

Conference

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Title

The Carpenter's Conundrum: From Muscle Weakness to Lipid Storage Myopathy

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Case Diagnosis

Lipid storage myopathy (LSM) due to impaired fatty acid oxidation

Case Description

A 70-year-old man with coronary artery disease, diabetic neuropathy, prior stroke (no deficits), and previous lumbar fusion presented to the ED with exertional dyspnea, increasing falls, and leg weakness. He is a retired carpenter and former smoker with unknown family history. Outpatient electrodiagnostic, thoracolumbar MRI, and cardiac stress testing were unremarkable. Examination revealed 4/5 hip flexor strength, ambulatory fatigue, and decreased foot sensation. Workup was negative except for mild, corrected hypokalemia. He was admitted to acute rehabilitation for presumed neuromuscular junction disorder requiring minimal assistance. Pulmonary function tests showed no evidence of restrictive lung disease. Myasthenia gravis and Lambert-Eaton antibodies were negative, so statin therapy was discontinued. Proximal thigh muscle edema on MRI, lipid accumulation on muscle biopsy, and elevated acylcarnitine (lipid derivative) confirmed a LSM. High-dose riboflavin and L-carnitine were started. After two weeks, he progressed to supervision-level function and was discharged home.

Discussion

This case demonstrates the complexity of diagnosing progressive proximal muscle weakness in older adults with comorbidities. Initial negative workup (A1C, CBC, BMP, inflammatory markers, muscle enzymes, thyroid, cortisol, respiratory viral panel, Lyme serology, chest radiograph, electrocardiogram, CT brain/pelvis, leg arterial/venous sonography) prompted testing of rare etiologies. Multiple acyl-CoA dehydrogenase deficiency and riboflavin-responsive myopathies are rare LSM associated with elevated acylcarnitines, and can mimic more common neuromuscular disorders. The patient's late onset proximal weakness is atypical and suggests a milder LSM with improved prognosis if treated promptly.

Conclusions

This case highlights the importance of a broad differential and thorough investigation. Clinicians should consider rare metabolic myopathies in the differential of unexplained proximal muscle weakness, particularly after excluding typical etiologies. Timely diagnosis allows for prompt initiation of disease-modifying therapy (riboflavin and L-carnitine supplementation), which can significantly improve function, quality of life, and guide patient-centered decision-making.