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RESEARCH ARTICLE

Acute Myositis as A Rare Manifestation of Mitochondrial Myopathy: Case Report

Dr Shobana Shiva*[1], Dr Sathyapriya SV[2], Dr Balambighai[3], Dr Padma V[4], Dr Gouri Gaur[1], Dr Gurucharan R[1] and Dr Albert Shaji[1]

[1]Postgraduate student, Department of General Medicine, Shree Balaji Medical College and Hospital, Chennai, Tamil Nadu, India
[2]Assistant Professor, Department of General Medicine, Sree Balaji Medical College and Hospital, Chennai, Tamil Nadu,
[3]Associate Professor, Department of Neurology Sree Balaji Medical College and Hospital, Chennai, Tamil Nadu, India
[4]Professor, Department of General Medicine, Sree Balaji Medical College and Hospital, Chennai, Tamil Nadu, India

*Corresponding Author
Shobana Shiva
(dr.shobana2022@gmail.com)

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Abstract: A 29-year-old male presented with acute onset of weakness of all four limbs, preceded by severe physical exertion. Upon examination, he was found to have proximal weakness of all four limbs (lower limbs >> upper limbs) and fully dependent for his ADL. Investigations showed elevated CPK levels. A provisional diagnosis of inflammatory myositis was made, and he was treated with IV methylprednisolone for 5 days, followed by a tapering dose of oral steroids. His upper and lower limb weakness improved and he was able to do ADL independently, but minimal proximal weakness persisted in both upper and lowerlimbs. On follow-up, he continued to have proximal weakness of the lower limbs. Hence, muscle biopsy was done which showed Few hypertrophic and atrophic fibres with Perifascicular atrophy and there was no granuloma or vasculitis suggestive of non-inflammatory myopathy. Electron microscopic picture of muscle biopsy showed Abundant mitochondria with few markedly enlarged forms (giant mitochondria) and shows many swollen cristae. Vessels were unremarkable, Endothelial tubuloreticular inclusions were not seen suggestive of mitochondrial myopathy He was advised genetic analysis, but he denied consent for genetic analysis. The patient is under close follow-up.This case is a rare presentation of mitochondrial myopathy as acute onset weakness.

Keywords: Mitochondrial myopathy, Acute myositis, Muscle biopsy, Electron microscopy, giant mitochondria with abnormal cristae.

INTRODUCTION

Mitochondrial myopathies are a heterogeneous group of neuromuscular disorders caused by defects in mitochondrial DNA or nuclear genes that affect oxidative phosphorylation (1). They typically present with progressive muscle weakness, exercise intolerance, or multisystem involvement. The clinical spectrum is broad and often overlaps with other neuromuscular conditions, which can make diagnosis challenging.

Acute myositis, characterized by sudden onset of muscle pain, tenderness, and weakness is more commonly associated with infectious, inflammatory, or autoimmune causes. Its occurrence in the context of mitochondrial myopathy is uncommon and may lead to diagnostic confusion, particularly in acute settings . Reporting such rare presentations is important to broaden clinical awareness and aid in timely recognition. This case highlights an unusual manifestation of mitochondrial myopathy presenting as acute myositis.

CASE PRESENTATION:

A 29-year-old male presented with an acute onset of pain in both shoulders and lower limbs for three days. He reported progressive difficulty in performing routine activities, including climbing stairs and lifting heavy-weight objects, which was preceded by diffuse myalgia and fatigue. He had history of lifting his autorickshaw and keeping the same over the thigh for 2 hrs for repair work.

On physical examination, the patient was alert and oriented. Vitals were stable. Neurological evaluation revealed symmetrical proximal muscle weakness, predominantly affecting the shoulder and hip girdle muscles. He had history of tremors for past 4 years, On clinical examination showed postural tremors and there was no intentional tremors. He has no co morbidities.

On examination, higher mental functions and cranial nerves were normal. The motor system showed proximal power of 3/5 in both upper limb , a power of 2/5 in both lower limbs and distal power was normal ,(5/5). DTRs were 1+, and bilateral plantar reflexes were flexor .There was muscle tenderness on palpation, and there was no cutaneous rash, joint swelling .The sensory system was normal.

