01. Beyond Polymyositis: Refractory Lumbar Spasms Revealing Underlying Isaac Syndrome

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Background: Polymyositis is an idiopathic inflammatory myopathy characterized by proximal muscle weakness, elevated creatine kinase (CK), and immune-mediated pathogenesis. Atypical features such as refractory shock-like paraspinal spasms should prompt consideration of a coexisting diagnosis. Isaac syndrome (neuromyotonia) is a rare autoimmune peripheral nerve hyperexcitability disorder classically associated with voltage-gated potassium channel (VGKC) antibodies, though antibodies against glutamic acid decarboxylase 65 (anti-GAD65) have also been implicated in peripheral nerve hyperexcitability syndromes. The clinical overlap between inflammatory myopathy and Isaac syndrome poses a significant diagnostic challenge, as their management and prognosis differ substantially. Early serological and clinical recognition is critical to guide appropriate immunotherapy and prevent systemic complications.

The Case: A 72-year-old male with inflammatory myopathy, scoliosis, and hypertension presented with repetitive shock-like lumbar spasms every 5 seconds. Neurological examination was otherwise unremarkable. Laboratory workup revealed CK 181 U/L, lactate 3.7 mmol/L, and mildly elevated troponin consistent with cardiac demand injury from sustained muscle hyperactivity. Importantly, anti-GAD65 antibodies returned positive at 6.9 nmol/L, providing serological support of an autoimmune neurological process. The disproportionately low CK relative to spasm severity, combined with anti-GAD65 seropositivity, pointed toward a neuromuscular rather than primary myopathic etiology. Initial treatment with intravenous methylprednisolone and intramuscular orphenadrine failed. Given clinical suspicion for autoimmune neuromuscular hyperexcitability, Isaac syndrome was diagnosed and plasma exchange (PLEX) was initiated. The patient demonstrated marked improvement in spasms, resolution of lactic acidosis, and normalization of cardiac biomarkers. He was discharged on prednisone taper and methotrexate with outpatient neurology and rheumatology follow-up. **Conclusion:** This case highlights that refractory shock-like spasms in the context of polymyositis should raise suspicion for coexisting autoimmune neuromyotonia. A low CK combined with poor steroid response are important clues toward an underlying peripheral nerve hyperexcitability syndrome. Anti-GAD65 seropositivity at high titer provided serological support for the autoimmune etiology and underscores the importance of a broad antibody workup in atypical inflammatory myopathy. Sustained involuntary muscle activity carries risk of serious metabolic complications including lactic acidosis and demand myocardial injury. Standard corticosteroids may be insufficient in GAD65-associated neuromyotonia, and early escalation to PLEX or other immunotherapies should be considered. A multidisciplinary approach involving neurology and rheumatology is essential to optimize outcomes.

Table 1. Polymyositis vs. Isaac Syndrome: Key Differentiating Features

Pathophysiology	
Immune-mediated muscle inflammation	
Autoimmune peripheral nerve hyperexcitability.	
Primary site	
Muscle (myopathy)	
Peripheral nerve (neuropathy)	
CK level	
Markedly elevated (often >1000 U/L)	
Normal or mildly elevated (181 U/L in this case)	
Spasm character	
Proximal weakness; spasms atypical	
Continuous, shock-like, every few seconds	
EMG pattern	
Myopathic units, fibrillations	
Myokymic / neuromyotonic discharges	
Key antibodies	
Anti-Jo-1, anti-Mi-2, anti-SRP	
Anti-VGKC (LGI1, CASPR2) or anti-GAD65; this patient: anti-GAD65 positive (6.9 nmol/L)	
Steroid response	
Usually responsive	
Often refractory; PLEX or IVIg needed	
Metabolic risk	
Low if treated	
High: lactic acidosis, demand cardiac injury	
Paraneoplastic	
Occasional	
~20% (thymoma most common, consider age-appropriate malignancy evaluation)	

VGKC: voltage-gated potassium channel; GAD65: glutamic acid decarboxylase 65; CK: creatine kinase; EMG: electromyography; PLEX: plasma exchange; IVIg: intravenous immunoglobulin. Anti-GAD65 antibodies (6.9 nmol/L) were positive in this patient, providing serological support for autoimmune neuromyotonia. EMG and VGKC-specific antibody testing were not performed; diagnosis was confirmed clinically and by response to PLEX.

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