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A novel case of transverse myelitis revealing undiagnosed McArdle disease

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Abstract

A 43-year-old woman contracted a presumed gastroenteritis and developed ascending numbness a week later. She had an unremarkable serum lab and spinal fluid evaluation and normal MRI brain. However, MRI of the spine showed an initial T7 lesion, and an additional C5–C6 lesion one month later. She was diagnosed with idiopathic transverse myelitis. Several months following this episode, she was hospitalized on 2 separate occasions for severe back pain following exertion. CK was elevated during both admissions (26,000 U/L and 40,000 U/L, respectively). A muscle biopsy of her right thigh showed absent myophosphorylase and increased glycogen stores, consistent with McArdle disease. Upon further questioning, the patient disclosed life-long exercise intolerance, contractures, and improved exercise capacity after rest. McArdle disease and transverse myelitis appeared to occur independently in the patient, but the truncal numbness from transverse myelitis might have masked the myopathic pain and her subjective warning signs of contractures, leading to her later hospitalizations for rhabdomyolysis.

Keywords

McArdle disease; transverse myelitis; contractures; rhabdomyolysis; truncal numbness.

Introduction

Transverse myelitis is a neuro-inflammatory disorder of the spinal cord that manifests with acute or subacute weakness, sensory deficits, and autonomic dysfunction. Causes of transverse myelitis include demyelinating disease (e.g. multiple sclerosis, neuromyelitis optica), post-infectious sequelae, systemic autoimmune disease, paraneoplastic processes, and toxin or drug exposures [1,2]. On MRI, transverse myelitis presents as T2-weighted hyperintense signals in the spinal cord in the absence of a compressive cord lesion. Cerebrospinal fluid studies may show oligoclonal bands, elevated IgG, or pleocytosis.

McArdle disease (myophosphorylase deficiency, glycogen storage disease V) is an autosomal recessive disorder of carbohydrate metabolism caused by a deficiency of myophosphorylase, which is responsible for liberating glucose from glycogen in skeletal muscle. McArdle disease is associated with exercise intolerance, characterized by early fatigue, muscle contractures, myoglobinuria, and rhabdomyolysis with trivial levels of exertion. Patients with McArdle disease also experience a "second-wind" phenomenon, with improvement in exercise capacity after ~8 minutes of aerobic exercise due to vascular delivery of liver-derived glucose to skeletal muscle [3,4].

Case Presentation

A 43-year-old woman presented to the emergency department with two days of ascending numbness and tingling from her feet to her waist, a week after an episode of presumed acute gastroenteritis. Serum and cerebrospinal fluid studies were unremarkable, and MRI of the brain showed no lesions. Spinal MRI showed a T2 hyperintense lesion in the posterior central spinal cord at T7, consistent with transverse myelitis. She received steroids and was discharged with improved symptoms. One month later, she developed new right-sided numbness, and a repeat MRI of the spine revealed a new spinal cord lesion at C5-C6 in addition to the stable T7 lesion. She was again given steroids and discharged after clinical improvement.

After the second hospitalization, she developed severe back pain after lifting some heavy objects, and again presented to the emergency department. Laboratory evaluation revealed rhabdomyolysis with CK elevation to 26,000 U/L. She was treated with intravenous fluids and discharged with a down trending CK.

Two months later, she was again admitted with rhabdomyolysis, with a CK of 40,000 U/L. This time, she had developed back pain and pigmenturia after participating in physical therapy. A muscle biopsy was obtained, which revealed absent myophosphorylase staining, leading to the diagnosis of McArdle disease (Figure 1). On further questioning, she did report a lifelong history of exercise intolerance, with episodes of muscles "locking up", early muscle fatigue, pigmenturia, and the "second wind" phenomenon.

Her physical exam at an outpatient clinic follow up was significant for normal strength, reduced sensation to pinprick, temperature, and proprioception in the toes, and increased reflexes with bilateral Hoffman's and crossed adductors at the knees.

MRI of the spine approximately two years after her initial transverse myelitis diagnosis did show a new C2 lesion, and spinal fluid analysis revealed elevated oligoclonal bands and IgG synthesis rate, leading to the diagnosis of presumed spinal predominant multiple sclerosis, which is currently being managed with ocrelizumab (Ocrevus; Genentech, South San Francisco, CA).

Additionally, she continues to have frequent episodes of rhabdomyolysis, and has had several additional hospitalizations for treatment with IV fluids.

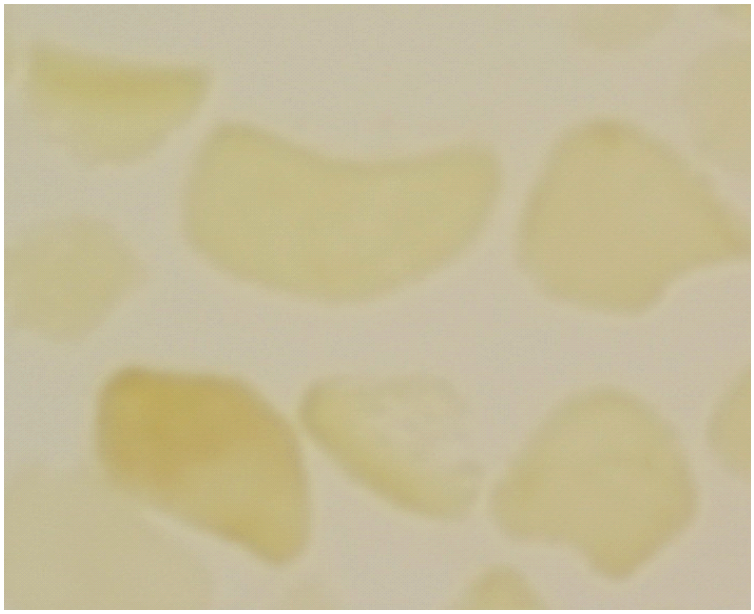

Figure 1: Myophosphorylase stain showing absence of myophosphorylase activity.

Discussion

This report details a case of a patient with undiagnosed McArdle disease and comorbid recurrent transverse myelitis who first began to recognize episodes of rhabdomyolysis only after the diagnosis of transverse myelitis. To our knowledge, this is the first reported case of a patient who presented with transverse myelitis and subsequently developed recurrent rhabdomyolysis, secondary to previously undiagnosed McArdle disease.

We theorize that diminished sensation secondary to transverse myelitis might have interfered with the case patient's ability to sense impending muscle contractures, which normally would have triggered rest and activation of the "second wind" phenomenon. Furthermore, we also question whether spasticity related to her spinal cord lesions could result in increased metabolic demand in skeletal muscle that could lower the threshold for developing muscle contractures and rhabdomyolysis.

We detail this case in order to increase awareness for the rare co-morbidity of McArdle disease and recurrent transverse myelitis, and to discuss how central nervous system lesions may negatively impact the disease course in patients with coexistent McArdle disease.

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