Initial laboratory investigations showed markedly elevated serum creatine phosphokinase (CPK) levels, 505 U/L, and mild elevation of lactate and liver transaminases. ANA profile shows positive for PCNA. Complete blood count and renal function tests were within normal limits. Autoimmune serology, including ANA and myositis-specific antibodies, was

negative. Viral serologies for common myotropic viruses were also non-reactive.

MRI brain and whole spine screening was unremarkable. Nerve conduction study of all 4 limbs were within normal limit. EMG demonstrated short-duration, small-amplitude motor unit potentials with early recruitment patterns. USG abdomen and 2D echo Shows normal. Acute inflammatory myositis was considered and he was started on IV methylprednisolone, following which his symptoms improved, and he was discharged with tapering dose of oral steroids.

On followup, he continues to have proximal muscle weakness of lower limb hence muscle biopsy was done.

Based on clinical, histopathological, and genetic findings, a diagnosis of mitochondrial myopathy presenting as acute myositis was confirmed. The patient was managed with supportive therapy, and physiotherapy. Over the following weeks, gradual symptomatic improvement was noted, with partial recovery of muscle strength and reduction in CPK levels.

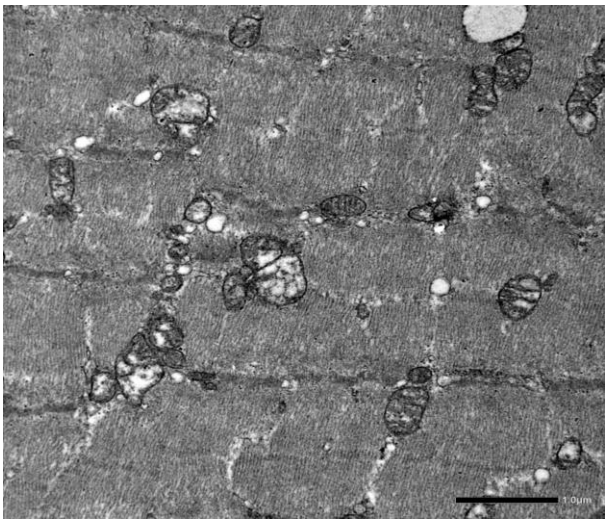

Figure 1 shows the electron microscopy image of skeletal muscle demonstrates fibers with largely preserved sarcomeric architecture but with abnormal mitochondrial accumulation. Several mitochondria appear enlarged, clustered, and show irregular morphology, some with disrupted cristae. Scattered vacuolated areas are also present, reflecting degenerative changes within the fibers. These alterations point toward a primary myopathic process, particularly suggestive of a mitochondrial myopathy, where structural mitochondrial abnormalities and accumulation are typical features. Correlation with clinical findings and biochemical markers is essential to establish a definitive diagnosis.

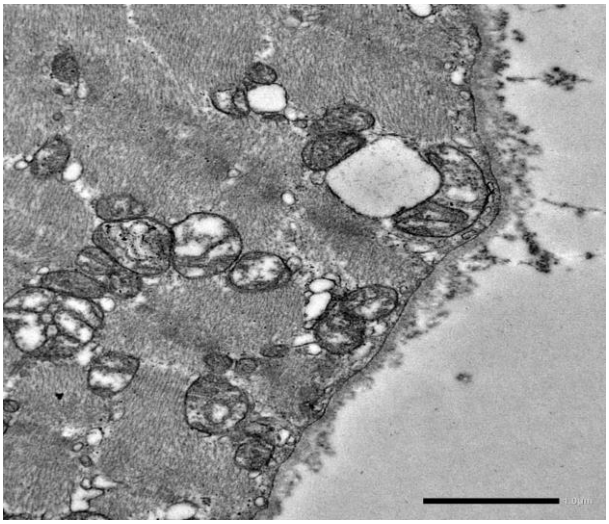

Figure 2 shows the electron microscopy image of skeletal muscle reveals disrupted fiber structure with clusters of abnormally enlarged and irregular mitochondria, many showing loss of normal cristae architecture. Several vacuolated regions are also evident, indicating ongoing degenerative changes. The peripheral location of these altered mitochondria, along with fiber damage, is highly suggestive of a mitochondrial myopathy. Such ultrastructural findings reflect impaired energy metabolism within muscle fibers and correlate with clinical presentations of muscle weakness and elevated muscle enzymes.

