Normal Enzymes, Hidden Myopathy: A Case of Sporadic Late-Onset Nemaline Myopathy

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Introduction

Sporadic late-onset nemaline myopathy (SLONM) is a rare acquired myopathy of adulthood, characterized by progressive proximal and axial weakness with nemaline rods on muscle biopsy. Unlike congenital forms, SLONM typically presents after age 40 and may be associated with monoclonal gammopathy of undetermined significance (MGUS) or, less commonly, HIV infection. Postoperative onset mimicking disuse or radicular weakness is unusual and can delay diagnosis.

Objectives

To highlight an atypical presentation of SLONM following total knee replacement and emphasize the importance of muscle biopsy and secondary workup in patients with unexplained proximal weakness.

Methods

We describe a case of a 70-year-old man who developed progressive bilateral proximal lower extremity weakness beginning shortly after right total knee replacement. Clinical evaluation included physical examination, laboratory testing, MRI imaging, and muscle biopsy. Secondary workup for associated conditions such as MGUS and HIV was also performed.

Results

The patient presented with a four-month history of progressive bilateral proximal lower extremity weakness that began shortly after surgery, resulting in significant functional limitation, particularly with ambulation and stair climbing. He reported severe exertional leg pain (10/10) along with burning and tingling sensations, but denied systemic symptoms such as fever, weight loss, or fatigue. Neurologic examination demonstrated symmetric proximal muscle weakness in the lower extremities with preserved distal strength, intact deep tendon reflexes, and normal sensory testing.

Results

Laboratory evaluation revealed normal creatine kinase, aldolase, and inflammatory markers, with negative HMG-CoA reductase antibodies and positive anti-SSA 52kD antibodies of unclear significance. MRI of the thighs showed nonspecific distal muscle edema, mild patchy fatty infiltration, and postsurgical changes including suprapatellar effusion and synovitis, without evidence of focal muscle atrophy or tendon injury.

Given persistent unexplained weakness, a **muscle biopsy** was performed, demonstrating **denervation-type changes with the presence of nemaline rods**, confirming the diagnosis of SLONM.

Comprehensive secondary evaluation for associated conditions was negative, including serum protein electrophoresis, immunofixation, immunoglobulin levels, kappa/lambda ratio, and HIV testing.
Cardiac evaluation with echocardiography showed preserved left ventricular ejection fraction (55–60%) and mild tricuspid regurgitation.
A 24-hour urine protein study remains pending.

The patient was referred for physical therapy and continues to be monitored for disease progression and potential respiratory involvement.

Conclusion

SLONM should be considered in older adults presenting with progressive proximal weakness, even in postoperative settings where symptoms may be attributed to disuse or radiculopathy. Muscle biopsy is essential for diagnosis, and evaluation for associated conditions such as MGUS and HIV is critical for prognosis and management. Early recognition and multidisciplinary care are key to optimizing patient outcomes.