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Acute Intermittent Porphyria Associated With Transverse Myelitis In A Young Patient : A Case Report

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Abstract

We report the case of a 30-year-old man with a progressive descending symmetric paresis, myalgia, and action tremor, initially investigated as Guillain-Barré syndrome. The absence of alterations in the initial tests and clinical worsening prompted treatment with intravenous immunoglobulin, followed by methylprednisolone and plasmapheresis, without satisfactory response. Neuroimaging suggested transverse myelitis, and the persistence of symptoms led to a metabolic investigation, which confirmed acute intermittent porphyria. The patient was referred for rehabilitation and specialized follow-up, showing gradual improvement. This case highlights the importance of including porphyria in the differential diagnosis of transverse myelitis in patients with refractory symptoms and atypical systemic manifestations.

Keywords: Acute Intermittent Porphyria; Transverse Myelitis; Rare Metabolic Diseases; Neuropathy; Differential Diagnosis

Introduction

Transverse myelitis (TM) is a rare and potentially debilitating neurological syndrome characterized by segmental inflammation of the spinal cord, resulting in motor, sensory, and autonomic deficits. The etiology of TM is multifactorial, involving autoimmune, infectious, post-vaccination processes, and, more rarely, metabolic and genetic conditions. Among these uncommon causes, the association with porphyrias stands out a heterogeneous group of hereditary disorders resulting from enzymatic deficiencies in specific steps of heme biosynthesis. Acute intermittent porphyria (AIP) is the most common form of acute porphyrias and presents with neurovisceral crises, including abdominal pain, vomiting, peripheral neuropathy, psychiatric disturbances, and autonomic dysfunction. During AIP crises, the accumulation of precursors such as delta-aminolevulinic acid (ALA) and porphobilinogen (PBG) in the central and peripheral nervous systems exerts direct neurotoxic effects, leading to clinical pictures that mimic other neurological diseases such as Guillain-Barré syndrome and transverse myelitis itself.

Although this association is rare and underreported in the literature, early identification is crucial for effective management and prevention of permanent neurological sequelae. Diagnosis can be challenging, especially in patients with atypical symptoms or who are refractory to conventional immunomodulatory therapies. Additionally, triggering factors

such as infections, surgical stress, medications, and prolonged fasting are common in porphyria patients and often precede the onset of acute crises. Treatment involves withdrawal of precipitating factors, intravenous administration of hemin, and appropriate nutritional support. The lack of clinical response to standard treatments—such as immunoglobulin, corticosteroids, or plasmapheresis should alert clinicians to underlying metabolic causes. In this context, the present case report describes a young patient with progressive paresis and autonomic symptoms initially suggestive of classic demyelinating diseases, but who, after thorough investigation, was diagnosed with neurological manifestations of acute intermittent porphyria. This case reinforces the importance of considering metabolic differential diagnoses in patients with refractory transverse myelitis, highlighting the role of rare diseases in the neurological spectrum.

Objectives

This study aims to report a clinical case of a young patient diagnosed with acute intermittent porphyria associated with transverse myelitis.

Materials And Methods

A retrospective case report was conducted through review of the electronic medical record, along with a brief literature review.

Case Report

A 30-year-old male patient was admitted with symmetric descending paresis, myalgia, and progressively worsening action tremor, eventually leading to inability to walk after more than 15 days of evolution. He reported a recent flu-like illness and postoperative recovery from cholecystectomy for biliary pancreatitis. Initially, Guillain-Barré syndrome was suspected; however, initial laboratory tests, CT scans, and cerebrospinal fluid analysis were unremarkable. Due to clinical worsening, intravenous immunoglobulin (IVIG) therapy was initiated and the patient was transferred to the intensive care unit. Despite slight initial improvement, the patient developed dysphagia, hyporeflexia, and worsening motor function. Follow-up neuroimaging suggested possible transverse myelitis, and high-dose methylprednisolone was administered, but without clinical response. Due to the persistent clinical picture, the patient was transferred to a tertiary hospital with more resources, where plasmapheresis was performed, also without improvement. After extensive investigations, metabolic evaluation revealed alterations in plasma and urinary porphyrin precursors, confirming the diagnosis of porphyria. The patient was then referred for rehabilitation, treatment, and specialized follow-up at a reference center, showing gradual improvement.