DISCUSSION:

Acute myositis as an initial presentation of primary mitochondrial myopathy (PMM) is unusual, as PMM more commonly presents with exercise intolerance and gradually progressive proximal weakness, often with unremarkable or nonspecific EMG findings (3). In the present case, the acute onset of myalgia with raised CPK initially resembled immune-mediated myositis; however, the EMG findings of short-duration, small-amplitude motor units with early recruitment and absence of fibrillation potentials were more compatible with a metabolic or mitochondrial process. Similar Study conducted by Mancuso et al. in the year 2013, reported that mitochondrial disease mimicking inflammatory myositis with ragged-red fibers and sparse inflammation that responded partially to IVIG and carnitine (6). Kyriakidou et al. in the year 2025 also documented a case of acute viral myositis occurring in the context of mitochondrial diabetes, demonstrating that mitochondrial dysfunction can predispose to acute exacerbations (7). Skeletal muscle is the most commonly biopsied tissue in mitochondrial disease, though genetic testing is reducing its need (8). A mosaic COX pattern reflects mutational heteroplasmy, while a quadruple immunofluorescent assay can reveal isolated complex I deficiency. Other nonspecific findings include fiber size variation, internal nuclei, and glycogen or lipid accumulation (9). Muscle biopsy, usually from the

quadriceps or deltoid, is key for diagnosing mitochondrial disorders. COX-deficient fibres (>5% or any in individuals <50 years) strongly suggest mitochondrial disease, especially when SDH-positive. Sub-sarcolemmal mitochondrial accumulations, seen as ragged blue or ragged red fibres, are classic features, with similar age-related thresholds for diagnostic significance (10). In contrast, our patient had no viral trigger and no inflammatory infiltrates, suggesting that mitochondrial pathology alone was responsible for the acute presentation. Idiopathic inflammatory myopathies, as emphasized by West et al in the year 2017, typically demonstrate inflammatory cell infiltrates, necrosis, regeneration, and MHC-I upregulation, none of which were observed here (11). While COX-negative fibers may appear in inflammatory myopathies due to secondary mitochondrial damage.

This usually occurs alongside clear inflammatory features, unlike in the current case. Imaging was nonspecific, which is often reported in mitochondrial disorders, as noted by Wang et al in the year 2015 (12). The transient improvement after steroid therapy should be interpreted cautiously, has been highlighted that glucocorticoids themselves may induce myopathy and mask the true course of disease. No disease-modifying therapy exists for mitochondrial myopathy (MM) (13). Nutritional supplements like carnitine, creatine, CoQ10, and dichloroacetate have shown no proven benefit in controlled trials, though rare CoQ10-deficient cases may respond to CoQ10. Experimental agents such as bezafibrate and MitoQ are under investigation (14). Structured aerobic, endurance, and resistance exercise programs are safe and can improve strength, fatigue, and quality of life (15). Current management principles for PMM emphasize physiotherapy, mitochondrial supplements, and supportive measures, with immunosuppression reserved only for confirmed overlap syndromes. This case underscores the importance of biopsy and genetic confirmation in distinguishing PMM from inflammatory myopathies to prevent unnecessary immunosuppressive therapy.

CONCLUSION:

Acute myositis as an initial manifestation of primary mitochondrial myopathy is extremely rare, as the condition typically presents with progressive proximal weakness and exercise intolerance. In this case, the acute onset of myalgia and elevated CPK levels initially suggested an inflammatory myopathy, abnormal mitochondria on histopathology and electron microscopy confirmed the mitochondrial origin. Imaging findings were nonspecific, and transient improvement with steroids highlighted the potential for misdiagnosis. An increasing number of clinical trials, usually designed to be double blinded and placebo controlled, have investigated the therapeutic effects of various vitamins, cofactors, and nutritional supplements, though often these trials have failed to show definitive beneficial primary and secondary outcomes. Moreover, new molecular and cellular strategies are being proposed that act on a molecular or cellular level, for example restriction endonucleases technologies. This case underscores the importance of combining clinical features with biopsy and genetic testing to distinguish mitochondrial myopathy from inflammatory myopathies, thereby preventing unnecessary immunosuppressive treatment and ensuring appropriate supportive management

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