & abdomen/pelvis with/without contrast were unremarkable. Multiple myeloma, neuro-sarcoidosis, Human Immunodeficiency Virus (HIV), syphillis, and lyme disease were also ruled out. Serum vitamin B12 level was 390 pg/mL (reference range: 211-911 pg/mL) and serum methylmalonic acid level was 103 nmol/L (reference range: 0-378 nmol/L).

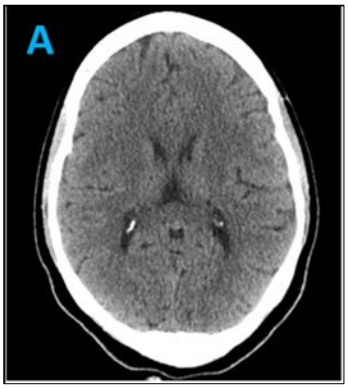

Figure 1 Non-contrast computed tomography (CT) scan of the head: demonstrates normal anatomy and no evidence of acute intracranial pathology; this includes a normal size of the brain for the patient's age, gray-white matter differentiation is maintained, no mass effect, no midline shift, or hydrocephalus

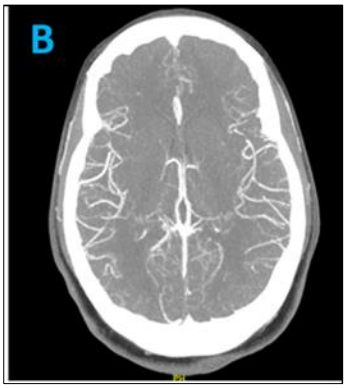

Figure 2 Computed tomography angiography (CTA) scan of the brain: shows no evidence of abnormalities in the major blood vessels supplying blood to the brain including no evidence of occlusion, hemodynamically significant stenosis, aneurysm or dissection

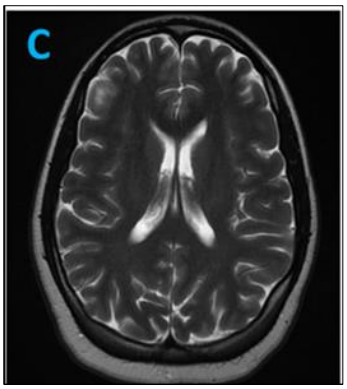

Figure 3 Magnetic resonance imaging (MRI) with/without intravenous (IV) contrast of the brain: demonstrates normal anatomy and no evidence of pathology, including unremarkable ventricles and subarachnoid spaces in size for the patient's given age, nor evidence of mass effect, midline shift or abnormal extra-axial fluid mass; additionally, there is no evidence of abnormal enhancement following intravenous contrast administration

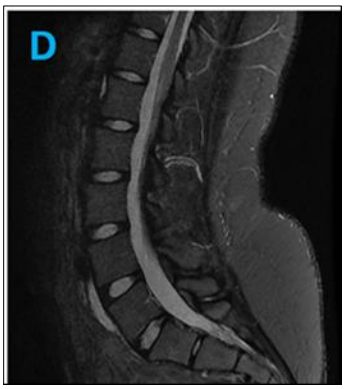

Figure 4 Magnetic resonance imaging (MRI) with/without IV contrast of the spinal cord: showed no abnormalities in the spine including normal height and appearance of the vertebral bodies & intervertebral discs, no evidence of soft tissue edema or disc herniation, no evidence of spinal or foraminal stenosis, no epidural collection or areas of abnormal enhancement on postcontrast images

The patient was initially started on high dose intravenous (IV) steroids and IV thiamine with no improvement in her symptoms. Psychiatry was consulted and no psychiatric issues were identified. The decision was made to administer parenteral vitamin B12 (cobalamin) that resulted in significant improvement of her symptoms within a few days. She was then discharged to a rehabilitation facility where she continued to improve clinically and was subsequently discharged home.

3. Discussion

Vitamin B12 deficiency is a well-recognized cause of neurological dysfunction, manifesting with a wide range of symptoms including ataxia, neuropathy, and cognitive decline [1]. However, a proportion of patients may present with neurological symptoms despite normal serum vitamin B12 levels. Diagnosis is typically based on a combination of clinical presentation, laboratory findings, and response to treatment [4]. Serum vitamin B12 levels are a cornerstone of diagnosis, but a significant proportion of patients, particularly those with early deficiency, may present with normal vitamin B12 levels [5]. This phenomenon, known as functional vitamin B12 deficiency, is attributed to impaired cellular uptake and utilization of vitamin B12 despite seemingly adequate serum concentrations [6].

Vitamin B12 supplementation has been previously used by some clinicians to successfully treat symptoms of peripheral neuropathy in patients with normal vitamin B12 levels, but acute cerebellar ataxia of unknown etiology responding to parenteral vitamin B12 supplementation has not been reported in the medical literature. The exact mechanism by which vitamin B12 supplementation improved the patient's symptoms in this case remains unclear. It's possible that borderline vitamin B12 deficiency, not detected by the initial test, was present. Alternatively, vitamin B12 supplementation may have addressed an underlying functional vitamin B12 deficiency despite normal serum levels.

Studies suggest that functional vitamin B12 deficiency can be caused by various factors, including genetic polymorphisms affecting vitamin B12 transport proteins or coexisting deficiencies in folate or other nutrients [7].

The decision to initiate vitamin B12 supplementation in our patient with normal vitamin B12 levels was based on the clinical suspicion of vitamin B12 deficiency and the potential for a therapeutic trial to be both safe and informative. This approach aligns with recommendations suggesting vitamin B12 supplementation can be a valuable diagnostic tool, particularly in patients with a high clinical suspicion [8]. As demonstrated in this case, a positive response to vitamin B12 supplementation can provide strong evidence for underlying vitamin B12 deficiency, even if initial serum levels are within the reference range. However, we need further research to explore pathophysiology of vitamin B12 deficiency and redefine appropriate cutoff levels of serum B12. We hope to contribute to the current literature and bring awareness to the medical community

4. Conclusion

This case report highlights the potential benefit of vitamin B12 supplementation in resolving acute cerebellar ataxia, even in the presence of normal serum vitamin B12 levels. It emphasizes the importance of considering a broader diagnostic approach that includes functional vitamin B12 deficiency and the potential utility of a therapeutic trial with vitamin B12 supplementation when clinical suspicion is high. However, this is a single case report, and generalizability of the findings is limited. Further research is needed to understand the role of vitamin B12 supplementation in patients with normal vitamin B12 levels presenting with ataxia. Additionally, exploring potential causes of functional vitamin B12 deficiency in such cases would be valuable.

Compliance with ethical standards

Disclosure of conflict of interest

No conflict of interest to be disclosed.

Statement of informed consent

Informed consent was obtained from all individual participants included in the study.

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