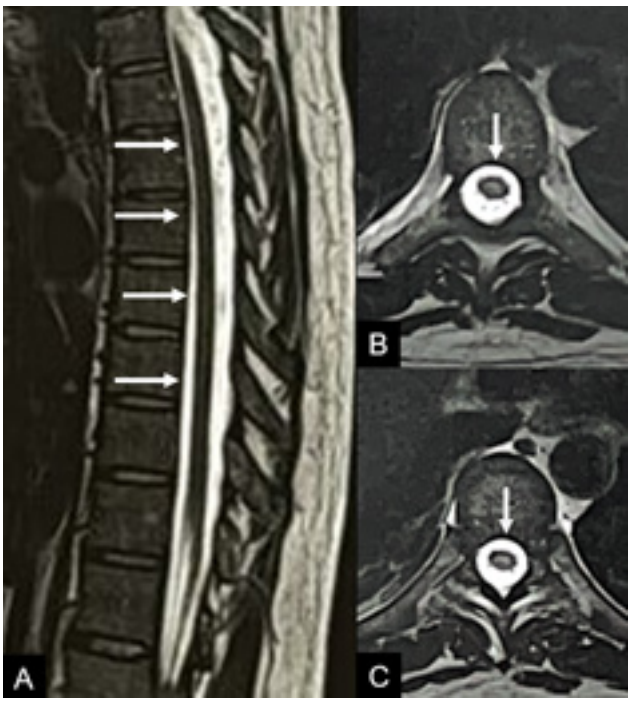

Figure 1: Sagittal (A) and axial (B) T2-weighted images of the thoracic spine, showing a slight area of signal alteration in the central portion of the spinal cord (arrows in A and B).

Discussion

Acute intermittent porphyria (AIP) is a rare autosomal dominant metabolic disorder resulting from partial deficiency of porphobilinogen deaminase, a key enzyme in the heme biosynthesis pathway. Although classically characterized by neurovisceral crises with abdominal pain, psychiatric symptoms, and peripheral neuropathy, central nervous system involvement remains underrecognized and poorly understood. The present case illustrates an uncommon but clinically significant association between AIP and transverse myelitis (TM), reinforcing the importance of considering metabolic etiologies in patients with atypical or refractory myelopathic syndromes.

Neurological manifestations of AIP are predominantly attributed to the neurotoxic effects of accumulated heme precursors, particularly delta-aminolevulinic acid (ALA) and porphobilinogen (PBG). These metabolites exert direct neurotoxicity through oxidative stress, mitochondrial dysfunction, impaired axonal transport, and disruption of gamma-aminobutyric acid (GABA) neurotransmission. While peripheral motor neuropathy is the most frequently reported neurological complication, central involvement such as seizures, encephalopathy, and, more rarely, spinal

cord dysfunction has been described. The pathophysiological mechanisms underlying myelitis in AIP may involve endothelial dysfunction, microvascular ischemia, and metabolic toxicity rather than classic immune-mediated inflammation, which may explain the poor response to immunomodulatory therapies observed in this case.

The clinical presentation of progressive symmetric paresis initially raised suspicion for Guillain-Barré syndrome, a frequent diagnostic consideration in patients with acute flaccid paralysis. However, the absence of albuminocytologic dissociation in cerebrospinal fluid analysis, normal initial neuroimaging, and lack of sustained response to intravenous immunoglobulin therapy suggested an alternative diagnosis. Subsequent neuroimaging findings compatible with transverse myelitis led to corticosteroid therapy and plasmapheresis, yet clinical refractoriness persisted. This therapeutic failure highlights a crucial diagnostic pitfall, as standard treatments for immune-mediated demyelinating disorders are ineffective in metabolic neuropathies such as AIP and may delay appropriate management.

Several triggering factors commonly associated with AIP crises were present in this patient, including recent infection, surgical stress, and postoperative fasting. These stressors likely increased hepatic heme demand, upregulated aminolevulinic acid synthase activity, and precipitated accumulation of neurotoxic intermediates. Recognition of such precipitating events is essential, particularly when neurological deterioration occurs in temporal association with systemic stressors and in the absence of clear autoimmune or infectious markers.

Although the association between AIP and TM is rarely reported, emerging evidence suggests that porphyria-related spinal cord involvement may be underdiagnosed, especially in cases classified as idiopathic or atypical myelitis. In such scenarios, metabolic screening for porphyria should be considered, particularly in patients presenting with multisystemic manifestations, autonomic dysfunction, hyponatremia, unexplained abdominal pain, or poor response to immunosuppressive therapies. Early diagnosis is critical, as prompt treatment with hemin, carbohydrate loading, and elimination of triggering factors can halt disease progression and significantly improve neurological outcomes.

This case underscores the need for heightened clinical suspicion and a multidisciplinary approach in evaluating acute myelopathies. Expanding the differential diagnosis to include rare metabolic disorders such as AIP may prevent misdiagnosis, avoid unnecessary and potentially harmful treatments, and facilitate timely institution of disease-specific therapy. Ultimately, recognizing porphyria as a potential cause of transverse myelitis contributes to improved patient outcomes and broadens the current understanding of the neurological spectrum of acute porphyrias.

Conclusion

This case emphasizes the importance of broadening the diagnostic spectrum of transverse myelitis to include rare metabolic causes, such as porphyria, in patients with neurological conditions refractory to conventional therapies and suggestive biochemical alterations. Although typically attributed to autoimmune or infectious processes, myelitis may result from dysfunction in the heme biosynthesis pathway. Undoubtedly, treatment with withdrawal of triggering factors, use of hemin, and metabolic support is essential to reverse the condition and prevent permanent sequelae.

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